

Aus dem Forschungsinstitut für die Biologie landwirtschaftlicher Nutztiere, FBN,
Forschungsbereich Genetik und Biometrie, Dummerstorf

INVESTIGATIONS ON THE IMPORTANCE OF GENOMIC IMPRINTING FOR GENETIC VARIATION IN LIVESTOCK

Dissertation

zur Erlangung des Doktorgrades
der Agrar- und Ernährungswissenschaftlichen Fakultät
der Christian-Albrechts-Universität zu Kiel

vorgelegt von

Dipl.-Ing.agr. Nadine Neugebauer

aus Hannover

Dekan: Prof. Dr. Uwe Latacz-Lohmann

1. Berichterstatter: Prof. Dr. Norbert Reinsch

2. Berichterstatter: Prof. Dr. Georg Thaller

Tag der mündlichen Prüfung: 11. Februar 2010

Allen, die ich liebe

TABLE OF CONTENTS

GENERAL INTRODUCTION	1
.....	
CHAPTER ONE	
Evidence for parent-of-origin effects on genetic variability of beef traits	
.....	20
CHAPTER TWO	
Parent-of-origin effects cause genetic variation in pig performance traits	
.....	48
CHAPTER THREE	
On-station heritabilities and parent-of-origin effects for beef traits in German Simmental	
.....	76
CHAPTER FOUR	
Parent-of-origin modulates direct and maternal genetic effects for birth weight and gestation length in German Holsteins	
.....	96
GENERAL DISCUSSION	112
.....	
SUMMARY	118
.....	
ZUSAMMENFASSUNG	119
.....	
APPENDIX	120
.....	

General introduction

Genomic imprinting is an epigenetic process, known to occur in nearly all mammalian species. Imprinted genes are involved in many aspects of development in mammals, plants and, perhaps, birds. The alteration of normal imprinting patterns gives rise to some genetic human diseases, including Beckwith-Wiedemann syndrome, Prader-Willi syndrome or Angelman syndrome and cancer (e.g. Pulford et al., 1999; Jirtle et al., 1999; Morison et al., 2005). The most well-known examples of imprinted genes in livestock are the *callipyge* mutation (major gene) occurring in sheep (Cockett et al., 1994; Smit et al., 2003) and the *IGF2* gene occurring in pigs (Jeon et al., 1999; Nezer et al., 1999; Van Laere et al., 2003). Genomic imprinting depends on the parent's sex: male (female) gametes may be imprinted by methylation, whereas the gametes of the opposite sex may not. Imprinted genes are expressed at a lower level than the other parent's copy. In each generation the imprint is newly established during gametogenesis. The important thing is that genes may not only be imprinted, but also polymorphic and therefore contribute to the total genetic variation. Thus an interesting research problem is to estimate the portion of the additive genetic variance contributed by imprinted genes. This part of the genetic variance will be termed the imprinting variance. In order to assess the relative importance of genomic imprinting for genetic variation Reinsch and Guiard (in preparation) proposed a model including two random genetic effects per individuals, which allows simultaneous consideration of paternal and maternal expression patterns.

The suggested gametic model summarises gene-action under different expression patterns on the gametic level. Basically the model in matrix notation is

$$y = X\beta + Z_s g_s + Z_d g_d + e \quad (1)$$

with the assumptions on the covariance of random effects being

$$\text{Var} \begin{bmatrix} g_s \\ g_d \\ e \end{bmatrix} = \begin{bmatrix} G\sigma_s^2 & G\sigma_{sd} & 0 \\ G\sigma_{sd} & G\sigma_d^2 & 0 \\ 0 & 0 & I\sigma_e^2 \end{bmatrix} \quad (2)$$

This results in the following mixed model equations

$$\begin{bmatrix} X'X & X'Z_s & X'Z_d \\ Z'_s X & Z'_s Z_s + G^{-1}\alpha_1 & Z'_s Z_d + G^{-1}\alpha_2 \\ Z'_d X & Z'_d Z_s + G^{-1}\alpha_2 & Z'_d Z_d + G^{-1}\alpha_3 \end{bmatrix} \begin{bmatrix} \tilde{\beta} \\ \hat{g}_s \\ \hat{g}_d \end{bmatrix} = \begin{bmatrix} X'y \\ Z'_s y \\ Z'_d y \end{bmatrix} \quad (3)$$

where y is the vector of observations, X is the design matrix for fixed effects with the corresponding vector β ; Z_s and Z_d are the design matrices for random genetic effects with the vectors g_s (paternal expression pattern) and g_d (maternal expression pattern) for parental gametic effects and e is the vector of random residuals. In the mixed model equation variance components are represented by

$$\begin{bmatrix} \alpha_1 & \alpha_2 \\ \alpha_2 & \alpha_3 \end{bmatrix}^{-1} = \frac{1}{\sigma_e^2} \begin{bmatrix} \sigma_s^2 & \sigma_{sd} \\ \sigma_{sd} & \sigma_d^2 \end{bmatrix} \quad (4)$$

The matrix G is the usual gametic relationship matrix (Schaeffer et al., 1989).

There is a possibility to use the additive genetic relationship matrix instead of the gametic relationship matrix and thereby reducing the number of equations. Again the model of analysis comprises two genetic effects per animal. The first one accounts for the effect on progeny of alleles inherited with a paternal expression pattern (genetic effect as sire, i.e. half of the breeding value as sire) and the second one is the effect of the same genotype, but mediated to progeny in connection with a maternal expression pattern (genetic effect as dam, i.e. half of the breeding value as dam). In matrix notation the model can be expressed as

$$y = X\beta + Z_s a_s + Z_d a_d + e \quad (5)$$

Now the assumptions on the covariance of random effects are

$$\text{Var} \begin{bmatrix} a_s \\ a_d \\ e \end{bmatrix} = \begin{bmatrix} \frac{1}{2} A \sigma_s^2 & \frac{1}{2} A \sigma_{sd} & 0 \\ \frac{1}{2} A \sigma_{sd} & \frac{1}{2} A \sigma_d^2 & 0 \\ 0 & 0 & W \sigma_R^2 \end{bmatrix} \quad (6)$$

Resulting in the following mixed model equations

$$\begin{bmatrix} X'W^{-1}X & X'W^{-1}Z_s & X'W^{-1}Z_d \\ Z_s'W^{-1}X & Z_s'W^{-1}Z_s + A^{-1}\alpha_1 & Z_s'W^{-1}Z_d + A^{-1}\alpha_2 \\ Z_d'W^{-1}X & Z_d'W^{-1}Z_s + A^{-1}\alpha_2 & Z_d'W^{-1}Z_d + A^{-1}\alpha_3 \end{bmatrix} \begin{bmatrix} \tilde{\beta} \\ \hat{a}_s \\ \hat{a}_d \end{bmatrix} = \begin{bmatrix} X'W^{-1}y \\ Z_s'W^{-1}y \\ Z_d'W^{-1}y \end{bmatrix} \quad (7)$$

where y , X and β are defined as before, Z_s and Z_d are the design matrices for random effects with the vectors a_s and a_d for half of the breeding values as sire (a_s) and as dam (a_d) and e is the vector of random residuals. In terms of gametic variances the variance components for the genetic effects are $\frac{1}{2}\sigma_s^2$ for the paternal one, $\frac{1}{2}\sigma_d^2$ for the maternal one and $\frac{1}{2}\sigma_{sd}$ for their covariance. In the mixed model equations the variance components are represented by

$$\begin{bmatrix} \alpha_1 & \alpha_2 \\ \alpha_2 & \alpha_3 \end{bmatrix}^{-1} = \frac{1}{\sigma_R^2} \begin{bmatrix} \sigma_s^2 & \sigma_{sd} \\ \sigma_{sd} & \sigma_d^2 \end{bmatrix} \quad (8)$$

The matrix A is the usual numerator-relationship matrix. The diagonal matrix W has the elements

$$w_{ii} = \left[\frac{\frac{1}{2}\sigma_s^2(1-F_{si}) + \frac{1}{2}\sigma_d^2(1-F_{di}) + \sigma_e^2}{\frac{1}{2}\sigma_s^2 + \frac{1}{2}\sigma_d^2 + \sigma_e^2} \right]^{-1} \quad (9)$$

and adjusts the error variance of each observation due to variation in the Mendelian sampling term according to the inbreeding coefficients (F_{si} , F_{di}) of the parents.

The imprinting variance can be expressed as the variance of the difference of the gametic effects as sire and as dam

$$\sigma_i^2 = \sigma_s^2 + \sigma_d^2 - 2\sigma_{sd}. \quad (10)$$

The equivalence of both models is demonstrated by a small example. The used pedigree is shown in Figure 1 and includes some inbred individuals, as it is usually found in livestock.

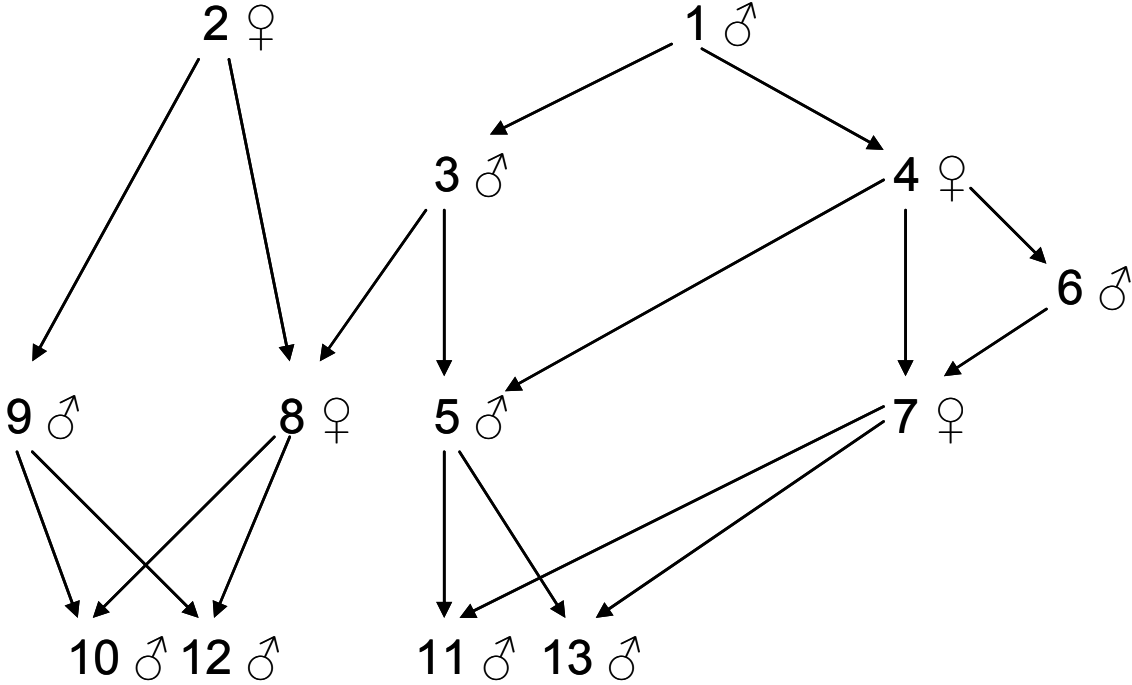


Figure 1: Example pedigree

The weights of bulls 10 to 13 after 350 days are shown in Table 1.

Table 1: Observations for the example

observation	animal	weight
y_1	10	625
y_2	11	375
y_3	12	400
y_4	13	355

Design matrices and vectors for the mixed model equations for the gametic imprinting model are $X' = [1 \ 1 \ 1 \ 1]$ for the overall mean, the design matrix for gametic effects with paternal expression pattern

$$Z_s = \begin{bmatrix} 0 & 0 & 0 & 0 & 0 & 0 & 0 & 0 & 0 & 0 & 0 & 0 & 0 & 0 & 0 & 0 & 0 & 0 & 1 & 0 & 0 & 0 & 0 & 0 & 0 & 0 \\ 0 & 1 & 0 & 0 & 0 & 0 & 0 \\ 0 & 1 & 0 & 0 & 0 & 0 \\ 0 & 1 & 0 \end{bmatrix}$$

the design matrix for gametic effects with maternal expression pattern

$$Z_d = \begin{bmatrix} 0 & 0 & 0 & 0 & 0 & 0 & 0 & 0 & 0 & 0 & 0 & 0 & 0 & 0 & 0 & 0 & 0 & 0 & 1 & 0 & 0 & 0 & 0 & 0 & 0 \\ 0 & 1 & 0 & 0 & 0 & 0 & 0 \\ 0 & 1 & 0 & 0 & 0 & 0 \\ 0 & 1 & 0 \end{bmatrix}$$

the vector of observations

$$Y' = [625 \quad 375 \quad 400 \quad 355]$$

and the gametic relationship matrix as shown on page 6.

Assume that $\sigma_e^2 = 5565$, $\sigma_s^2 = 2552$, $\sigma_d^2 = 11830$ and $\sigma_{sd} = 2040$; therefore

$$\alpha = \begin{bmatrix} \alpha_1 & \alpha_2 \\ \alpha_2 & \alpha_3 \end{bmatrix} = \sigma_e^2 \begin{bmatrix} \sigma_s^2 & \sigma_{sd} \\ \sigma_{sd} & \sigma_d^2 \end{bmatrix}^{-1} = \begin{bmatrix} 2.529 & -0.436 \\ -0.436 & 0.546 \end{bmatrix}$$

The results of the gametic imprinting model are shown in Table 2.

Now we have a look on the design matrices for the imprinting model with additive genetic relationship matrix (reduced model):

The design matrix for the breeding value as sire

$$Z_s = \begin{bmatrix} 0 & 0 & 0 & 0 & 0 & 0 & 0 & 0 & 1 \\ 0 & 0 & 0 & 0 & 1 & 0 & 0 & 0 & 0 \\ 0 & 0 & 0 & 0 & 0 & 0 & 0 & 0 & 1 \\ 0 & 0 & 0 & 0 & 1 & 0 & 0 & 0 & 0 \end{bmatrix}$$

	1	0	0	0	0.5	0	0.5	0	0.25	0.25	0	0.25	0.125	0.25	0.25	0	0	0	0	0.125	0.25	0.188	0	0.125	0.25	0.188
	0	1	0	0	0.5	0	0.5	0	0.25	0.25	0	0.25	0.125	0.25	0.25	0	0	0	0	0.125	0.25	0.188	0	0.125	0.25	0.188
	0	0	1	0	0	0	0	0	0	0	0	0	0	0	0	0.5	0	0.5	0.25	0.25	0	0	0.25	0.25	0	0
	0	0	0	1	0	0	0	0	0	0	0	0	0	0	0	0.5	0	0.5	0.25	0.25	0	0	0.25	0.25	0	0
	0.5	0.5	0	0	1	0	0.5	0	0.5	0.25	0	0.25	0.125	0.25	0.5	0	0	0	0	0.25	0.375	0.188	0	0.25	0.375	0.188
	0	0	0	0	0	1	0	0	0.5	0	0	0	0	0	0.5	0	0	0	0	0.25	0.25	0	0	0.25	0.25	0
	0.5	0.5	0	0	0.5	0	1	0	0.25	0.5	0	0.5	0.25	0.5	0.25	0	0	0	0	0.125	0.375	0.375	0	0.125	0.375	0.375
	0	0	0	0	0	0	0	1	0	0.5	0	0.5	0.25	0.5	0	0	0	0	0	0	0.25	0.375	0	0	0.25	0.375
	0.25	0.25	0	0	0.5	0.5	0.25	0	1	0.125	0	0.125	0.062	0.125	0.5	0	0	0	0	0.25	0.562	0.094	0	0.25	0.562	0.094
	0.25	0.25	0	0	0.25	0	0.5	0.5	0.125	1	0	0.5	0.25	0.5	0.125	0	0	0	0	0.062	0.562	0.375	0	0.062	0.562	0.375
	0	0	0	0	0	0	0	0	0	0	1	0	0.5	0	0	0	0	0	0	0	0	0.25	0	0	0	0.25
	0.25	0.25	0	0	0.25	0	0.5	0.5	0.125	0.5	0	1	0.5	0.5	0.125	0	0	0	0	0.062	0.312	0.5	0	0.062	0.312	0.5
	0.125	0.125	0	0	0.125	0	0.25	0.25	0.062	0.25	0.5	0.5	1	0.25	0.062	0	0	0	0	0.031	0.156	0.625	0	0.031	0.156	0.625
	0.25	0.25	0	0	0.25	0	0.5	0.5	0.125	0.5	0	0.5	0.25	1	0.125	0	0	0	0	0.062	0.312	0.625	0	0.062	0.312	0.625
	0.25	0.25	0	0	0.5	0.5	0.25	0	0.5	0.125	0	0.125	0.062	0.125	1	0	0	0	0	0.5	0.312	0.094	0	0.5	0.312	0.094
	0	0	0.5	0.5	0	0	0	0	0	0	0	0	0	0	0	1	0	0.5	0.25	0.5	0	0	0.25	0.5	0	0
	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	1	0	0.5	0	0	0	0.5	0	0	0
	0	0	0.5	0.5	0	0	0	0	0	0	0	0	0	0	0	0.5	0	1	0.5	0.25	0	0	0.5	0.25	0	0
	0	0	0.25	0.25	0	0	0	0	0	0	0	0	0	0	0	0.25	0.5	0.5	1	0.125	0	0	0.5	0.125	0	0
	0.125	0.125	0.25	0.25	0.25	0.25	0.125	0	0.25	0.062	0	0.062	0.031	0.062	0.5	0.5	0	0.25	0.125	1	0.156	0.047	0.125	0.5	0.156	0.047
	0.25	0.25	0	0	0.375	0.25	0.375	0.25	0.562	0.562	0	0.312	0.156	0.312	0.312	0	0	0	0	0.156	1	0.234	0	0.156	0.562	0.234
	0.188	0.188	0	0	0.188	0	0.375	0.375	0.094	0.375	0.25	0.5	0.625	0.625	0.094	0	0	0	0	0.047	0.234	1	0	0.047	0.234	0.625
	0	0	0.25	0.25	0	0	0	0	0	0	0	0	0	0	0	0.25	0.5	0.5	0.5	0.125	0	0	1	0.125	0	0
	0.125	0.125	0.25	0.25	0.25	0.25	0.125	0	0.25	0.062	0	0.062	0.031	0.062	0.5	0.5	0	0.25	0.125	0.5	0.156	0.047	0.125	1	0.156	0.047
	0.25	0.25	0	0	0.375	0.25	0.375	0.25	0.562	0.562	0	0.312	0.156	0.312	0.312	0	0	0	0	0.156	0.562	0.234	0	0.156	1	0.234
	0.188	0.188	0	0	0.188	0	0.375	0.375	0.094	0.375	0.25	0.5	0.625	0.625	0.094	0	0	0	0	0.047	0.234	0.625	0	0.047	0.234	1

G =

and the design matrix for the breeding value as dam is

$$Z_d = \begin{bmatrix} 0 & 0 & 0 & 0 & 0 & 0 & 0 & 1 & 0 \\ 0 & 0 & 0 & 0 & 0 & 0 & 1 & 0 & 0 \\ 0 & 0 & 0 & 0 & 0 & 0 & 0 & 1 & 0 \\ 0 & 0 & 0 & 0 & 0 & 0 & 1 & 0 & 0 \end{bmatrix}$$

The design matrix for fixed effects and the vector of observations are identical in both models.

The additive genetic relationship matrix is

$$A = \begin{bmatrix} 1.000 & 0 & 0.5000 & 0.500 & 0.50000 & 0.2500 & 0.37500 & 0.25000 & 0.00 \\ 0.000 & 1 & 0.0000 & 0.000 & 0.00000 & 0.000 & 0.00000 & 0.50000 & 0.50 \\ 0.500 & 0 & 1.0000 & 0.250 & 0.62500 & 0.1250 & 0.18750 & 0.50000 & 0.00 \\ 0.500 & 0 & 0.2500 & 1.000 & 0.62500 & 0.5000 & 0.75000 & 0.12500 & 0.00 \\ 0.500 & 0 & 0.6250 & 0.625 & 1.12500 & 0.3125 & 0.46875 & 0.31250 & 0.00 \\ 0.250 & 0 & 0.1250 & 0.500 & 0.31250 & 1.0000 & 0.75000 & 0.06250 & 0.00 \\ 0.375 & 0 & 0.1875 & 0.750 & 0.46875 & 0.7500 & 1.25000 & 0.09375 & 0.00 \\ 0.250 & 0.5 & 0.5000 & 0.125 & 0.31250 & 0.0625 & 0.09375 & 1.00000 & 0.25 \\ 0.000 & 0.5 & 0.0000 & 0.000 & 0.00000 & 0.0000 & 0.00000 & 0.25000 & 1.00 \end{bmatrix}$$

In terms of gametic variances the variance components for the reduced imprinting model are

$$\sigma_R^2 = 12756 \quad (\text{equal to } \sigma_e^2 + \frac{1}{2}\sigma_s^2 + \frac{1}{2}\sigma_d^2), \quad \frac{1}{2}\sigma_s^2 = 1276, \quad \frac{1}{2}\sigma_d^2 = 5915 \quad \text{and} \quad \frac{1}{2}\sigma_{sd} = 1020;$$

$$\text{consequently } \alpha = \begin{bmatrix} \alpha_1 & \alpha_2 \\ \alpha_2 & \alpha_3 \end{bmatrix} = \frac{1}{\sigma_R^2} \begin{bmatrix} \sigma_s^2 & \sigma_{sd} \\ \sigma_{sd} & \sigma_d^2 \end{bmatrix}^{-1} = \begin{bmatrix} 11.595 & -1.999 \\ -1.999 & 2.501 \end{bmatrix}$$

The diagonal matrix W has two equal elements for the animals 11 and 13, because their sire has an inbreeding coefficient of 0.125 and their dam of 0.25.

$$w_{ii} = \left[\frac{1276(1-0.125) + 5915(1-0.25) + 5565}{1276 + 5915 + 5565} \right]^{-1} = (0.8715702)^{-1}$$

The inbreeding coefficients of the parents of animal 10 and 12 are zero, thus the observations y_1 and y_3 have a weighting factor of 1 and W becomes

$$W^{-1} = \begin{bmatrix} 1 & 0 & 0 & 0 \\ 0 & 1.147354 & 0 & 0 \\ 0 & 0 & 1 & 0 \\ 0 & 0 & 0 & 1.147354 \end{bmatrix}$$

The results of this reduced imprinting model are also shown in Table 2 (page: 9). The breeding values as sire (as dam) from the gametic imprinting model are the sum of both gametic effects as sire (as dam) per animal. The breeding values as sire (as dam) from the reduced imprinting model were calculated as twice the estimates of the genetic effects as sire (as dam). Estimates of genetic effects from the gametic model are summarized in column four and five. The reduced imprinting model (using the additive genetic relationship matrix) provides estimates of half of the breeding values as sire and as dam (column six and seven). For comparison the breeding values from the reduced model (twice the corresponding genetic effects, no estimates for final offspring) are shown in Table 2, column eight and nine. As expected, the reduced imprinting model and the gametic imprinting model yield equivalent results.

Table 2: Results of the gametic imprinting model and the reduced imprinting model.

animal	gametic effects		breeding values from the gametic model		parental effect from the reduced imprinting model		breeding values from the reduced imprinting model	
	as sire	as dam	as sire	as dam	as sire	as dam	as sire	as dam
1	-4.080	-6.658	-8.159	-13.317	-4.080	-6.658	-8.159	-13.317
	-4.080	-6.658						
2	6.118	18.480	12.236	36.959	6.118	18.480	12.236	36.959
	6.118	18.480						
3	-4.421	-0.137	-5.103	12.907	-2.551	6.454	-5.103	12.907
	-0.682	13.044						
4	-7.818	-19.839	-15.295	-46.199	-7.648	-23.099	-15.295	-46.199
	-7.477	-26.360						
5	-5.952	3.736	-16.999	-22.082	-8.500	-11.041	-16.999	-22.082
	-11.048	-25.817						
6	-2.718	-15.762	-11.725	-46.742	-5.862	-23.371	-11.725	-46.742
	-9.007	-30.980						
7	-8.580	-39.133	-18.946	-77.994	-9.473	-38.997	-18.946	-77.994
	-10.366	-38.861						
8	0.167	22.215	9.003	56.456	4.501	28.228	9.003	56.456
	8.836	34.241						
9	6.800	5.436	16.319	26.634	8.159	13.317	16.319	26.634
	9.518	21.198						
10	22.813	25.031	39.028	121.187				
	16.215	96.156						
11	-10.471	-12.616	-21.294	-59.444				
	-10.823	-46.828						
12	0.306	7.039	-1.471	-1.138				
	-1.776	-8.177						
13	-12.479	-14.222	-24.679	-69.030				
	-12.200	-54.808						

Including maternal effects into the reduced imprinting model (5) the model can be expressed

as

$$y = X\beta + Z_s a_s + Z_d a_d + Z_m a_m + e \quad (11)$$

and yields covariances of genetic effects as

$$\text{Var} \begin{bmatrix} m \\ a_d \\ a_s \\ e \end{bmatrix} = \begin{bmatrix} A\sigma_m^2 & A\frac{1}{2}\sigma_{m,d} & A\frac{1}{2}\sigma_{m,s} & 0 \\ A\frac{1}{2}\sigma_{m,d} & A\frac{1}{2}\sigma_d^2 & A\frac{1}{2}\sigma_{s,d} & 0 \\ A\frac{1}{2}\sigma_{m,s} & A\frac{1}{2}\sigma_{s,d} & A\frac{1}{2}\sigma_s^2 & 0 \\ 0 & 0 & 0 & W\sigma_R^2 \end{bmatrix} \quad (12)$$

Here m denotes maternal genetic effects and σ_m^2 is the corresponding genetic variance; $\sigma_{m,d}$ and $\sigma_{m,s}$ are covariances between maternal and both direct effects and all other symbols are defined as before.

With equal weights the variance of the observations y equals

$$\begin{aligned} \text{Var}(y) = & Z_m AZ_m' \sigma_m^2 + 2Z_m AZ_d' \frac{1}{2} \sigma_{m,d} + 2Z_m AZ_s' \frac{1}{2} \sigma_{m,s} + \\ & Z_d AZ_d' \frac{1}{2} \sigma_d^2 + 2Z_d AZ_s' \frac{1}{2} \sigma_{s,d} + Z_s AZ_s' \frac{1}{2} \sigma_s^2 + I\sigma_e^2 \end{aligned} \quad (13)$$

Since here the design matrix for the maternal genetic effects Z_m always equals the design matrix for the genetic effects as dam (half of the breeding values as dam)

$M = Z_m AZ_m' = Z_m AZ_d' = Z_d AZ_d'$. Likewise we set $C = Z_m AZ_s' = Z_d AZ_s'$. As a result $\text{Var}(y)$ becomes

$$\text{Var}(y) = M \left[\sigma_m^2 + 2\frac{1}{2}\sigma_{m,d} + \frac{1}{2}\sigma_d^2 \right] + Z_s AZ_s' \frac{1}{2}\sigma_s^2 + 2C \left[\frac{1}{2}\sigma_{m,s} + \frac{1}{2}\sigma_{s,d} \right] + I\sigma_e^2 \quad (14)$$

The latter expression clearly shows that M has an associated variance component, which is composed by the gametic variance as dam (σ_d^2) plus the genetic maternal variance (σ_m^2) and the covariance between maternal genetic effects and direct genetic effects as dam ($\sigma_{m,d}$). Also matrix C has an associated variance component, which itself is a function of the covariance between maternal genetic effects and direct genetic effects as sire ($\sigma_{m,s}$) and the covariance between direct genetic effects as sire and as dam ($\sigma_{s,d}$). Therefore at the very end the reduced imprinting model cannot be applied for separating maternal and direct genetic variances as sire and dam, because those variances can not be distinguished.

As a way out of this variance component separation problem a gametic model was applied, where both the direct and the maternal genetic effects are replaced by gametic effects. Furthermore, in order to allow for genomic imprinting both for the direct and maternal effect, both gametic effects occur as sire as well as as dam

$$y = X\beta + Z_{d_s}g_{d_s} + Z_{d_d}g_{d_d} + Z_{m_s}g_{m_s} + Z_{m_d}g_{m_d} + e \quad (15)$$

with the assumptions on the covariance of random effects being

$$\text{Var} \begin{bmatrix} g_{d_s} \\ g_{d_d} \\ g_{m_s} \\ g_{m_d} \\ e \end{bmatrix} = \begin{bmatrix} G\sigma_{d_s}^2 & G\sigma_{d_{sd}} & G\sigma_{d_s m_s} & G\sigma_{d_s m_d} & 0 \\ G\sigma_{d_{sd}} & G\sigma_{d_d}^2 & G\sigma_{d_d m_s} & G\sigma_{d_d m_d} & 0 \\ G\sigma_{d_s m_s} & G\sigma_{d_d m_s} & G\sigma_{m_s}^2 & G\sigma_{m_{sd}} & 0 \\ G\sigma_{d_s m_d} & G\sigma_{d_d m_d} & G\sigma_{m_{sd}} & G\sigma_{m_d}^2 & 0 \\ 0 & 0 & 0 & 0 & I\sigma_e^2 \end{bmatrix} \quad (16)$$

This results in the mixed model equations (17) on page 12, with y being the vector of observations, X being the design matrix for fixed effects with the corresponding vector β ; Z_{d_s} and Z_{d_d} are the design matrices for direct random gametic effects with the vectors g_{d_s} (paternal expression pattern) and g_{d_d} (maternal expression pattern) for parental gametic effects; Z_{m_s} and Z_{m_d} are the design matrices for maternal random gametic effects with the vectors g_{m_s} (paternal expression pattern) and g_{m_d} (maternal expression pattern) for parental gametic effects of the dam and e is the vector of random residuals. In the mixed model equation the variance components are represented by

$$\begin{bmatrix} \alpha_1 & \alpha_2 & | & \alpha_3 & \alpha_4 \\ \alpha_2 & \alpha_5 & | & \alpha_6 & \alpha_7 \\ \hline \alpha_3 & \alpha_6 & | & \alpha_8 & \alpha_9 \\ \alpha_4 & \alpha_7 & | & \alpha_9 & \alpha_{10} \end{bmatrix}^{-1} = \frac{1}{\sigma_e^2} \begin{bmatrix} \sigma_{d_s}^2 & \sigma_{d_{sd}} & | & \sigma_{d_s m_s} & \sigma_{d_s m_d} \\ \sigma_{d_{sd}} & \sigma_{d_d}^2 & | & \sigma_{d_d m_s} & \sigma_{d_d m_d} \\ \hline \sigma_{d_s m_s} & \sigma_{d_d m_s} & | & \sigma_{m_s}^2 & \sigma_{m_{sd}} \\ \sigma_{d_s m_d} & \sigma_{d_d m_d} & | & \sigma_{m_{sd}} & \sigma_{m_d}^2 \end{bmatrix} = \frac{1}{\sigma_e^2} \mathcal{S} \quad (18)$$

$$\begin{bmatrix}
\mathbf{X}'\mathbf{X} & \mathbf{X}'\mathbf{Z}_{d_s} & \mathbf{X}'\mathbf{Z}_{d_d} & \mathbf{X}'\mathbf{Z}_{m_s} & \mathbf{X}'\mathbf{Z}_{m_d} \\
\mathbf{Z}_{d_s}'\mathbf{X} & \mathbf{Z}_{d_s}'\mathbf{Z}_{d_s} + \mathbf{G}^{-1}\alpha_1 & \mathbf{Z}_{d_s}'\mathbf{Z}_{d_d} + \mathbf{G}^{-1}\alpha_2 & \mathbf{Z}_{d_s}'\mathbf{Z}_{m_s} + \mathbf{G}^{-1}\alpha_3 & \mathbf{Z}_{d_s}'\mathbf{Z}_{m_d} + \mathbf{G}^{-1}\alpha_4 \\
\mathbf{Z}_{d_d}'\mathbf{X} & \mathbf{Z}_{d_d}'\mathbf{Z}_{d_s} + \mathbf{G}^{-1}\alpha_2 & \mathbf{Z}_{d_d}'\mathbf{Z}_{d_d} + \mathbf{G}^{-1}\alpha_5 & \mathbf{Z}_{d_d}'\mathbf{Z}_{m_s} + \mathbf{G}^{-1}\alpha_6 & \mathbf{Z}_{d_d}'\mathbf{Z}_{m_d} + \mathbf{G}^{-1}\alpha_7 \\
\mathbf{Z}_{m_s}'\mathbf{X} & \mathbf{Z}_{m_s}'\mathbf{Z}_{d_s} + \mathbf{G}^{-1}\alpha_3 & \mathbf{Z}_{m_s}'\mathbf{Z}_{d_d} + \mathbf{G}^{-1}\alpha_6 & \mathbf{Z}_{m_s}'\mathbf{Z}_{m_s} + \mathbf{G}^{-1}\alpha_8 & \mathbf{Z}_{m_s}'\mathbf{Z}_{m_d} + \mathbf{G}^{-1}\alpha_9 \\
\mathbf{Z}_{m_d}'\mathbf{X} & \mathbf{Z}_{m_d}'\mathbf{Z}_{d_s} + \mathbf{G}^{-1}\alpha_4 & \mathbf{Z}_{m_d}'\mathbf{Z}_{d_d} + \mathbf{G}^{-1}\alpha_7 & \mathbf{Z}_{m_d}'\mathbf{Z}_{m_s} + \mathbf{G}^{-1}\alpha_9 & \mathbf{Z}_{m_d}'\mathbf{Z}_{m_d} + \mathbf{G}^{-1}\alpha_{10}
\end{bmatrix}
\begin{bmatrix}
\tilde{\boldsymbol{\beta}} \\
\hat{\boldsymbol{\sigma}}_{d_s} \\
\hat{\boldsymbol{\sigma}}_{d_d} \\
\hat{\boldsymbol{\sigma}}_{m_s} \\
\hat{\boldsymbol{\sigma}}_{m_d}
\end{bmatrix}
=
\begin{bmatrix}
\mathbf{X}'\mathbf{y} \\
\mathbf{Z}_{d_s}'\mathbf{y} \\
\mathbf{Z}_{d_d}'\mathbf{y} \\
\mathbf{Z}_{m_s}'\mathbf{y} \\
\mathbf{Z}_{m_d}'\mathbf{y}
\end{bmatrix}
\quad (17)$$

The imprinting variance for the direct effect $\sigma_{i_d}^2$ can be expressed as the variance of the difference of the gametic effects as sire and as dam

$$\sigma_{i_d}^2 = \sigma_{d_s}^2 + \sigma_{d_d}^2 - 2\sigma_{d_{sd}}. \quad (19)$$

The imprinting variance $\sigma_{i_d}^2$ is a part of the total direct additive genetic variance $\sigma_{a_d}^2$ ($\sigma_{d_s}^2 + \sigma_{d_d}^2$). The imprinting variance of the maternal effect $\sigma_{i_m}^2$ can be calculated in the same way

$$\sigma_{i_m}^2 = \sigma_{m_s}^2 + \sigma_{m_d}^2 - 2\sigma_{m_{sd}}. \quad (20)$$

Again the imprinting variance $\sigma_{i_m}^2$ is part of the total maternal additive genetic variance $\sigma_{a_m}^2$ ($\sigma_{m_s}^2 + \sigma_{m_d}^2$). The other parts of both additive genetic variances are called Mendelian variance ($\sigma_{M_d}^2$ and $\sigma_{M_m}^2$), which describes the “unimprinted” component of the additive genetic variance (not to be confused with the *Mendelian* sampling variance).

Additionally, the imprinting variance for a particular effect per trait can be divided into a paternal contribution $\sigma_s^2 - \sigma_{sd}$ and a maternal contribution $\sigma_d^2 - \sigma_{sd}$.

With regard to separation of covariance components we express the covariance of the gametic effects, as

$$\text{Var}(g) \begin{bmatrix} g_{d_s} \\ g_{d_d} \\ g_{m_s} \\ g_{m_d} \end{bmatrix} = S \otimes G \quad (21)$$

setting

$$Z = \begin{bmatrix} Z_{d_s} & Z_{d_d} & Z_{m_s} & Z_{m_d} \end{bmatrix} \quad (22)$$

the variance of the observations becomes

$$\begin{aligned} \text{Var}(y) = Z \cdot \text{Var}(g) \cdot Z' + I\sigma_e^2 = \\ Z_{d_s} GZ_{d_s}' \sigma_{d_s}^2 + 2Z_{d_s} GZ_{d_d}' \sigma_{d_s d_d} + 2Z_{d_s} GZ_{m_s}' \sigma_{d_s m_s} + \\ 2Z_{d_s} GZ_{m_d}' \sigma_{d_s m_d} + Z_{d_d} GZ_{d_d}' \sigma_{d_d}^2 + 2Z_{d_d} GZ_{m_s}' \sigma_{d_d m_s} + \\ 2Z_{d_d} GZ_{m_d}' \sigma_{d_d m_d} + Z_{m_s} GZ_{m_s}' \sigma_{m_s}^2 + 2Z_{m_s} GZ_{m_d}' \sigma_{m_s m_d} + \\ Z_{m_d} GZ_{m_d}' \sigma_{m_d}^2 + I\sigma_e^2 \end{aligned} \quad (23)$$

In general the matrices associated with gametic covariance components are not null nor are they linear dependent. For the example data these matrices ($Z_i GZ_j'$) are shown in the appendix (1). The resulting breeding values are presented in Table 3 (page no. 16). The design matrices are the design matrix of direct paternal expressed gametic effects

$$Z_{d_s} = \begin{bmatrix} 0 & 0 & 0 & 0 & 0 & 0 & 0 & 0 & 0 & 0 & 0 & 0 & 0 & 0 & 0 & 0 & 0 & 1 & 0 & 0 & 0 & 0 & 0 & 0 & 0 \\ 0 & 0 & 0 & 0 & 0 & 0 & 0 & 0 & 0 & 0 & 0 & 0 & 0 & 0 & 0 & 0 & 0 & 0 & 0 & 1 & 0 & 0 & 0 & 0 & 0 \\ 0 & 1 & 0 & 0 & 0 \\ 0 & 1 & 0 \end{bmatrix}$$

and the design matrix of direct maternal expressed gametic effects

$$Z_{d_d} = \begin{bmatrix} 0 & 0 & 0 & 0 & 0 & 0 & 0 & 0 & 0 & 0 & 0 & 0 & 0 & 0 & 0 & 0 & 0 & 0 & 1 & 0 & 0 & 0 & 0 & 0 & 0 \\ 0 & 1 & 0 & 0 & 0 & 0 \\ 0 & 1 & 0 & 0 \\ 0 & 1 \end{bmatrix}$$

The design matrix of genetic maternal paternal expressed gametic effects

$$Z_{m_s} = \begin{bmatrix} 0 & 0 & 0 & 0 & 0 & 0 & 0 & 0 & 0 & 0 & 0 & 0 & 0 & 0 & 1 & 0 & 0 & 0 & 0 & 0 & 0 & 0 & 0 & 0 & 0 \\ 0 & 0 & 0 & 0 & 0 & 0 & 0 & 0 & 0 & 0 & 0 & 0 & 0 & 1 & 0 & 0 & 0 & 0 & 0 & 0 & 0 & 0 & 0 & 0 & 0 \\ 0 & 0 & 0 & 0 & 0 & 0 & 0 & 0 & 0 & 0 & 0 & 0 & 0 & 0 & 1 & 0 & 0 & 0 & 0 & 0 & 0 & 0 & 0 & 0 & 0 \\ 0 & 0 & 0 & 0 & 0 & 0 & 0 & 0 & 0 & 0 & 0 & 0 & 1 & 0 & 0 & 0 & 0 & 0 & 0 & 0 & 0 & 0 & 0 & 0 & 0 \end{bmatrix}$$

and the design matrix of genetic maternal maternal expressed gametic effects

$$Z_{m_d} = \begin{bmatrix} 0 & 0 & 0 & 0 & 0 & 0 & 0 & 0 & 0 & 0 & 0 & 0 & 0 & 0 & 1 & 0 & 0 & 0 & 0 & 0 & 0 & 0 & 0 & 0 & 0 \\ 0 & 0 & 0 & 0 & 0 & 0 & 0 & 0 & 0 & 0 & 0 & 0 & 1 & 0 & 0 & 0 & 0 & 0 & 0 & 0 & 0 & 0 & 0 & 0 & 0 \\ 0 & 0 & 0 & 0 & 0 & 0 & 0 & 0 & 0 & 0 & 0 & 0 & 0 & 1 & 0 & 0 & 0 & 0 & 0 & 0 & 0 & 0 & 0 & 0 & 0 \\ 0 & 0 & 0 & 0 & 0 & 0 & 0 & 0 & 0 & 0 & 0 & 1 & 0 & 0 & 0 & 0 & 0 & 0 & 0 & 0 & 0 & 0 & 0 & 0 & 0 \end{bmatrix}$$

We assume that $\sigma_e^2 = 5723$, $\sigma_{d_s}^2 = 2588$, $\sigma_{d_d}^2 = 11478$ with $\sigma_{d_{sd}} = 2164$, $\sigma_{m_s}^2 = 252$,

$\sigma_{m_d}^2 = 252$ with $\sigma_{m_{sd}} = -46$, $\sigma_{d_s m_s} = 26$, $\sigma_{d_s m_d} = 24$, $\sigma_{d_d m_s} = 28$ and $\sigma_{d_d m_d} = 22$, therefore

$$\alpha = \begin{bmatrix} \alpha_1 & \alpha_2 & \alpha_3 & \alpha_4 \\ \alpha_2 & \alpha_5 & \alpha_6 & \alpha_7 \\ \alpha_3 & \alpha_6 & \alpha_8 & \alpha_9 \\ \alpha_4 & \alpha_7 & \alpha_9 & \alpha_{10} \end{bmatrix} = \begin{bmatrix} 2.630 & -0.495 & -0.263 & -0.255 \\ -0.495 & 0.592 & -0.016 & -0.007 \\ -0.263 & -0.016 & 25.528 & 4.321 \\ -0.255 & -0.007 & 4.321 & 23.524 \end{bmatrix}$$

Resulting estimates are shown in Table 3 (page: 16), direct trait effects in columns two and three, breeding values for the direct trait (sum of gametic effects as sire/as dam) in columns four and five, maternal trait gametic effects in columns six and seven and, finally, breeding values for the maternal trait in the second last and the last column.

Table 3: Results of the gametic direct and maternal effects imprinting model. The breeding values as sire (as dam) for the direct and the maternal effects per animal.

animal	direct trait				maternal trait			
	gametic effects		breeding values		gametic effects		breeding values	
	as sire	as dam	as sire	as dam	as sire	as dam	as sire	as dam
1	-4.061	-6.521	-8.122	-13.042	0.180	-0.394	0.36	-0.788
	-4.061	-6.521			0.180	-0.394		
2	6.208	17.702	12.416	35.404	-0.049	0.711	-0.098	1.422
	6.208	17.702			-0.049	0.711		
3	-4.302	-0.461	-4.783	11.658	0.507	-0.455	1.161	-0.577
	-0.481	12.119			0.654	-0.122		
4	-7.882	-19.101	-15.523	-44.262	0.032	-0.727	-0.263	-1.393
	-7.641	-25.161			-0.295	-0.666		
5	-5.739	3.030	-16.848	-21.900	0.547	-0.319	0.382	-1.047
	-11.109	-24.930			-0.165	-0.728		
6	-2.866	-14.918	-12.061	-44.508	-0.688	0.091	-1.163	-0.56
	-9.195	-29.590			-0.475	-0.651		
7	-8.897	-37.172	-19.520	-74.206	-1.270	-0.190	-1.319	-1.567
	-10.623	-37.034			-0.049	-1.377		
8	0.475	20.747	9.544	53.351	1.269	-0.379	1.137	1.013
	9.069	32.604			-0.132	1.392		
9	6.695	5.598	16.251	26.099	0.067	0.062	0.051	0.804
	9.556	20.501			-0.016	0.742		
10	22.885	25.391	39.999	117.527	0.174	0.539	0.902	1.171
	17.114	92.136			0.728	0.632		
11	-10.338	-12.551	-21.470	-56.931	0.172	-0.541	-0.505	-1.338
	-11.132	-44.380			-0.677	-0.797		
12	0.060	6.306	-1.912	-2.787	-0.055	0.327	0.426	0.765
	-1.972	-9.093			0.481	0.438		
13	-12.368	-14.247	-24.954	-66.341	0.151	-0.560	-0.545	-1.372
	-12.586	-52.094			-0.696	-0.812		

Chapter one to three of the following thesis include the use of the reduced imprinting model in order to assess the relative importance of genomic imprinting for the genetic variation of slaughter traits.

Chapter one describes the analysis of imprinting variances in German dual-purpose Simmentals in order to assess the relative importance of genomic imprinting for the genetic variation of slaughter weight, net daily gain, killing out percentage and a set of 22 carcass composition traits. Most of them were recorded by using a video-image analysis (VIA) device.

In chapter two, 33 traits for growth, carcass composition and meat quality of Large White fattening pigs reared on a test station in Switzerland were analysed regarding the influence of genomic imprinting.

The analysis of imprinting variances and heritabilities for Bavarian progeny testing station data is presented in chapter three. The data set included up to 46 traits per animal for growth, carcass composition, meat quality and also a full fatty acid profile of Simmental fattening bulls.

Chapter four includes the use of the gametic direct and maternal effect imprinting model (see above) to separate the imprinting variances for a direct trait and maternal trait simultaneously.

The model was applied to reproduction traits (in German Holstein) which are often affected by maternal effects (e.g. Willham, 1972; Meyer, 1992; Roehe, 1993).

Literature

- Cockett, N. E., Jackson, S. P., Shay, T. L., Nielsen, D., Moore, S. S., Steele, M. R., Barendse, W., Green, R. B., Georges, M., 1994: Chromosomal localization of the callipyge gene in sheep (*ovis aries*) using bovine DNA markers. Proc. Natl. Acad. Sci. USA 91: 3019-3023.
- Engelland, T., Tier, B., 2002: Genetic variances due to imprinted genes in cattle. J. Anim. Breed. Genet. 119 (3): 154-165.
- Jeon, J.-T., Carlborg, Ö., Törnsten, A., Giuffra, E., Amarger, V., Chardon, P., Andersson-Eklund, L., Andersson, K., Hansson, I., Lundström, K., Andersson, L., 1999: a paternally expressed QTL affecting skeletal and cardiac muscle mass in pigs maps to the *IGF2* locus. Nature Genetics 21: 157-158.
- Jirtle, R. L. and Weidman, J. R., 2007: Imprinted and more equal. Am. Sci. 95: 143-149.
- Jirtle, R. L., 1999: Genomic imprinting and cancer. Exp. Cell Res., 248: 18-24.
- Meyer, K., 1992: Bias and sampling covariances of estimates of variance components due to maternal effects. Genet. Sel. Evol. 24: 487-509.
- Morison, I. M., Ramsay, J. P., Spencer, H. G., 2005: A census of mammalian imprinting. Trends Genet. 21(8): 457-465.
- Nezer, C., Moreau, L., Brouwers, B., Coppeters, W., Detilleux, J., Hanset, R., Karim, L., Kvasz, A., Leroy, P., Georges, M., 1999: An imprinted QTL with major effect on muscle mass and fat deposition maps to the *IGF2* locus in pigs. Nature Genetics 21: 155-156.
- Pulford, D. J., Falls, J. G., Killian, J. K., Jirtle, R. L., 1999: Polymorphisms, genomic imprinting and cancer susceptibility. Mutat Res. 436: 59-67.
- Reinsch, N. and Guiard, V.: Estimable genetic covariance components under mixed Mendelian and imprinted inheritance (in preparation).

- Roehe, R., Kennedy, B. W., 1993: The influence of maternal effects on accuracy of evaluation of litter size in swine. *J. Anim. Sci.* 71: 2353-2364.
- Schaeffer, L. R., Kennedy, B. W., Gibson, J. P., 1989: The inverse of the gametic relationship matrix. *J. Dairy Sci.* 72(5): 1266-1272.
- Smit, M., Segers, K., Carrascosa, L. G., Shay, T., Baraldi, F., Gyapay, G., Snowder, G., Georges, M., Cockett, N., Charlier, C., 2003: Mosaicism of solid gold supports the causality of a noncoding A-to-G transition in the determinism of the callipyge phenotype. *Genetics* 163: 453-456.
- Van Laere, A.-S., Nguyen, M., Braunschweig, M., Nezer, C., Colette, C., Moreau, L., Archibald, A. L., Haley, C. S., Buys, N., Tally, M., Andersson, G., Georges, M., Andersson, L., 2003: A regulatory mutation in *IGF2* causes a major QTL effect on muscle growth in the pig. *Nature* 425: 823-836.
- Willham, R. L., 1972: The role of maternal effects in animal breeding: III. Biometrical aspects of maternal effects in animals. *J. Anim. Sci.* 35: 1288-1293.

Chapter 1

Evidence for parent-of-origin effects on genetic variability of beef
traits

Accepted for publication in *Journal of Animal Science*

Evidence for parent-of-origin effects on genetic variability of beef traits

N. Neugebauer*, I. Räder*, H. J. Schild[†], D. Zimmer*, N. Reinsch*

*) Forschungsinstitut für die Biologie landwirtschaftlicher Nutztiere, FBN, Forschungsbereich Genetik und Biometrie, Wilhelm-Stahl-Allee 2, 18196 Dummerstorf, Germany

†) LKV Bayern, Haydnstraße 11, 80336 München, Germany

ABSTRACT

Imprinted genes are involved in many aspects of development in mammals, plants and perhaps birds and may play a role in growth and carcass composition of slaughter animals. In the presence of genomic imprinting the expression and, consequently, the effect on the phenotype of maternal and paternal alleles is different. For genetic evaluation genomic imprinting can be accounted for by incorporating 2 additive genetic effects per animal; the first corresponds to a paternal and the second to a maternal expression pattern of imprinted genes. This model holds whatever the mode of imprinting may be: paternal or maternal, full or partial, or any combination thereof. A set of slaughter data from 65,233 German Simmental fattening bulls was analyzed with respect to the relative importance of the genetic imprinting variance. Besides of slaughter weight, net daily BW gain, and killing out percentage there were 22 other traits describing the carcass composition. The latter traits were evaluated by automatic video-imaging (VIA) devices and were composed of weights of valuable cuts as well as fat and meatiness grade. The number of ancestors in the pedigree was 356,880. Genomic imprinting significantly contributed to the genetic variance of 10 traits, with estimated proportions between 8 and 25% of the total additive genetic variance. For 6 of these traits the maternal contribution to the imprinting variance was larger than the paternal, whereas for all other traits the reverse was true. Fat grade only showed a paternal contribution to the imprinting variance. Estimates

of animal model heritabilities of video-imaging-recorded carcass traits ranged between 20 and 30%.

INTRODUCTION

Genomic imprinting refers to an epigenetic marking of alleles, which are differentially expressed when maternally or paternally inherited. In each generation the imprint is newly established during gametogenesis, either with a parent-specific DNA-methylation of cytosine-phosphate-guanine-rich domains or with an acetylation and methylation of histones (Barlow, 1997; Reik and Walter, 1998; Jirtle and Weidman, 2007). The usual result of paternal (maternal) DNA-methylation is the silencing of paternal (maternal) alleles, which may be full or partial. In the latter case both alleles remain active, but the level of activity and the respective impact on the phenotype differs between alleles.

The perhaps best known examples of imprinted genes in livestock are the *callipyge* mutation (major gene) in sheeps (Cockett et al., 1994; Smit et al., 2003) and the *IGF2* gene in pigs (Jeon et al., 1999; Nezer et al., 1999; Van Laere et al., 2003). The *callipyge* phenotype is characterised by a muscular hypertrophy especially in the pelvic limbs and loin, becoming apparent after an age of 2 to 3 months. A polymorphism in the *IGF2* gene of the pig, also known to be imprinted in the mouse, was identified as the root cause of a previously mapped imprinted QTL in an F₂-line-cross experiment (summarized in Vuocolo et al., 2007). Imprinted QTL in pigs were reported for body compositions (e.g. de Koning et al., 2000), meat quality (e.g. de Koning et al., 2001a) and growth and reproduction traits (e.g. de Koning et al., 2001b). All these mapping experiments were conducted in F₂ families derived by crossing different pig lines because this family type allows to separate *Qq*- and *qQ*-genotypes at the QTL, where the first allele is paternal, and to statistically test for phenotypic differences between them, as they are expected under imprinted inheritance.

The very first results on the importance of imprinted genes for genetic variation in livestock were however obtained by a variance components approach: De Vries et al. (1994) fitted an animal model with an additional paternal or maternal gametic effect, as initially proposed by Gibson et al. (1988), Schaeffer et al. (1989), and later by Tier and Sölkner (1993), to slaughter data of 2 commercial Australian pig lines and attached roughly 10% of the phenotypic variance of backfat thickness and growth rate to both types of parent-specific allelic variation. By using the same methodology, Engellandt and Tier (2002) found significant effects of paternal gametes for pelvic fat (14% of the phenotypic variance), kidney fat (16%) and meat content of the carcass (15%) in German Gelbvieh finishing bulls. Furthermore Essl and Voith (2002) reported significant imprinting effects for protein content and milk yield in different lactations, persistency, days open as well as for true and functional herd life in Austrian dual-purpose Simmental. The latter authors analyzed each trait twice, the first time genetic effects were fitted for sires and the second time for dams. Results on imprinting were derived by a comparison of the respective variance components. All mentioned approaches regard either only paternal or maternal imprinting.

In this report we describe an analysis of beef traits from German dual-purpose Simmentals. Most of them were evaluated by using a video-image analysis (VIA) device. To assess the relative importance of genomic imprinting for the genetic variation of these traits we applied a model including 2 random genetic effects for each animal, as outlined in N. Reinsch und V. Guiard (FBN, unpublished data) that allows simultaneous estimation of paternal and maternal imprinting as well as of any combination of full and partial imprinting. The simultaneous estimation of 3 uncorrelated variance components (Mendelian, paternally imprinted, maternally imprinted), which corresponds to a scenario with loci either fully imprinted (maternal or paternal) or not imprinted at all and results in zero covariances between the corresponding random genetic effects, was proposed by Hill and Keightley (1988). In the case of partial imprinting their genetic covariance matrix is no longer diagonal, and it would become necessary

to estimate covariances, which is impossible. In this report the parameterization circumvents this problem by defining the variance components in a different way. The total additive genetic variance is thereby subdivided into 2 components, the Mendelian and the imprinting variance. The outcomes of significance tests for the presence of genomic imprinting effects are provided as well as REML estimates of genetic covariance components.

MATERIAL AND METHODS

Principle of Analysis

In general the model of analysis is composed of fixed effects and, among possible additional random effects, 2 genetic effects per animal. The first one accounts for the effect on the progeny when alleles are inherited with a paternal expression pattern (genetic effect as sire; i.e., one-half of the breeding value as sire) and the second one is the effect of the same genotype, but mediated to progeny in connection with a maternal expression pattern (genetic effect as dam; i.e., one-half of the breeding value as dam). Basically the model in matrix notation is

$$y = X\beta + Z_s a_s + Z_d a_d + e, \quad (1)$$

where the assumptions on the covariance of random effects are

$$\text{Var} \begin{bmatrix} a_s \\ a_d \\ e \end{bmatrix} = \begin{bmatrix} 1/2 A\sigma_s^2 & 1/2 A\sigma_{sd} & 0 \\ 1/2 A\sigma_{sd} & 1/2 A\sigma_d^2 & 0 \\ 0 & 0 & W\sigma_R^2 \end{bmatrix}. \quad (2)$$

This results in the following mixed model equations

$$\begin{bmatrix} X'W^{-1}X & X'W^{-1}Z_s & X'W^{-1}Z_d \\ Z_s'W^{-1}X & Z_s'W^{-1}Z_s + A^{-1}\alpha_1 & Z_s'W^{-1}Z_d + A^{-1}\alpha_2 \\ Z_d'W^{-1}X & Z_d'W^{-1}Z_s + A^{-1}\alpha_2 & Z_d'W^{-1}Z_d + A^{-1}\alpha_3 \end{bmatrix} \begin{bmatrix} \beta \\ a_s \\ a_d \end{bmatrix} = \begin{bmatrix} X'W^{-1}y \\ Z_s'W^{-1}y \\ Z_d'W^{-1}y \end{bmatrix}, \quad (3)$$

where Y is the vector of observations, X is the design matrix for fixed effects with the corresponding vector β , Z_s and Z_d are the design matrices for random genetic effects with the

vectors a_s and a_d for half of the breeding values as sire and as dam, and e is the vector of random residuals. In terms of gametic variances the variance components for the genetic effects are $1/2\sigma_s^2$ for the paternal one, $1/2\sigma_d^2$ for the maternal one, and $1/2\sigma_{sd}$ for their covariance. In the mixed model equation the variance components are represented by

$$\begin{bmatrix} \alpha_1 & \alpha_2 \\ \alpha_2 & \alpha_3 \end{bmatrix}^{-1} = \frac{1}{2\sigma_R^2} \begin{bmatrix} \sigma_s^2 & \sigma_{sd} \\ \sigma_{sd} & \sigma_d^2 \end{bmatrix} \quad (4)$$

The matrix A is the usual numerator-relationship matrix. The diagonal matrix W has elements

$$w_i = \left[\frac{1/2\sigma_s^2(1-F_{si}) + 1/2\sigma_d^2(1-F_{di}) + \sigma_e^2}{1/2\sigma_s^2 + 1/2\sigma_d^2 + \sigma_e^2} \right]^{-1} \quad (5)$$

and adjusts the error variance of each observation due to variation in the Mendelian sampling term according to the inbreeding coefficients (F_{si}, F_{di}) of both parents. The model therefore is a special kind of reduced model (Quaas and Pollak, 1980). The imprinting variance can be expressed as the variance of the difference of the gametic effects as sire and as dam,

$$\sigma_i^2 = \sigma_s^2 + \sigma_d^2 - 2\sigma_{sd}. \quad (6)$$

Additionally, the imprinting variance for a particular trait can be divided into a paternal contribution $\sigma_s^2 - \sigma_{sd}$ and a maternal contribution $\sigma_d^2 - \sigma_{sd}$. The imprinting variance σ_i^2 is a part of the total additive genetic variance, $\sigma_a^2 = \sigma_s^2 + \sigma_d^2$, which is a generalization of the fact that $\sigma_a^2 = 2\sigma_g^2$ (gametic variance; Schaeffer et al., 1989). The Mendelian variance $\sigma_M^2 = \sigma_a^2 - \sigma_i^2 = 2\sigma_{sd}$, which describes the “unimprinted” component of the additive genetic variance (not to be confused with the *Mendelian* sampling variance), is the other part; thus the additive genetic variance is the sum of Mendelian variance and imprinting variance.

Hypothesis Testing

The genetic covariance components are summarized by the matrix S

$$S = \begin{bmatrix} \sigma_s^2 & \sigma_{sd} \\ \sigma_{sd} & \sigma_d^2 \end{bmatrix}. \quad (7)$$

Given the null hypothesis of nonimprinting ($\sigma_i^2 = 0$) applies, then all variances are equal and S is not positive definite. Alternatively, in the presence of imprinting, the variances and the covariance differ from each other and S is positive definite (i.e., σ_i^2 becomes positive, either because of different variances, because of an imperfect correlation, or both).

For each trait 2 models, the imprinting model and an equivalent non-imprinting model (animal model), were applied and the REML log likelihood was calculated for both models. A REML likelihood ratio test (**RLRT**) with two degrees of freedom was used to test the hypothesis of imprinting against the absence of imprinting.

If ℓ_{R2} is the REML log-likelihood of the more general model and ℓ_{R1} is the REML log-likelihood of the restricted model (i.e., the REML log-likelihood under the null hypothesis), then the RLRT is given by

$$RLRT = 2(\ell_{R2} - \ell_{R1}). \quad (8)$$

Under the null hypothesis and the assumption of between-subject independence (here, no relationships between animals) RLRT is asymptotically distributed as a 1:1 mixture of 2 χ^2 -distributions with 1 and 2 df (Self and Liang, 1987). Because of the violation of such assumptions, we used a χ^2 distribution with 2 df for a conservative test.

Animals and Traits

The used pedigree only included the ancestors of the slaughtered animals. Three hundred fifty-six thousand, eight hundred eighty ancestors were extracted from a comprehensive Simmental pedigree and resulted in a pedigree depth of up to 17 generations. Slaughter data were

recorded in 2 abattoirs. After editing there were 65,233 records from German (dual-purpose) Simmental fattening bulls for analysis. In both slaughter houses, VIA-devices (VBS2000, E+V Technology GmbH, 16515 Oranienburg, Germany) had been installed during the data-collection period from 1998 to 2004. Animal Care and Use Committee approval was not obtained for this study because the data were obtained from an existing database of LKV Bayern. The dataset contained 25 traits (Table 1, page: 28) describing the estimated weight of valuable cuts, carcass composition, and growth. Most of the traits were evaluated by the VIA, only carcass weight and finishing weight were collected conventionally, pistola cut/slaughter weight, killing out percentage and net weight BW gain were calculated traits. The VIA uses the bending of light stripes to catch the information from each carcass (Figure 1, side 29). Analyses about the quality of VIA data evidence high correlations between body weight and the estimated weights of individual cuts (Schild, 2004).

The definition of the valuable cuts is specified according to DLG (Deutsche Landwirtschafts-Gesellschaft, German agricultural association) guidelines (Scheper and Scholz, 1985). The entirety of traits is presented in detail by Table 1 (page: 28). We distinguished between hot weight of the entire round and round-4-cut, a particular German cut composed of the following cuts: topside, silverside, rump, and knuckle. Rose and topside are components of the round. The silverside forms also a part of the round and is located below the topside. The pistola cut is defined as the sum of the hindquarter and strip loin. In contrast to the subjective European Union grading scheme, beefiness and fat scores were objectively evaluated through VIA and composed of 15 grades providing 3 times greater resolution than conventional grading.

Table 1: Overview over all analyzed traits: number of observation (n), raw mean and SD

Trait	n	Mean	SD	Heritability ¹
Beefiness score (1 to 15)	66,506	7.85	1.61	0.31
Meat percentage, %	66,506	73.70	2.82	0.27
Weight of grade 1 meat, kg	66,506	72.90	8.33	0.24
Weight of grade 2 meat, kg	66,506	25.14	3.08	0.21
Weight of grade 3 meat, kg	66,506	41.44	4.08	0.24
Weight of pistola cut, kg	66,506	80.20	7.37	0.27
Pistola cut / slaughter weight, %	66,506	20.37	0.69	0.17
Weight of strip loin, kg	66,506	10.28	1.10	0.24
Weight of strip loin, four cut, kg	66,506	25.26	2.75	0.25
Weight of strip loin + weight, kg	66,506	17.09	2.11	0.17
Round weight, kg	66,506	51.32	5.07	0.26
Hot round weight, kg	66,506	60.20	5.54	0.26
Weight of round, four cut, kg	66,506	43.62	4.42	0.25
Topside weight, kg	66,506	11.59	1.14	0.22
Silverside weight, kg	66,506	12.11	1.30	0.26
Tenderloin weight, kg	66,506	4.07	0.42	0.22
Rose weight, kg	66,506	9.31	1.06	0.26
Shoulder weight, kg	66,506	28.56	2.97	0.26
Weight of forequarter, kg	66,506	94.15	10.56	0.25
Weight of forequarter, 3 cut, kg	66,506	16.18	1.74	0.26
Total saleable meat weight, kg	66,506	139.62	15.04	0.25
Killing out percentage, %	64,642	58.59	5.12	0.22
Fat score (1 to 15)	66,490	7.09	1.78	0.25
Carcass weight, kg	66,506	394.38	40.09	0.26
Net BW gain, g/d	64,786	691.82	93.80	0.28

1) The heritability in the last column is from an animal model, assuming no imprinting.

SE of heritabilities between 0.006 and 0.008.



Figure 1: Video image of a carcass. The bending of light stripes is used to cover information on color and conformation (picture provided by E+V Technology GmbH, 16515 Oranienburg, Germany).

Effects in the Model

The estimation of imprinting effects in carcass traits was based on the following linear model (formula (9)) representing to the standard genetic evaluation of beef traits in German Simmentals (Schild and Niebel, 1998).

$$y_{ijklmnop} = SM_i + GT_j + KN_k + b_1x + b_2x^2 + b_3x^3 + s_l + d_m + yc_n + m_o + e_{ijklmnop} \quad (9)$$

Where y is an observation; SM_i is the fixed effect of the interaction between farm and slaughterhouse; GT_j is the fixed effect of birth type (single-, twin- and multiple births); KN_k is the fixed effect of calving number of the dam, subdivided into first, second and all later

births; b_1, b_2, b_3 are linear, quadratic and cubic regressions on slaughter age (x); s_l is the random additive genetic effect as sire; d_m the random additive genetic effect as dam; yc_n is the random y-chromosomal effect, and m_o the random mitochondrial effect; $e_{ijklmnop}$ is the random residual.

The paternal inheritance in the pedigree was traced back until the very first male founder was identified. The number of this male founder was assigned to each observation as the corresponding y-chromosomal effect. It was assumed that the variance of the y-chromosomal effects was $I\sigma_{yc}^2$, where I is an identity matrix with dimension YC , and YC is the number of distinct founder Y-chromosomes in the pedigree. In the same manner the mitochondrial inheritance was traced back in maternal lines. The variance of the mitochondrial effects was assumed $I\sigma_{mi}^2$ where I is an identity matrix with dimension MI , and MI is the number of distinct founder mitochondria in the pedigree.

The ASReml program was used for variance component estimation (Gilmour et al., 2006); approximative standard errors for heritabilities and other functions of variance components were derived by applying the delta method.

Estimation of Imprinting Effects

Estimates for the imprinting effects of all animals can be derived as differences between estimated breeding values as sire and as dam. For animal i in the pedigree,

$$\hat{d}_i = \hat{a}_d^i - \hat{a}_s^i \quad (10)$$

with the corresponding precision

$$\begin{aligned} \text{Var}(\hat{d}_i - d_i)^{-1} = \\ \left[\text{Var}(\hat{a}_s^i - a_s^i) + \text{Var}(\hat{a}_d^i - a_d^i) - 2\text{Cov}(\hat{a}_s^i - a_s^i, \hat{a}_d^i - a_d^i) \right]^{-1}, \end{aligned} \quad (11)$$

the prediction error variances and covariances of both genetic effects for each animal were retrieved from the inverted left-hand side of the mixed model equations as described for AS-Reml in Gilmour et al. (2004) and Welham et al. (2004).

Genetic correlations between imprinting effects of different traits were not directly estimated. For multiple traits it is however important to know if their imprinting effects are concordant in the same direction or not. To get an approximate picture, we calculated pair-wise correlations between estimated imprinting effects for all traits with a significant imprinting variance.

RESULTS AND DISCUSSION

Genetic Parameters

For 10 traits shown in Table 2 (page: 32) we found significant imprinting variances. Imprinting variance σ_i^2 of these traits accounted for 8 to 25% of the total additive genetic variance. Killing out percentage and fat grade reached the greatest proportion with more than 24% imprinting variance, followed by meat percentage with 18.8%. Most of the estimated imprinting variances took around 11% of the additive genetic variance (strip loin, tenderloin, rose, top-side of the valuable traits and daily BW gain). The traits with the smallest imprinting variance were round and hot round weight with around 8%.

Obviously, the incomplete genetic correlation between both genetic effects (Table 2, page: 32) contributed to the estimated imprinting variances. For the significant traits, these correlations ranged from 0.76 for killing out percentage to 0.92 for round and hot round weight. This correlation tended to decline together with the proportion of the imprinting variance (Table 2; e.g., hot round weight also has the smallest estimated relative imprinting variance of 7.8%).

Table 2: Results from the imprinting analysis for all traits with a significant genetic imprinting variance.

Trait	Genetic SD	Relative imprinting variance, ¹ %	Heritability ²	Correlation ³	Y-chromosomal variance, ⁴ %	Mitochondrial variance, ⁴ %
Beefiness score (1 to 15)	0.74	10.75 (15.22)	0.28	0.89	0.00	0.46
Meat percentage, %	0.43	18.67 (16.66)	0.26	0.81	0.04	0.08
Weight of strip loin, kg	0.43	11.87 (17.76)	0.22	0.88	0.00	1.78
Round weight, kg	2.96	7.82 (15.95)	0.24	0.92	0.00	1.97
Hot round weight, kg	2.26	7.76 (15.78)	0.24	0.92	0.00	1.97
Topside weight, kg	1.31	11.53 (17.68)	0.21	0.89	0.00	1.44
Tenderloin weight, kg	1.17	11.91 (18.12)	0.20	0.88	0.00	1.58
Rose weight, kg	0.16	11.66 (16.84)	0.24	0.88	0.00	1.96
Killing out percentage, %	1.07	24.56 (19.75)	0.20	0.76	0.00	0.32
Fat score (1 to 15)	0.67	24.77 (20.60)	0.21	0.78	0.00	1.23

- 1) The imprinting variance is expressed relative to the total additive genetic variance (SE in parentheses). The SE for the absolute imprinting variances were all less than 1 % of the estimates.
- 2) SE of heritabilities from the imprinting model are between 0.012 and 0.013.
- 3) Correlations between breeding values as sire and as dam.
- 4) Estimates for y-chromosomal and mitochondrial variances are provided as the percentage of the phenotypic variance (SE for y-chromosomal variances are between 0.0 and 0.001 and SE for mitochondrial variances are between 0.003 and 0.004).

The first comprehensive analysis of animal model heritabilities (i.e., no imprinting was assumed) of a VIA collected data set is presented in Table 1 (page: 28) for all traits. The estimated heritabilities of the traits ranged from 17% for strip loin + weight and pistola cut /slaughter weight, up to 30% for beefiness score. For this form of data ascertainment there are virtually no evaluations, Schild (2004) used only a subset of the dataset for slaughter weight, net daily BW gain, round, strip loin, tenderloin, forequarter, pistola cut, topside, silver side, weight of meat quality grade 1 to 3, meat percentage and total meat. The results of both analyses were very similar, but the heritabilities of net daily BW gain and forequarter in this study were 3% greater. In the literature, few comparable heritability estimates exist for our traits because definition of cuts varies considerably across countries. For American Simmental bulls, Woodward et al. (1992) reported a heritability of 17% for percentage of cutability. This trait is comparable with our killing out percentage (22%). Crews et al. (2003) estimated for the heritability of the carcass weight 48%. Marshall (1994) summarized the heritabilities of hot carcass weight from many studies. The values ranged between 31 and 68% in different breeds. Compared with these results, we found a slightly smaller value. Engellandt and Tier (2002) estimated a heritability of 34% of net BW gain in German Gelbvieh finishing bulls. We achieved almost the same level in our results (28%). The best possible comparisons are with the results of Engellandt et al. (1999) and Reinsch et al. (1999) because they analyzed Simmental young bulls from beef progeny field tests. The design is similar to our data set. The heritabilities for net BW gain ranged from 12 to 15%. Reinsch et al. (1999) estimated for E-U-R-O-P (European Union meat grading system) a heritability of 20%. This trait it is comparable with our beefiness grade (31%). Smaller heritabilities found earlier might have been caused by poorer data quality (scale with only 5 grades instead of 15) and application of a threshold-model, reducing the validity of such comparisons.

Heritabilities in Table 2 (page: 32) were calculated for all significant traits in the imprinting model. The estimated heritabilities ranged from 20 to 28%, beefiness grade showed the high-

est one. In comparison with heritabilities of the animal model, current heritability estimates were less. Engellandt and Tier (2002) noticed the same result. Heritabilities of the traits from an imprinting model dropped down. Engellandt and Tier found a reduction of 15% for pelvic and of 30% for kidney fat in a German Gelbvieh finishing bulls. The reduction of the heritability for meat percentage was 20%.

The heritability for our fat grade showed a reduction of 4%, the meat percentage of 1%. However, direct comparison with reduction of the heritabilities found by Engellandt and Tier (2002) is not possible, because they used $h_M^2 = \sigma_M^2 / \sigma_p^2$ (i.e., the ratio between Mendelian and phenotypic variance) as their definition of heritability. When recalculating heritabilities with our definition $h_a^2 = \sigma_a^2 / \sigma_p^2$ with the estimated variance components of Engellandt and Tier (2002), we found the reductions of the imprinting-model heritabilities compared to the animal model heritabilities smaller than originally reported: the reduction for meat percentage dwindled to 13%, for pelvic fat to 4% and for kidney fat to 21%. These reductions are still larger than our results, what may also be due to the generally greater level of heritabilities for those traits.

Variation of y-chromosomal and mitochondrial inheritance (Table 2, page: 32) usually turned out to be small, whereby the variances for the mitochondrial inheritance were on a greater level as the variances of y-chromosomal inheritance. No information about mitochondrial inheritance in carcass traits seems to be present in literature. The influence of the y-chromosomal inheritance in carcass traits was analyzed by Reinsch et al. (1999) and Engellandt and Tier (2002). In agreement with our results, no significant impacts of the y-chromosomal inheritance in carcass traits were found at that time.

The estimated imprinting variances show that imprinting noticeably affects slaughter traits. For this reason they should not be neglected. Evaluation of paternal as well as maternal breed-

ing values would account for this problem. In this respect paternal breeding values would improve the validity of genetic proofs if specific bulls are used as terminal sires.

Parental Contributions to the Imprinting Variance

Different parental contributions to the expression of carcass traits are shown by Figure 2 (page: 37). The x-axis indicates the proportion of the total imprinting variance, on the left side the maternal contribution $\left[\frac{(\sigma_d^2 - \sigma_{sd})}{\sigma_i^2} \right]$, on the right side the paternal contribution $\left[\frac{(\sigma_s^2 - \sigma_{sd})}{\sigma_i^2} \right]$.

Figure 2 contains all significant traits. Greater maternal contribution ($\sigma_d^2 - \sigma_{sd} > \sigma_s^2 - \sigma_{sd}$) is shown in the upper area (hot round weight, round, topside, rose, tenderloin). The maternal contribution on hot round weight added up to 94 %, whereas the weight of the (cold) round, topside, rose and tenderloin achieved values of 87, 78, 72 and 66%, respectively. Beefiness score (31%) and killing out percentage (30%) showed a minor maternal contribution ($\sigma_s^2 - \sigma_{sd} > \sigma_d^2 - \sigma_{sd}$), whereas an exclusive paternal contribution was found for fat score.

Two of the traits had almost the same maternal and paternal contribution to the imprinting variance; meat percentage had 53% and strip loin 43% maternal contribution. The traits meat percentage and strip loin almost exclusively attain their imprinting variances from the incomplete genetic correlation. In all other traits the estimated imprinting variances were caused both by the incomplete genetic correlation and by the difference in the parental contribution.

In an extreme example we found only one parental contribution like in the data set for fat score ($\sigma_d^2 - \sigma_{sd} = 0$). In this case it is possible to use the traditional models with 1 gametic effect for 1 parent like Engellandt and Tier (2002), who found high paternal imprinting effects for pelvic and kidney fat weight. Our results suggest that the imprinting variance for fat grade is only affected by paternal alleles and are in good agreement with Engellandt and Tier

(2002), who reported significant imprinting effects from their analysis of fat grade from a less complex model. For any other analyzed trait there is evidence that both kinds of parental origin contribute to the imprinting variance, if significant.

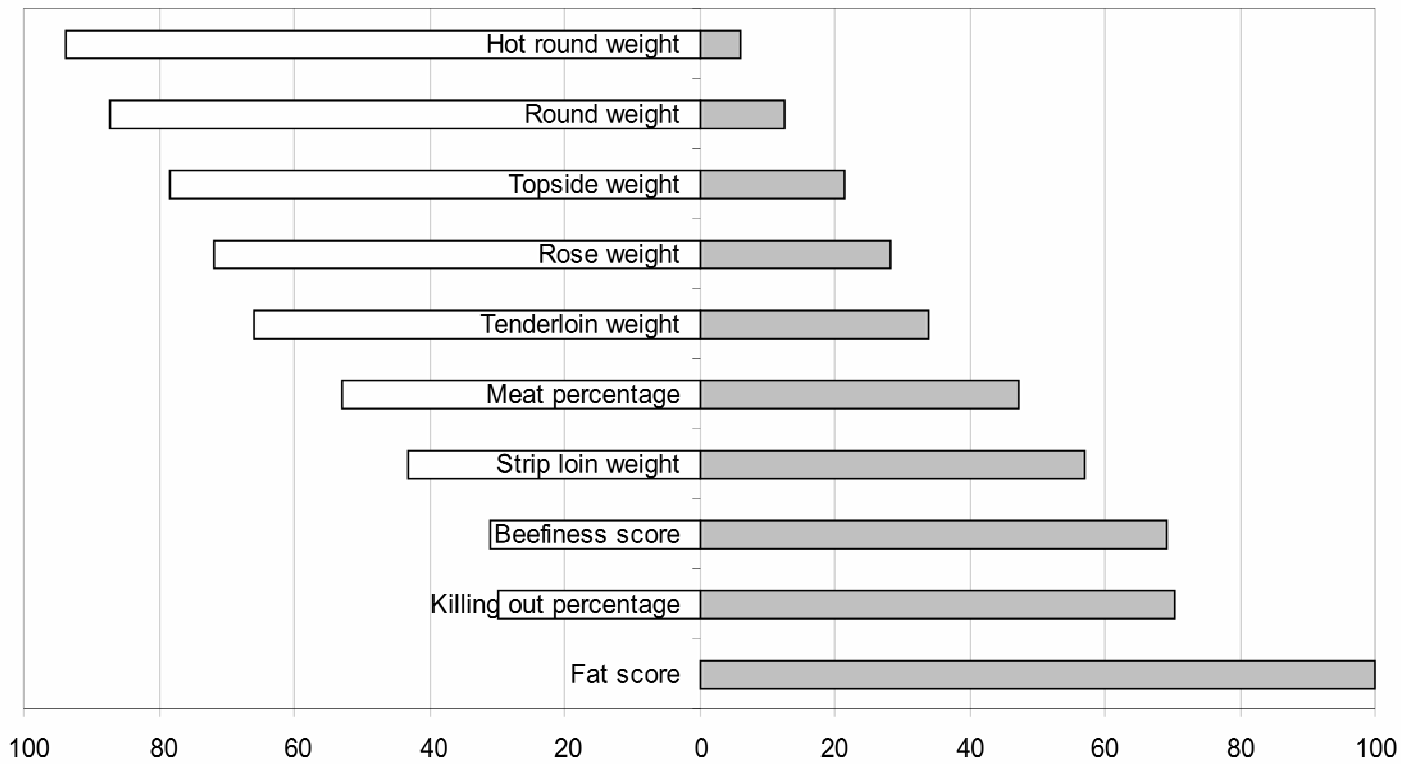


Figure 2: Parental contributions to imprinting variance of significant traits (expressed in percentage of the total imprinting variance): Maternal contributions on the left, paternal contributions on the right.

Individual Estimates of Imprinting Effects

As already mentioned the estimated imprinting effects were calculated as the difference between the paternal and maternal breeding values. Figure 3 shows the individual estimates of imprinting effects [formula (10), horizontal axis] of all animals in the pedigree for the example traits killing out percentage, meat percentage, fat grade, and strip loin weight. The vertical axis represents the log-precision of the estimates [formula (11)].

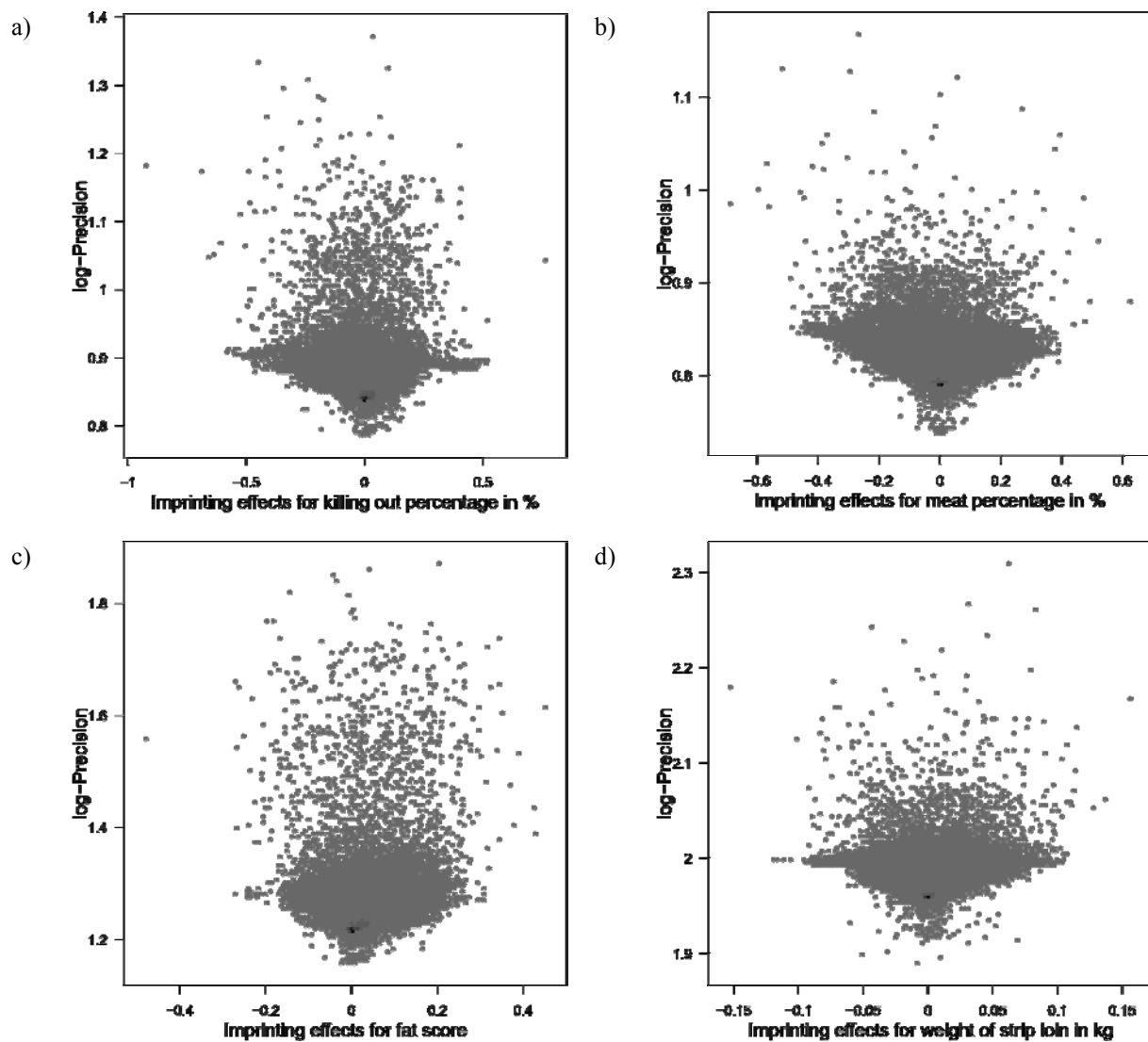


Figure 3: Scatter plots of imprinting effects (estimated as differences between genetic effects as sire and as dam) and their log-precision. The traits are killing out percentage (a), meat percentage (b), fat score (c) and weight of strip loin (d). Units for log-precisions are $\ln(\%)^{-2}$ (a and b) and $\ln(\text{kg})^{-2}$ (d).

Most of the estimates are concentrated around zero irrespective of their precision, in accordance with their zero expectation. In the lower part of each plot where the precision of esti-

mates is relatively poor, we observe only small imprinting effects because the estimates are regressed to the mean because of lack of information (e.g., for animals of older generations). All displayed example traits show a similar pattern. With increasing precision the spread of estimates becomes larger. The estimated imprinting effect for killing out percentage reached up to 0.5% and for fat grade up to 0.4 units. Meat percentage showed differences of more than 0.6% and strip loin weight of more than 150g.

Genetic Correlation Between Imprinting Effects

Because we performed a series of univariate analyses, there were no results for the genetic correlations between imprinting effects of different traits. As a convenient surrogate for a comprehensive multi-trait analysis pair-wise correlations between estimates for imprinting effects of different traits were computed and collected in Table 3 (page: 40). Only traits with a significant imprinting variance were included. Many of these correlations were low to moderate, but the traits strip loin, tenderloin and round weight showed high correlations between 0.75 and 0.98, indicating a high degree of concordance of the imprinting effects of valuable cuts from the back and the hindquarter. As parts of the round, rose and topside also fit into this pattern. A rough estimate for the total imprinting variance of the summed weight of strip

loin, tenderloin and round was obtained by $\hat{\sigma}_{i,total}^2 = \sum_{i=1}^3 \hat{\sigma}_i^2 + \sum_{i=1}^3 \sum_{\substack{j=1 \\ j \neq i}}^3 \hat{r}_{ij} \hat{\sigma}_i \hat{\sigma}_j$, where r_{ij} is the cor-

relation between estimated imprinting effects for traits i and j ; resulted in a SD of 1.13 kg. It is interesting to note that the pattern of high correlations between imprinting effects for valuable cuts from the back and the hindquarter corresponds well with the effects of the *callipyge* mutation in sheep, which induces muscular hypertrophy in the same parts of the body.

Significance Testing

We used the REML likelihood ratio test [formula (8)] with $\chi_{df=2}^2$ and $\alpha=5\%$ to show the significance of the imprinting variance. In 10 cases the null hypothesis was rejected, that means the imprinting model represents the better model for these traits. The exact χ^2 distribution of $RLRT \sim p\chi_1^2 + (1-p)\chi_2^2$ is a mixture with unknown proportion p , which can, in principle, be estimated by applying a bootstrap method (Crainiceanu and Ruppert, 2004). This was, however, computationally unfeasible for our large data set. Therefore we decided to treat $p = 0$ and used a conservative significance test with 2 df.

Separation of Variance Components

The covariance structure of the y-chromosomal and the mitochondrial variances differs from other random effects (diagonal matrices: $I\sigma_{yc}^2$ and $I\sigma_{mi}^2$ with proper dimensions) thus their separation is possible. The separation of the genetic maternal effects variance is, however, different, if such a variance exists. The variance of the observations Y can in this case be

$$\text{written as: } \text{Var}(Y) = M \left[\sigma_m^2 + 2\frac{1}{2}\sigma_{m,d} + \frac{1}{2}\sigma_d^2 \right] + S \frac{1}{2}\sigma_s^2 + 2C \left[\frac{1}{2}\sigma_{m,s} + \frac{1}{2}\sigma_{sd} \right] + I\sigma_e^2,$$

where M , S and C are matrices derived from the design matrices for genetic maternal effects, genetic effects as sire, and genetic effects as dam and I is an identity matrix. Matrix M has an associated variance component, which is composed by the gametic variance as dam σ_d^2 plus the genetic maternal variance σ_m^2 and the covariance between maternal effects and genetic effects as dam $\sigma_{m,d}$. Associated with C is a function of the covariance between genetic maternal effects and genetic effects as sire $\sigma_{m,s}$ and the covariance between genetic effects as sire and as dam σ_{sd} as mentioned in Neugebauer et al. (2009). Thus our results for the imprinting variances can be interpreted as upper bounds and may be contaminated by a

maternal variance contribution. But even in this case the model with 2 breeding values per animal remains applicable and the problem for the breeder can be solved by allowing for 2 parent-specific breeding values per animal.

Conclusion

There is evidence that some parts of the additive genetic variance in beef traits are influenced by imprinting effects. Genetic evaluation could account for such imprinting by estimation of 2 parental breeding values per animal. This might become practically important in particular when bulls are used as terminal sires. It is possible to derive the imprinting variance as the variance of the difference between both breeding values per animal. In a Simmental data set we found 10 beef traits that are significantly influenced by imprinting, whereby between 8 and 25% of the total additive genetic variance was attributed to be induced by imprinted loci.

Acknowledgement

We thank E+V Technology GmbH, 16515 Oranienburg, Germany for their helpful information about VIA-devices and for providing Figure 1. We thank W. Brandscheid for his help with the DLG cuts procedure and R. Pfuhl, who provided assistance for the systematic of carcass traits.

LITERATURE CITED

- Barlow, D. P., 1997: Competition - a common motif for the imprinting mechanism? *EMBO J*, 16. 6899-6905.
- Cockett, N. E., Jackson, S. P., Shay, T. L., Nielsen, D., Moore, S. S., Steele, M. R., Barendse, W., Green, R. B., Georges, M., 1994: Chromosomal localization of the callipyge

- gene in sheep (*ovis aries*) using bovine DNA markers. Proc. Natl. Acad. Sci. USA 91: 3019-3023.
- Crainiceanu, C. M., Ruppert, D., 2004: Likelihood ratio tests in linear mixed models with one variance component. J. R. Statist. Soc. B 66(1): 165-185.
- Crews Jr., D. H., Pollak, E. J., Weaber, R. L., Quaas, R. L. Lipsey, R. J., 2003: Genetic parameters for carcass traits and their live animal indicators in Simmental cattle. J. Anim. Sci. 81: 1427-1433.
- de Koning, D. J., Harlizius, B., Rattink, A. P., Groenen, M. A. M., Brascamp, E. W., van Arendonk, J. A. M., 2001a: Detection and characterization of quantitative trait loci for meat quality traits in pigs. J. Anim. Sci. 79: 2812-2819.
- de Koning, D. J., Rattink, A.P., Harlizius, B., Groenen, M. A. M., Brascamp, E. W., van Arendonk, J. A. M., 2001b: Detection and characterization of quantitative trait loci for growth and reproduction traits in pigs. Livestock Production Science 72: 185-198.
- de Koning, D. J., Rattink, A. P., Harlizius, B., van Arendonk, J. A. M., Brascamp, E. W., Groenen, M. A. M., 2000: Genome-wide scan for body composition in pigs reveals important role of imprinting. Proc. Natl. Acad. Sci. USA 97, 7947-7950.
- De Vries, A. G., Kerr, R., Tier, B., Long, T., Meuwissen T. H. E., 1994: Gametic imprinting effects on rate and composition of pig growth. Theor. Appl. Genet. 88: 1037-1042.
- Engelland, T., Reinsch, N., Schild, H. J., Kalm, E., 1999: Progeny test for beef traits in German Simmental – organization, genetic parameters and breeding values. J. Anim. Breed. Genet. 116: 47-60.
- Engelland, T., Tier, B., 2002: Genetic variances due to imprinted genes in cattle. J. Anim. Breed. Genet. 119 (3): 154-165.
- Essl, A., Voith, K., 2002: Genomic imprinting effects on dairy- and fitness-related traits in cattle. J. Anim. Breed. Genet. 119 (3): 182-189.

- Gibson, J. P., Kennedy, B. W., Schaeffer, L. R., Southwood, O. I., 1988: Gametic models for estimation of autosomally inherited effects that are expressed only when received from either male or female parent (abstract). *J. Dairy Sci.* 71 (suppl 1): 143.
- Gilmour, A. R., Cullis, B. R., Welham, S. J., Gogel, B. J. and Thompson, R., 2004: An efficient computing strategy for prediction in mixed linear models. *Computational Statistics and Data Analysis* 44, 571-586.
- Gilmour, A.R., Gogel, B.J., Cullis, B.R., and Thompson, R., 2006: ASReml User Guide Release 2.0 VSN International Ltd, Hemel Hempstead, HP1 1ES, UK.
- Hill, W.G. and Keightley, P.D., 1988: Interaction between molecular and quantitative genetics. pp. 41-55 in *Advances in Animal Breeding*, edited by Korver, S., van der Steen, H.A.M., van Arendonk, J.A.M., Bakker, H., Brascamp, E.W., and Dommerholt, J.. Proceedings of the World Symposium in Honour of Professor Politiek, RD, Wageningen, The Netherlands, 11-14 September 1988. PUDOC, Wageningen, The Netherlands.
- Jeon, J.-T., Carlborg, Ö., Törnsten, A., Giuffra, E., Amarger, V., Chardon, P., Andersson-Eklund, L., Andersson, K., Hansson, I., Lundström, K., Andersson, L., 1999: a paternally expressed QTL affecting skeletal and cardiac muscle mass in pigs maps to the *IGF2* locus. *Nature Genetics* 21: 157-158.
- Jirtle, R. L. and Weidman, J. R., 2007: Imprinted and more equal. *Am. Sci.* 95: 143-149.
- Marshall, D. M., 1994: Breed differences and genetic parameters for body composition traits in beef cattle. *J. Anim. Sci.* 72: 2745-2755.
- Neugebauer, N., Luther, H., Reinsch, N., 2009: Parent-of-origin effects cause genetic variation in pig performance traits. (accepted for publication in *Animal*).
- Nezer, C., Moreau, L., Brouwers, B., Coppieters, W., Detilleux, J., Hanset, R., Karim, L., Kvasz, A., Leroy, P., Georges, M., 1999: An imprinted QTL with major effect on

- muscle mass and fat deposition maps to the *IGF2* locus in pigs. *Nature Genetics* 21: 155-156.
- Quaas, R. L., Pollak, E. J., 1980. Mixed model methodology for farm and ranch beef cattle testing programs. *Journal of Animal Science* 51, 1277-1287.
- Reik, W., Walter, J., 1998: Imprinting mechanisms in mammals. *Curr. Opin. Genet. Dev.* 8: 154-164.
- Reinsch, N. and Guiard, V.: Estimable genetic covariance components under mixed Mendelian and imprinted inheritance (in preparation).
- Reinsch, N., Engellandt, T., Schild, H. J., Kalm, E., 1999: Lack of evidence for bovine Y-chromosomal variation in beef traits. A Bayesian analysis of Simmental data. *J. Anim. Breed. Genet.* 116: 437-445.
- Schaeffer, L. R., Kennedy, B. W., Gibson, J. P., 1989: The inverse of the gametic relationship matrix. *J. Dairy Sci.* 72(5): 1266-1272.
- Scheper, J., Scholz, W., 1985: DLG-Schnittführung für die Zerlegung der Schlachtkörper von Rind, Kalb, Schwein und Schaf. DLG Verlag, Frankfurt/M.
- Schild, H. J. and Niebel, E., 1998: Zuchtwertschätzung auf Fleischleistung. Aktueller Stand und neue Entwicklungen der Zuchtwertschätzung beim Rind, Verden.
- Schild, H. J., 2004: Apparative Klassifizierung beim Rind: Informationen zum Sachstand aus der Projektgruppe der DGfZ "Klassifizierung Rind". *Züchtungskunde* 76: 1-5.
- Self, S. G., Liang, K., 1987: Asymptotic properties of maximum likelihood estimators and likelihood ratio tests under nonstandard conditions. *J. Am. Stat. Assoc.* 82: 605-610.
- Smit, M., Segers, K., Carrascosa, L. G., Shay, T., Baraldi, F., Gyapay, G., Snowder, G., Georges, M., Cockett, N., Charlier, C., 2003: Mosaicism of solid gold supports the causality of a noncoding A-to-G transition in the determinism of the callipyge phenotype. *Genetics* 163: 453-456.

- Tier, B. and Sölkner, J., 1993: Analysing gametic variation with an animal model. *Theor. Appl. Genet.* 85: 868-872.
- Van Laere, A.-S., Nguyen, M., Braunschweig, M., Nezer, C., Colette, C., Moreau, L., Archibald, A. L., Haley, C. S., Buys, N., Tally, M., Andersson, G., Georges, M., Andersson, L., 2003: A regulatory mutation in *IGF2* causes a major QTL effect on muscle growth in the pig. *Nature* 425: 823-836.
- Vuocolo, T., Byrne, K., White, J., McWilliam, S., Reverter, A., Cockett, N. E., Tellam, R. L., 2007: Identification of a gene network contributing to hypertrophy in *callipyge* skeletal muscle. *Physiol Genomics* 28: 253-272.
- Welham, S. J., Cullis, B. R., Gogel, B. J., Gilmour, A. R. and Thompson, R. 2004: Prediction in linear mixed models. *Australian and New Zealand Journal of Statistics* 46: 325-347.
- Woodward, B. W., Pollak, E. J., Quaas, R. L., 1992: Parameter estimation for carcass traits including growth information of Simmental beef cattle using restricted maximum likelihood with a multiple-trait model. *J. Anim. Sci.* 70: 1098-1109.

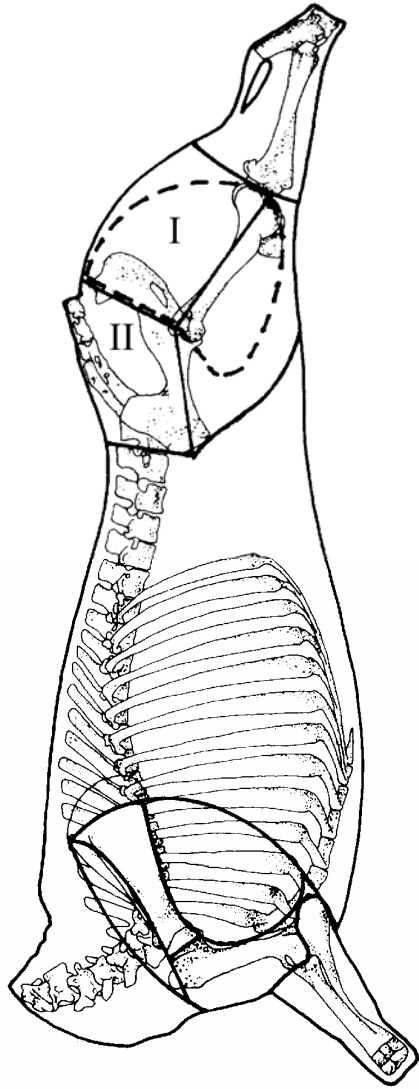


Figure 4: Meat segmentation (adapted from Scheper and Scholz, 1982): I) Topside, II) Rose (supplemental material).

Chapter 2

Parent-of-origin effects cause genetic variation in pig performance
traits

Accepted for publication in *Animal*

Parent-of-origin effects cause genetic variation in pig performance traits

N. Neugebauer*, H. Luther[†], N. Reinsch*

*) Forschungsinstitut für die Biologie landwirtschaftlicher Nutztiere, FBN, Forschungsbereich Genetik und Biometrie, Wilhelm-Stahl-Allee 2, 18196 Dummerstorf, Germany

†) SUISAG, AG für Dienstleistungen in der Schweineproduktion, Allmend, CH-6204 Sempach, Switzerland

Abstract

In order to assess the relative importance of genomic imprinting for the genetic variation of traits economically relevant for pork production, a data set containing 21,209 records from Large White pigs was analysed. A total of 33 traits for growth, carcass composition and meat quality were investigated. All traits were recorded between 1997 and 2006 at a test station in Switzerland and the pedigree included 15,747 ancestors.

A model with two genetic effects for each animal was applied: the first corresponds to a paternal and the second to a maternal expression pattern of imprinted genes. The imprinting variance was estimated as the sum of both corresponding genetic variances per animal minus twice the covariance. The null hypothesis of no imprinting was tested by a restricted maximum likelihood ratio test with two degrees of freedom. Genomic imprinting significantly contributed to the genetic variance of 19 traits. The proportion of the total additive genetic variance that could be attributed to genomic imprinting was of the order of between 5% and 19%.

Introduction

Genomic imprinting is an epigenetic process and depends on the sex of the parent. Imprinted genes are expressed at a lower level than the copy from the other parent. Silencing of one pa-

rental allele may be complete or, in the case of partial imprinting, there may be some remaining activity and also an effect on the phenotype. In each generation the imprint is newly established during gametogenesis. The first results on how much imprinted genes contribute to genetic variation in livestock were presented by De Vries *et al.* (1994) with estimates of genetic variance components of carcass and growth traits in pigs. They found that about 5% of the phenotypic variance in back fat thickness and up to 4% of growth rate variance were affected by imprinting. For the analysis De Vries *et al.* (1994) used an animal model augmented by either an additional paternal or maternal gametic effect, an approach which was adopted by nearly all researchers in this field. The same method was used by Stella *et al.* (2003), who reported very small imprinting effects for reproduction traits in pigs. Engellandt and Tier (2002) found a significant paternal gametic variance for two fatness traits and, economically most important, for carcass meat content of German Gelbvieh finishing bulls. Another approach was used by Essl and Voith (2002). They analysed the data twice, first with a sire model and second with a dam model. By comparing variance components from both analyses they inferred imprinting effects for protein content and days open. Beyond that, several studies exist in which significant imprinted QTL effects for body compositions, meat quality, growth and reproduction traits were mapped in pig F_2 families (e.g. De Koning *et al.* 2000; 2001a and 2001b).

In order to assess the relative importance of genomic imprinting for the genetic variation of traits economically relevant for pork production, we applied a model including two random genetic effects for each animal, as outlined in Reinsch and Guiard (in preparation) and applied in Neugebauer *et al.* (2009) for slaughter traits in cattle. Hill and Keightley (1988) proposed an approach to estimate three uncorrelated variance components simultaneously (*Mendelian*, paternally imprinted and maternally imprinted), which corresponds to a scenario with loci either fully imprinted (maternal or paternal) or not imprinted at all and results in zero covariances between the corresponding random genetic effects. With partial imprinting their genetic

covariance matrix is, however, no longer diagonal and it would become necessary to estimate covariances, which is not possible. Our parameterisation circumvents this problem by defining the variance components in a different way. The model allows for paternal and maternal imprinting as well as of any combination of full and partial imprinting simultaneously. This model was applied for the analysis of a comprehensive dataset originating from the Swiss herd book breeding program.

Material and Methods

Principle of analysis

Inheritance was modelled by two random genetic effects per animal: one for the genetic effect as sire, that is half of the breeding value as sire, and one for the genetic effect as dam, that is half of the breeding value as dam. The model in matrix notation is

$$Y = X\beta + Z_s a_s + Z_d a_d + e \quad (1)$$

where assumptions on the covariance of random effects are

$$\text{Var} \begin{bmatrix} a_s \\ a_d \\ e \end{bmatrix} = \begin{bmatrix} \frac{1}{2} A\sigma_s^2 & \frac{1}{2} A\sigma_{sd} & 0 \\ \frac{1}{2} A\sigma_{sd} & \frac{1}{2} A\sigma_d^2 & 0 \\ 0 & 0 & W\sigma_R^2 \end{bmatrix}. \quad (2)$$

Resulting in the following general mixed model equations

$$\begin{bmatrix} X'W^{-1}X & X'W^{-1}Z_s & X'W^{-1}Z_d \\ Z_s'W^{-1}X & Z_s'W^{-1}Z_s + A^{-1}\alpha_1 & Z_s'W^{-1}Z_d + A^{-1}\alpha_2 \\ Z_d'W^{-1}X & Z_d'W^{-1}Z_s + A^{-1}\alpha_2 & Z_d'W^{-1}Z_d + A^{-1}\alpha_3 \end{bmatrix} \begin{bmatrix} \beta \\ a_s \\ a_d \end{bmatrix} = \begin{bmatrix} X'W^{-1}y \\ Z_s'W^{-1}y \\ Z_d'W^{-1}y \end{bmatrix} \quad (3)$$

where Y is the vector of observations, X is the design matrix for fixed effects with the corresponding solution vector β ; Z_s and Z_d are the design matrices for random genetic effects with solution vectors a_s and a_d ; e is the vector of random residuals. The matrix A is the numerator relationship matrix. Variance ratios are

$$\begin{bmatrix} \alpha_1 & \alpha_2 \\ \alpha_2 & \alpha_3 \end{bmatrix}^{-1} = \frac{1}{2\sigma_R^2} \begin{bmatrix} \sigma_s^2 & \sigma_{sd} \\ \sigma_{sd} & \sigma_d^2 \end{bmatrix}. \quad (4)$$

Genetic variance components are expressed in terms of gametic variance as sire σ_s^2 , gametic variance as dam σ_d^2 and their gametic covariance σ_{sd} . The diagonal matrix W has elements

$$w_i = \left[\frac{\frac{1}{2}\sigma_s^2(1-F_{si}) + \frac{1}{2}\sigma_d^2(1-F_{di}) + \sigma_e^2}{\frac{1}{2}\sigma_s^2 + \frac{1}{2}\sigma_d^2 + \sigma_e^2} \right]^{-1} \quad (5)$$

and adjusts the error variance of each observation to the variation in the *Mendelian* sampling term according to the inbreeding coefficients of both parents. The model, therefore, is a special kind of reduced model (Quaas and Pollak, 1980).

The variance of the difference of the gametic effects as sire and as dam

$$\sigma_s^2 + \sigma_d^2 - 2\sigma_{sd} = \sigma_i^2 \quad (6)$$

corresponds to the imprinting variance σ_i^2 , which is a part of the total additive genetic variance $\sigma_a^2 = \sigma_s^2 + \sigma_d^2$. The other part of the total additive genetic variance is termed as *Mendelian* variance $\sigma_M^2 = \sigma_a^2 - \sigma_i^2 = 2\sigma_{sd}$ which describes the “unimprinted” component of the additive genetic variance (not to be confused with the *Mendelian* sampling variance). Thus, the additive genetic variance is the sum of *Mendelian* and imprinting variance. Moreover, the imprinting variance for a particular trait can be divided into a paternal contribution $\sigma_s^2 - \sigma_{sd}$ and a maternal contribution $\sigma_d^2 - \sigma_{sd}$.

Animals and traits

The data set was provided by the Swiss pig breeding organisation SUISAG. Between 1997 and 2006, 21209 Large White pigs (females and castrates) were fattened and slaughtered at the central test station. The 33 traits were split into three groups: fattening traits (6), carcass traits (24) and meat quality traits (3). A comprehensive overview is given in Table 1 (page: 53). The pedigree included 15747 ancestors of the slaughtered animals with a pedigree depth

of up to 20 generations. The number of sows represented in the data with progeny from a single litter was 5188, 1213 sows had two litters, 284 had three litters and 93 contributed progeny from four or even more litters.

Table 1: An overview of analysed traits: raw means with standard deviations, heritabilities, proportions of the phenotypic variance due to common litter environment, test statistic RLRT for significance of the imprinting variance and corresponding error probabilities.

Trait (unit)	Mean (s.d.)	h^2 ⁽¹⁾	c^2 ⁽²⁾	RLRT ⁽³⁾	p ⁽⁴⁾
Life weight at the end of the test (kg)	104.12 (3.44)	0.12	0.03	1.06	0.589
Average daily gain in lifetime (g/d)	622.20 (45.78)	0.41	0.16	16.94	0.000
Average daily gain during test (g/d)	870.94 (95.13)	0.34	0.07	1.82	0.402
Feed conversion (kg/kg)	2.54 (0.19)	0.40	0.09	8.18	0.017
Average daily feed intake (kg/d)	2.21 (0.24)	0.51	0.05	9.62	0.008
Total feed intake during test (kg)	189.82 (14.73)	0.38	0.07	8.718	0.013
Carcass weight (kg)	82.26 (2.72)	0.12	0.03	0.016	0.602
Carcass length (cm)	96.25 (2.70)	0.75	0.03	0.96	0.619
Percentage of premiums cuts (%)	57.31 (2.52)	0.74	0.02	8.48	0.014
Intramuscular fat content of chop (%)	1.98 (0.60)	0.78	0.04	12.96	0.002
Meat quality (unit)	3.841 (0.29)	0.10	0.04	5.22	0.074
pH 45 minutes <i>post mortem</i>	6.27 (0.18)	0.16	0.03	0.76	0.684
pH 30 hours <i>post mortem</i>	5.43 (0.05)	0.15	0.02	3.46	0.177
Meat reflectance	32.69 (3.19)	0.22	0.03	6.82	0.033
Back fat thickness at ham (cm)	1.43 (0.40)	0.51	0.03	9.66	0.008
Back fat thickness at mid of back (cm)	1.79 (0.34)	0.38	0.04	2.16	0.340
Trimmed back fat (kg)	2.84 (0.55)	0.61	0.03	2.84	0.242
Head (kg)	5.74 (0.33)	0.66	0.03	0.88	0.644

Trait (unit)	Mean (s.d.)	h^2 ⁽¹⁾	c^2 ⁽²⁾	RLRT ⁽³⁾	p ⁽⁴⁾
Head (%)	6.78 (0.36)	0.66	0.03	3.4	0.182
Feet (kg)	0.73 (0.05)	0.88	0.02	8.14	0.017
Trimmed kidney fat (kg)	0.79 (0.17)	0.81	0.01	16.14	0.000
Kidney fat (%)	1.93 (0.40)	0.81	0.01	15.3	0.000
Pork belly (kg)	7.32 (0.48)	0.59	0.03	21.48	0.000
Pork belly (%)	17.82 (0.93)	0.57	0.04	21.84	0.000
Trimmed shoulder fat (kg)	0.81 (0.12)	0.46	0.02	1.44	0.487
Shoulder (kg)	5.07 (0.28)	0.57	0.04	8.44	0.015
Shoulder (%)	12.34 (0.62)	0.60	0.04	8.88	0.012
Pork chop (kg)	10.65 (0.62)	0.61	0.02	7.9	0.019
Pork chop (%)	25.94 (1.29)	0.67	0.02	10.0	0.007
Trimmed ham fat (kg)	1.19 (0.18)	0.59	0.03	2.03	0.363
Ham (kg)	7.81 (0.49)	0.72	0.03	6.8	0.033
Ham (%)	19.03 (1.09)	0.73	0.03	6.4	0.041
Total trimmed fat (%)	11.78 (1.77)	0.67	0.03	2.22	0.330

The number of observations was always 21,209, except total feed intake during test with 17,033

- (1) Heritabilities from models assuming no imprinting, their s.e. were uniformly very close to 0.01
- (2) Fraction of the phenotypic variance due to common litter environment, from models assuming no imprinting, their s.e. were uniformly very close to 0.01
- (3) $2(\text{LogL}(\text{imprinting model}) - \text{LogL}(\text{Mendelian model}))$
- (4) Error probabilities

The structure of the data changed during time. In the beginning up to five animals per litter were tested. Since 2000, test groups consisted mostly of two animals per litter (one female and one male castrate). All pigs were generally slaughtered at 103 kg and cut according to the *Swiss Pig Performance Testing Manual* (internal documentation). The left half of the carcass was divided into 11 cuts and the weight of each cut was recorded. In addition to the percentage of the cut in comparison to the complete weight of the carcass side was calculated for selected cuts. Thus, for ham, pork belly, kidney fat, shoulder and pork chop there were two observations in the data set, weight of the cut in kilogram and as a percentage.

Models

A linear model with one of three different combinations of fixed effects was used according to the kind of trait, thereby following the standard genetic evaluations in the Swiss herd book breeding program. For carcass and fattening traits (except live end weight) the following linear model was employed

$$y_{ijklmnopqr} = S_i + B_j + b_1x_1 + p_k + f_l + l_m + s_n + d_o + yc_p + m_q + e_{ijklmnopqr} \quad (9)$$

where y is the vector of observations, S_i is the fixed effect of sex, B_j is the fixed effect of the interaction between barn and cycle, b_1 is the linear regression on carcass weight (x_1), p_k is the random effect of pen, f_l is the random effect of the interaction between farm of origin and year, l_m is the random effect of litter, s_n is the random additive genetic effect as sire, d_o the random additive genetic effect as dam, yc_p is the random y-chromosomal effect and m_q the random mitochondrial effect and $e_{ijklmnopqr}$ is the random residual.

For meat quality traits the interaction between barn and cycle was replaced by SD_j the fixed effect of the slaughter day as this contemporary group effect is far more important for these traits than the interaction between barn and cycle.

$$y_{ijklmnopqr} = S_i + SD_j + b_1x + p_k + f_l + l_m + s_n + d_o + yc_p + m_q + e_{ijklmnopqr} \quad (10)$$

In the analysis of carcass weight and live end weight the linear regression on carcass weight was replaced by the linear regression (b_2) on age (x_2)

$$y_{ijklmnopqr} = S_i + SD_j + b_2x_2 + p_k + f_l + l_m + s_n + d_o + yc_p + m_q + e_{ijklmnopqr} \quad (11)$$

y-chromosomal and mitochondrial genetic effects were included to avoid inflated estimates of the imprinting variance as a consequence of contributions of the y-chromosomal or mitochondrial variance to the covariance between paternal or maternal half sibs. For each observation paternal inheritance in the pedigree was traced back until the first male founder was identified. The number of this male founder was assigned to this observation as the corre-

sponding y-chromosomal effect. It was assumed that the variance of the y-chromosomal effects was $I\sigma_{yc}^2$, where I is an identity matrix with dimension YC , and YC is the number of distinct founder y-chromosomes in the pedigree. We used the same system for the mitochondrial inheritance and traced back the maternal inheritance. The variance of the mitochondrial effect was assumed as $I\sigma_{mi}^2$ where I is an identity matrix with dimension MI , and MI is the number of distinct founder mitochondria in the pedigree.

The ASReml program was used for variance component estimation (Gilmour *et al.*, 2004). Approximative standard errors for heritabilities and other functions of variance components were derived by applying the delta method.

Hypothesis testing

The matrix S

$$S = \begin{bmatrix} \sigma_s^2 & \sigma_{sd} \\ \sigma_{sd} & \sigma_d^2 \end{bmatrix} \quad (12)$$

contains the genetic covariance components. Under the null hypothesis of no imprinting ($\sigma_i^2 = 0$) all variances are equal and S is not positive definite. Two models were fitted to each trait in our data set, the imprinting model and an equivalent non-imprinting model (*Mendelian* model). From the restricted maximum (REML) log-likelihood of both models a REML likelihood ratio test (RLRT) with two degrees of freedom was calculated to test the hypothesis of imprinting against the absence of imprinting.

If λ_{R2} is the REML log-likelihood of the more general model and λ_{R1} is the REML log-likelihood of the restricted model (REML log-likelihood under the null hypothesis), the RLRT is given by

$$RLRT = 2(\lambda_{R2} - \lambda_{R1}). \quad (13)$$

With the assumption of between-subject independence (in this case no relationships between animals), the RLRT under the null hypothesis is asymptotically distributed as a 1:1 mixture of two χ^2 distributions with a single and two degrees of freedom (Self and Liang, 1987). Since this condition is not fulfilled and the ratio of the mixture is unknown in our case we used a conservative test with a χ^2 distribution with two degrees of freedom to minimize the risk of falsely rejecting the null hypothesis.

Estimation of imprinting effects

The differences between estimated genetic effects as sire and as dam correspond to the imprinting effect; for animal i in the pedigree,

$$\hat{d}_i = \hat{a}_d^i - \hat{a}_s^i \quad (14)$$

with the corresponding precision

$$\begin{aligned} \text{Var}(\hat{d}_i - d_i)^{-1} = \\ \left[\text{Var}(\hat{a}_s^i - a_s^i) + \text{Var}(\hat{a}_d^i - a_d^i) - 2\text{Cov}(\hat{a}_s^i - a_s^i, \hat{a}_d^i - a_d^i) \right]^{-1} \end{aligned} \quad (15)$$

where the prediction error variances and covariances of both genetic effects for each animal were retrieved from the inverted left-hand side of the mixed model equations as described for ASReml in Gilmour *et al.* (2004) and Welham *et al.* (2004).

Results and Discussion

Genetic parameters

Significant imprinting variances were found for 19 traits (Table 2, page: 61). Imprinting variances of significant traits were between 5% and 19% of the total additive genetic variance. Average daily gain in lifetime reached the highest proportion with nearly 19%. Many of the estimated imprinting variances accounted for around 12% to 14% of the total additive genetic variance (back fat thickness at ham, pork belly (kg) and pork belly (%), feed conversion, average daily feed intake, average daily gain during test). For another group of traits imprinted inheritance contributed a proportion between 6% and 10% to the additive genetic variance (kidney fat (kg), kidney fat (%), shoulder (kg), shoulder (%), ham (kg) and ham (%), percentage of premiums cuts, intramuscular fat content of chop, meat reflectance). Among the traits with the smallest imprinting variance were feet, pork chop (kg) and pork chop (%) with around 5%. Standard errors for the absolute imprinting variances of significantly imprinted traits were all below 1% of the estimates.

The estimated imprinting variances are a result of the incomplete genetic correlation between both genetic effects (Table 2) and the differences between their variances. For significant traits these correlations ranged from 0.86 for pork belly (%) to 0.98 for average daily gain during lifetime. Several traits (e.g. daily gain during test) had a genetic correlation close to one but showed a significant imprinting variance, which is mainly caused by a difference between both genetic variance components.

In the extreme case the genetic correlation could be exactly one with both gametic variances different and a ratio of $f^2 = \sigma_s^2 / \sigma_d^2$, resulting in $\sigma_i^2 = \sigma_s^2(1-f)^2$. It should be mentioned that matrix S would not be positive definite in that case. A reviewer's proposal was to fit gametic models that explicitly account for a perfect genetic correlation by employing a matrix $G^* = DGD\sigma_g^2$ as covariance matrix of gametic effects, where G is the gametic relationship

matrix and D is a diagonal matrix with one and f on alternate rows. The inverse of G^* can be computed from the pedigree for known f . With a proper incidence matrix assigning observations to gametes f could be estimated, for example, by a grid search. For the non-significant traits correlations between both genetic effect were between 0.87 and 0.99 and f -coefficients (derived from estimated variance components) were always smaller than 1.07. It may be expected that we did not miss a significant result by not applying this kind of model because $f = 1.07$ corresponds to less than 1% of the additive genetic variance due to imprinting. When applied to the significant traits it would, however, provide a separate test for a perfect v. a non-perfect genetic correlation between both kinds of breeding values.

Heritability estimates from *Mendelian* models (i.e. assuming no imprinting) ranged from 12% for live end weight up to 88% for feet weight (Table 1); standard errors were very uniform and close to 1%. Reported heritability estimates from the literature vary widely for economically important traits in pigs. Most of the estimates for back fat thickness are between 50% and 70%, for carcass length between 52% and 63%, for intramuscular fat content of chop between 38% and 67% and the results for pH 45 minutes post mortem are between 15% and 40% (e.g. Enfield and Whatley, 1961; Cameron, 1990; Hovenier *et al.*, 1992; Lo *et al.*, 1992; Knapp *et al.*, 1997; Cassady *et al.*, 2002). In general, estimates from the literature tend to be smaller than our estimates.

The imprinting model heritabilities for all significant traits are presented in Table 2, together with their approximative standard errors. The estimated heritabilities ranged from 37% to 73% and were somewhat lower than the corresponding *Mendelian* model heritabilities, whereby reductions were between 1% and 15%. Animal model and imprinting model heritabilities were also compared by Engellandt and Tier (2002). However they used the *Mendelian* genetic variance divided by the phenotypic variance $h_M^2 = \sigma_M^2 / \sigma_p^2$ and found lower values for the imprinting model heritabilities, while we looked at the proportion of the phenotypic vari-

ance, which is explained by the additive genetic variance, that is the sum of *Mendelian* and imprinting variance $h_a^2 = \sigma_a^2 / \sigma_p^2$. Reductions of heritabilities reported by Engellandt and Tier (2002) was up to 30% for kidney fat (pelvic fat, 15% and meat percentage, 20%) in their imprinting analysis of data from German Gelbvieh finishing bulls. Applying our definition of heritability to their results yields smaller reductions of imprinting model heritabilities (kidney fat 21%, pelvic fat 4% and meat percentage 13% relative to the larger animal model estimates).

Generally, results for mitochondrial and y-chromosomal variance components show very small estimates (< 1%, not shown). This is in good agreement with results from quantitative analyses of beef traits (Reinsch *et al.*, 1999; Engellandt and Tier, 2002), who found no significant influence of y-chromosomal inheritance in cattle carcass traits.

Jeon *et al.* (1999), Nezer *et al.* (1999) and Van Laere *et al.* (2003) reported effects of a polymorphism of the imprinted *IGF2* locus on carcass traits in pigs. In the Swiss Large White population this polymorphism seems to be of minor or no importance, since all of about 200 genotyped boars had the same homozygous genotype. Therefore the *IGF2* locus does not contribute to the observed imprinting variances.

Table 2: Results from the imprinting analysis for all traits with a significant genetic imprinting variance.

Trait (unit)	Genetic s.d.	Gametic s.d. as sire	Gametic s.d. as dam	Relative imprinting variance ¹ (%)	Heritability ²	Correlation ³
Average daily gain in lifetime (g/d)	23.82	17.39	16.28	18.55 (16.02)	0.37 (0.02)	0.93 (0.07)
Feed conversion (kg/kg)	0.10	0.071	0.074	12.55 (17.31)	0.38 (0.02)	0.88 (0.05)
Average daily feed intake (kg/d)	0.13	0.091	0.094	12.05 (15.50)	0.46 (0.02)	0.88 (0.04)
Total feed intake during test (kg)	7.81	5.189	5.833	12.29 (17.02)	0.37 (0.02)	0.98 (0.05)
Percentage of premiums cuts (%)	1.68	1.156	1.223	8.99 (12.52)	0.63 (0.02)	0.91 (0.03)
Intramuscular fat content of chop (%)	0.44	0.299	0.321	6.83 (11.37)	0.67 (0.01)	0.93 (0.02)
Meat reflectance	1.31	0.968	0.886	9.68 (20.30)	0.21 (0.01)	0.91 (0.05)
Back fat thickness at ham (cm)	0.23	0.155	0.173	13.63 (13.50)	0.47 (0.02)	0.87 (0.03)
Feet (kg)	0.04	0.028	0.029	4.97 (10.78)	0.73 (0.01)	0.95 (0.01)
Trimmed kidney fat (kg)	0.12	0.083	0.089	8.26 (11.37)	0.68 (0.01)	0.92 (0.02)
Kidney fat (%)	0.29	0.199	0.212	7.99 (11.34)	0.68 (0.01)	0.92 (0.02)
Pork belly (kg)	0.26	0.174	0.192	13.77 (13.75)	0.53 (0.02)	0.87 (0.04)
Pork belly (%)	0.60	0.404	0.444	13.93 (13.92)	0.51 (0.02)	0.86 (0.04)

Trait (unit)	Genetic s.d.	Gametic s.d. as sire	Gametic s.d. as dam	Relative imprinting variance ¹ (%)	Heritability ²	Correlation ³
Shoulder (kg)	0.16	0.105	0.118	8.49 (13.86)	0.51 (0.02)	0.92 (0.04)
Shoulder (%)	0.41	0.274	0.306	9.37 (13.69)	0.53 (0.02)	0.91 (0.04)
Pork chop (kg)	0.32	0.214	0.283	5.2 (12.75)	0.54 (0.02)	0.95 (0.02)
Pork chop (%)	0.83	0.547	0.620	4.62 (12.15)	0.58 (0.02)	0.96 (0.02)
Ham (kg)	0.30	0.210	0.221	9.98 (12.84)	0.62 (0.02)	0.90 (0.03)
Ham (%)	0.78	0.538	0.561	9.53 (12.75)	0.62 (0.02)	0.91 (0.03)

1) the imprinting variance is expressed relative to the total additive genetic variance (s.e. in brackets)

2) heritability is from the imprinting model (s.e. in brackets)

3) estimates for the correlations between breeding values as sire and as dam (s.e. in brackets)

Parental contributions to the imprinting variance

Different parental contributions to the imprinting variance are graphed in Figure 1 (page: 64). The x-axis shows the contribution in percent; the maternal contribution $\left(\left(\sigma_d^2 - \sigma_{sd}\right) / \sigma_i^2\right)$ is on the right, the paternal contribution $\left(\left(\sigma_s^2 - \sigma_{sd}\right) / \sigma_i^2\right)$ is on the left. The traits in the upper area (meat reflectance and average daily gain in lifetime) presented a higher paternal contribution ($\sigma_s^2 - \sigma_{sd} > \sigma_d^2 - \sigma_{sd}$). The paternal contribution for meat reflectance amounted to 96 % and for average daily gain in lifetime to 68%. All other traits showed a higher maternal contribution ($\sigma_s^2 - \sigma_{sd} < \sigma_d^2 - \sigma_{sd}$) to the imprinting variance: average daily feed intake (62%), feed conversion (66%), feet weight (71%), ham percent (72%), ham kilogram (76%), percentage of premiums cuts (81%), pork belly percent (85%), pork belly kilogram (86%), back fat thickness at ham (90%), kidney fat percent (91%), kidney fat kilogram (92%) and average daily gain during test (97%). For the traits intramuscular fat content of chop, shoulder (kilogram and percent) and pork chop (kilogram and percent), the calculation resulted in positive maternal contributions and negative paternal contributions, which is unexpected, but mathematically possible if large differences between both genetic variances occur and the covariances are higher than one of the parental variances. For these traits the covariance was higher than the paternal variance. We assume that the paternal contribution is actually very small or zero and negative estimates for one of the parental contributions are a result of estimation errors. Exactly equal parental contributions or a contribution exclusively from a single parental side are rather special cases.

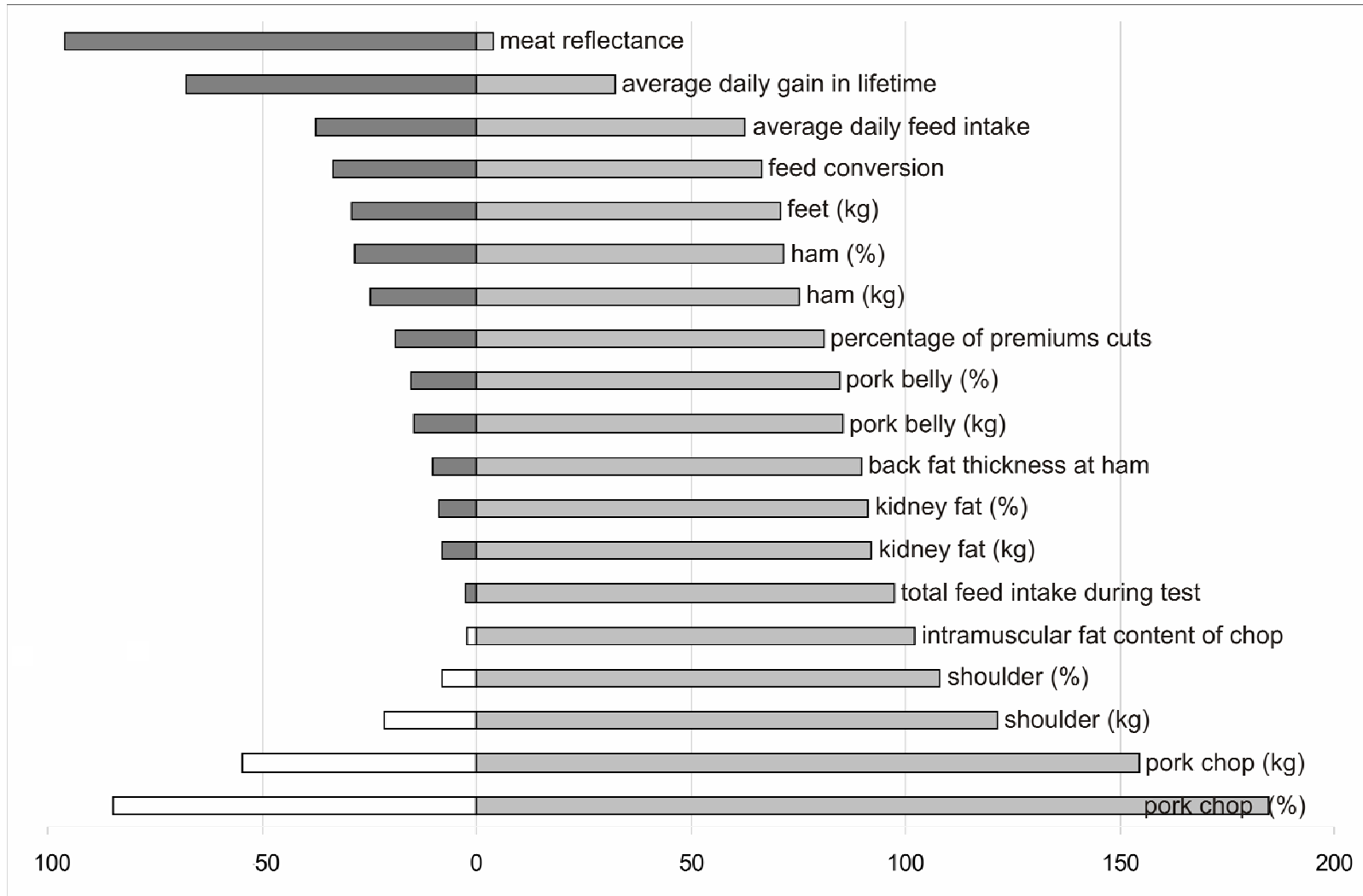
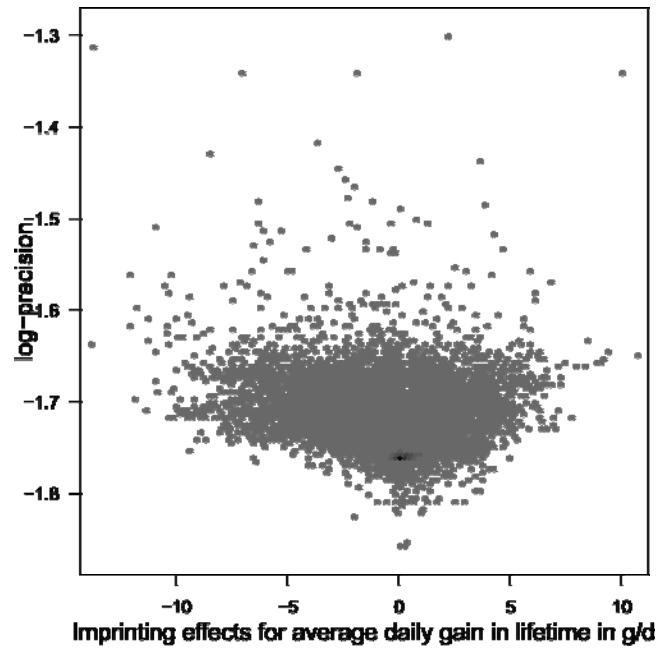


Figure 1: Parental contributions to the imprinting variance of significant traits (expressed in percentage of the total imprinting variance): Paternal contributions on the left, maternal contributions on the right. Negative estimates were obtained for the paternal contributions of the last five traits, therefore the paternal contributions appear empty and the maternal contributions numerically are larger than 100%. The difference of these bars equates to 100%.

Imprinting effects

Estimates of imprinting effects (i.e., differences between paternal and maternal breeding values) for all animals are shown as scatter plots together with their log-precision in Figure 2 for the example traits average daily gain in lifetime and feed conversion.

a)



b)

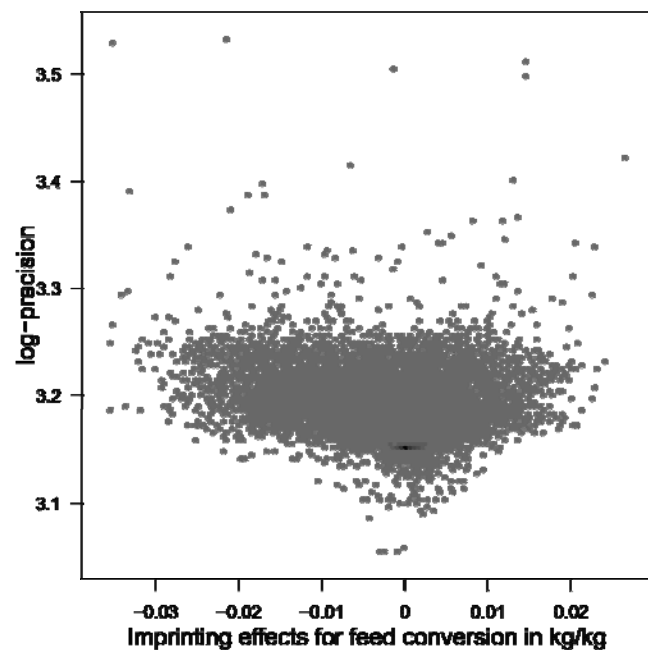


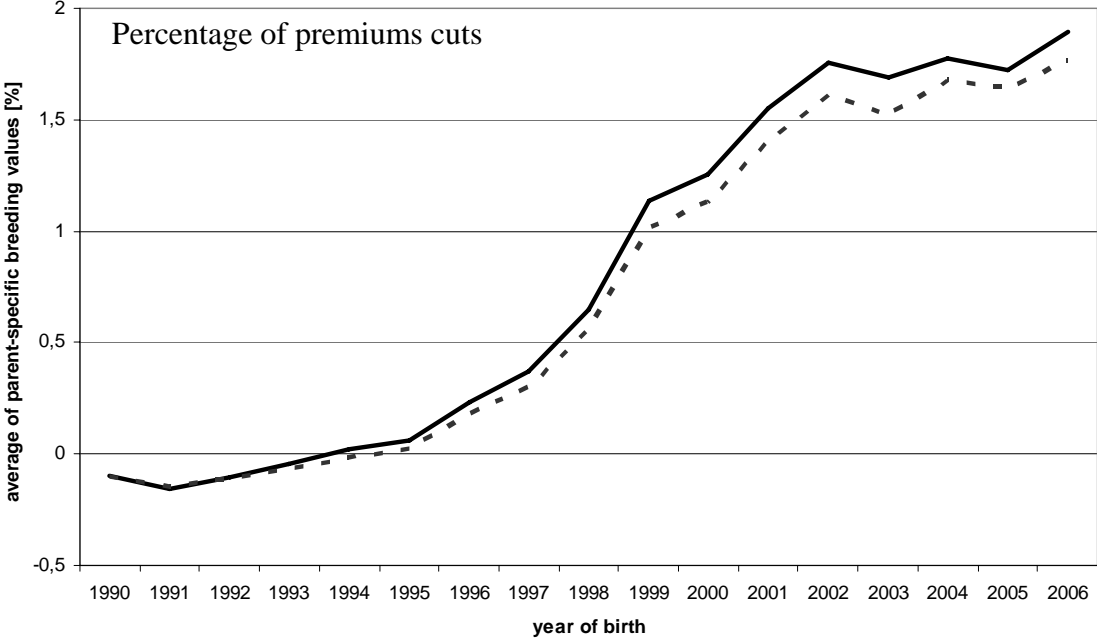
Figure 2: Two-dimensional plot of estimated imprinting effects for all animals in the pedigree and their log-precision for the traits average daily gain in lifetime in g/day (a) and for feed conversion in kg/kg (b). The left side corresponds to higher breeding values as sire. Units for log-precisions are $\ln(\text{g/day})^{-2}$ (a) and $\ln(\text{kg/kg})^{-2}$ (b).

Average daily gain in lifetime showed positive and negative imprinting effects of up to 15 g/day and for feed conversion of more than 0.03 kg/kg. Both plots contain estimates for all animals in the pedigree. Most estimates are concentrated around zero, as expected for the random imprinting effect. In the lower parts of both plots, where precision is low, the differences between breeding values deviate less from zero since estimates are regressed towards zero. This is due to the lack of information, for example, for animals many generations back in the pedigree without direct progeny with records in the data. The spread of estimated imprinting effects for animals with sufficient precision is roughly two thirds of a genetic standard deviation for average daily gain and one third of a genetic standard deviation for feed conversion.

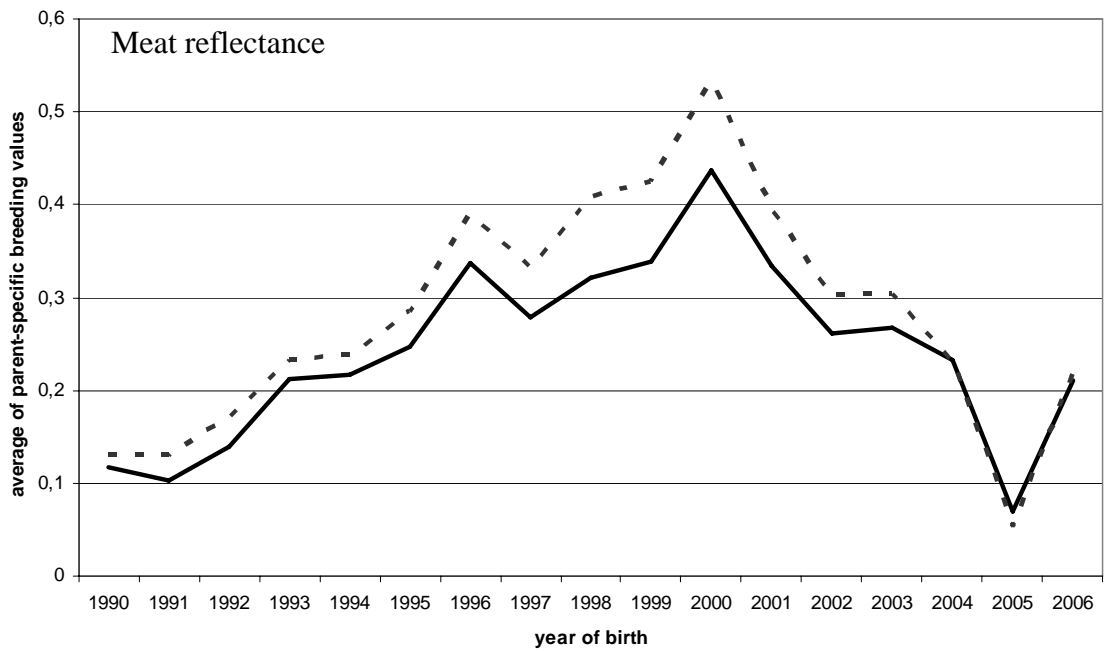
Parent-of-origin-specific genetic trends

Parent-of-origin-specific genetic trends are shown for the percentage of premiums cuts, meat reflectance and average daily gain in lifetime in Figure 3 (a-c).

a)



b)



c)

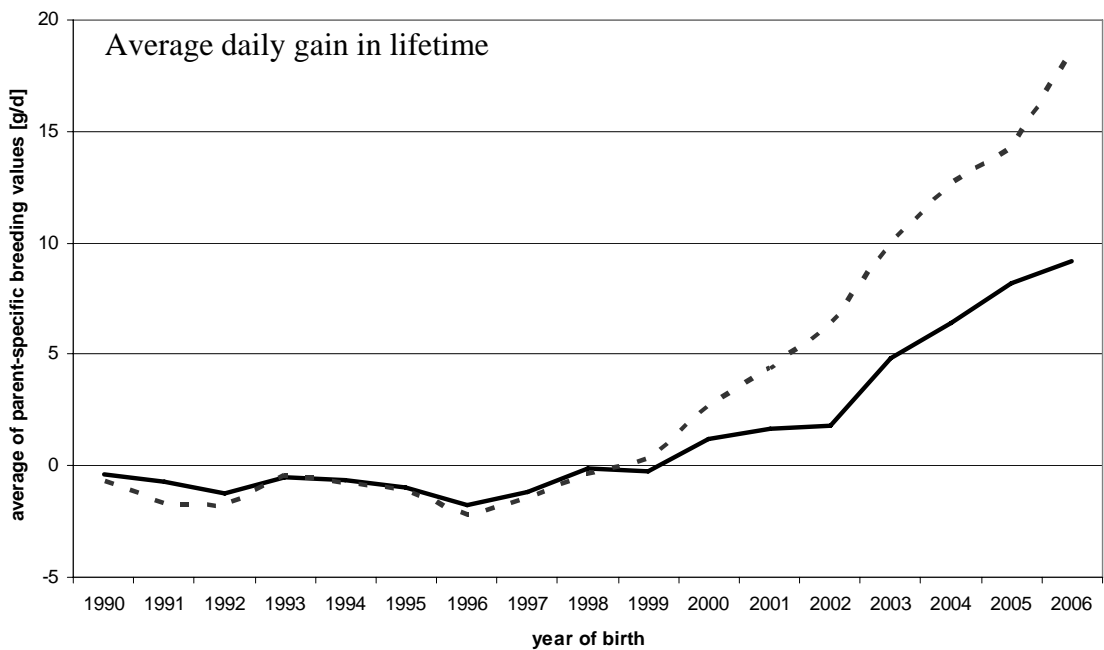


Figure 3: Parent-of-origin-specific genetic trends for percentage of premiums cuts (a) in percent, meat reflectance (b) and average daily gain in lifetime (c) in g/day. Genetic trends as dam (as sire) are represented as black lines (dotted lines).

Genetic trends as sire and as dam for percentage of premiums cuts (Figure 3a) nearly coincide closely until 1993. In 1994, genetic trends split and the trend as dam evolved somewhat steeper than that as sire. In later years (1999 to 2006) the difference between both curves re-

mained more or less constant. The pattern for meat reflectance is quite different (Figure 3b): during the 1990s the gap between both trends increased continuously until a maximum divergence was reached in the year 2000. With the beginning of the new millennium both trends turned negative with a shrinking difference until they coincided for the last 3 years. Meat reflectance was a selection goal during the beginning of the 1990s. Breeders stopped selecting for this trait for a while and started again in the last years.

Positive trends for average daily gain in lifetime (Figure 3c) have been observed since 1999 with virtually no difference between both trends during the years before. Since 1999, the slope of the genetic trend as sire was roughly twice as that as dam and the absolute difference between both trends is still increasing. Apparent separation of estimated genetic trends is influenced by the fact that observations in the data set were not collected earlier than 1997. The overall picture of genetic trends was very similar to that for the trait percentage of premium cuts in Figure 3a. The shape of genetic trends for reflectance is presumably a product of the selection history of this trait, which was a component of the breeding goal at the beginning of the 1990s, but later it was not considered in the breeding goal for some years although recording of the trait was maintained. Since 2000, selection pressure was put again on meat reflectance and obviously forced both genetic trends together, whereas a lack of selection pressure in the years before allowed some divergence of trends. Average daily gain in lifetime is not a component of the total breeding value in Switzerland, whereas daily gain during test is. Therefore, since both growth traits are genetically correlated, there is only an indirect selection on gain during life time. If we further assume that genomic imprinting mainly affects the early growth (for gain during test no significant imprinting variance was found) this may explain the divergent evolution of genetic trends as sire and as dam in this particular case. For the majority of imprinted traits there was, however, nearly no visible divergence of genetic trends.

Correlations between imprinting effects

In order to get insight into the genetic correlations between imprinting effects we computed pair-wise correlations between estimates for imprinting effects of different significant traits, as a convenient surrogate for a comprehensive multi-trait analysis. Table 3 (page: 70) shows the correlation between the significant traits; seven proportion traits (e.g. ham (%)) are not shown, because they were nearly perfectly (0.98-0.99) correlated with their respective weight traits (e.g. ham (kg)). Many of the correlations in Table 3 are low-to-moderate. The highest value of 0.87 was observed between feed conversion and total feed intake during test. Larger correlations between 0.7 and 0.8 were also observed between the percentage of premium cuts and the single cuts shoulder, pork chop and ham, while the correlations with the fatness-related traits back fat thickness at ham, kidney fat and pork belly were of a similar magnitude but of opposite sign. A similar pattern was observed for the correlations between imprinting effects for ham (kg) and other lean cuts (correlations with shoulder 0.47 and with pork chop 0.42) on one hand and fat cuts on the other hand (negative correlations of about 0.55 with back fat at ham, kidney fat and pork belly). The overall picture is, that correlations between imprinting effects for different traits show the same direction as genetic correlations as reported in the literature (e.g. Hermesch *et al.*, 2000; Kadarmideen *et al.*, 2004).

Table 3: Correlations between estimated imprinting effects of different traits

	FC	ADF	TFI	PPC	IFC	MF	BFT	FEET	KFkg	PBkg	SHkg	PCkg	HAkg
ADG	0.13	0.55	0.16	-0.03	-0.14	-0.13	0.16	0.06	0.25	0.21	-0.19	-0.36	-0.20
FC		0.51	0.87	-0.47	0.18	-0.16	0.26	0.05	0.39	0.22	-0.36	-0.43	-0.29
ADF			0.50	-0.55	0.11	-0.16	0.34	0.04	0.44	0.32	-0.38	-0.49	-0.40
TFI				-0.46	0.16	-0.10	0.24	0.09	0.35	0.26	-0.37	-0.48	-0.28
PPC					-0.30	0.01	-0.70	0.27	-0.72	-0.72	0.74	0.77	0.78
IFC						0.14	0.25	-0.25	0.24	0.30	-0.21	-0.16	-0.37
MF							0.01	-0.22	-0.12	0.11	0.01	0.06	-0.11
BFT								-0.31	0.60	0.42	-0.56	-0.52	-0.57
FEET									-0.26	-0.31	0.27	0.10	0.24
KFkg										0.46	-0.58	-0.49	-0.55
PBkg											-0.57	-0.59	-0.56
SHkg												0.58	0.47
PCkg													0.42

ADG: average daily gain in lifetime, FC: feed conversion, ADF: average daily feed intake, TFI: total feed intake during test, PPC: percentage of premiums cuts, IFC: intramuscular fat content of chop, MF: meat reflectance, BFT: back fat thickness at ham, FEET: feet (kg), KFkg: kidney fat (kg), PBkg: pork belly (kg), SHkg: shoulder (kg), PCkg: pork chop (kg), HAkg: ham (kg).

Separation of variance components

Separation of y-chromosomal and the mitochondrial variances from the imprinting variance is possible because their covariance structure differs from other random effects (diagonal matrices: $I_{(YC*YC)}\sigma_{yc}^2$ and $I_{(MI*MI)}\sigma_{mi}^2$).

The case is, however, different for genetic maternal effects variance. Partitioning maternal genetic variance, if existent, from the imprinting variance causes problems. In this case the variance of the observations Y can be written as:

$$Var(Y) = M \left[\sigma_m^2 + 2\frac{1}{2}\sigma_{m,d} + \frac{1}{2}\sigma_d^2 \right] + S \frac{1}{2}\sigma_s^2 + 2C \left[\frac{1}{2}\sigma_{m,s} + \frac{1}{2}\sigma_{sd} \right] + I\sigma_e^2, \quad (16)$$

where M , S and C are matrices derived from the design matrices for genetic maternal effects, genetic effects as sire and genetic effects as dam and I is an identity matrix. It can be seen that the variance component associated with M is a function of the gametic variance as dam σ_d^2 plus the genetic maternal variance σ_m^2 and the covariance between maternal effects and genetic effects as dam $\sigma_{m,d}$. Matrix C has an associated variance component, which is composed by the covariance between genetic maternal effects and genetic effects as sire $\sigma_{m,s}$ and the covariance between genetic effects as sire and as dam σ_{sd} . As a consequence our estimates for the imprinting variances of different traits may be contaminated by fractions of maternal genetic variances. These are, however, considered to be absent or of minor importance for the spectrum of traits analysed; for example, Tholen *et al.* (2005) and Habier *et al.* (2007) estimated very small common environmental litter variances, which are indicators for minor importance of maternal effects. Our estimates are in good agreement with these results (Table 1), since the relative proportion of common litter variance (c^2) was always beyond 10%, with the exception of average daily gain in lifetime with $c^2=0.16$.

Conclusion

There is evidence that parts of the additive genetic variance in pig traits are influenced by imprinting effects. Genetic evaluation could account for such imprinting by estimation of two parental breeding values per animal. It is possible to derive the imprinting variance as the variance of the difference between both breeding values per animal. In a comprehensive data set on Large White pigs, significant genetic imprinting variance was observed for 19 traits. Between 5% and 19% of the total additive genetic variance was controlled by imprinted loci. Therefore, different breeding values for each animal that account for imprinting effects would be helpful especially in cross breeding schemes. In dam lines one should use the breeding values as dam and in sire lines the breeding values as sire to better attain the breeding goal.

Acknowledgement

We thank an anonymous reviewer for helpful and constructive comments, R. Pfuhl for assistance with the terminology of carcass traits and I. Räder for technical assistance.

Literature

- Cameron, ND 1990. Genetic and phenotypic parameters for carcass traits, meat and eating quality traits in pigs. *Livestock Production Science* 26, 119-135.
- Cassady JP, Young LD and Leymaster KA 2002. Heterosis and recombination effects on pig growth and carcass traits. *Journal of Animal Science* 80, 2286-2302.
- De Koning DJ, Rattink AP, Harlizius B, van Arendonk JAM, Brascamp EW and Groenen MAM 2000. Genome-wide scan for body composition in pigs reveals important role of imprinting. *Proceedings of the National Academy of Sciences* 97, 7947-7950.

- De Koning DJ, Rattink AP, Harlizius B, Groenen MAM, Brascamp EW and van Arendonk JAM 2001a. Detection and characterization of quantitative trait loci for growth and reproduction traits in pigs. *Livestock Production Science* 72, 185-198.
- De Koning DJ, Harlizius B, Rattink AP, Groenen MAM, Brascamp EW and van Arendonk JAM 2001b. Detection and characterization of quantitative trait loci for meat quality traits in pigs. *Journal of Animal Science* 79, 2812-2819.
- De Vries AG, Kerr R, Tier B, Long T and Meuwissen THE 1994. Gametic imprinting effects on rate and composition of pig growth. *Theoretical and Applied Genetics*. 88, 1037-1042.
- Enfield FD and Whatley Jr JA 1961. Heritability of Carcass Length, Carcass Back fat Thickness and Loin Lean Area in Swine. *Journal of Animal Science* 20, 631-634.
- Engellandt T and Tier B 2002. Genetic variances due to imprinted genes in cattle. *Journal of Animal Breeding and Genetics* 119 (3), 154-165.
- Essl A and Voith K 2002. Genomic imprinting effects on dairy- and fitness-related traits in cattle. *Journal of Animal Breeding and Genetics* 119 (3), 182-189.
- Gilmour AR, Cullis BR, Welham SJ, Gogel BJ and Thompson R 2004. An efficient computing strategy for prediction in mixed linear models. *Computational Statistics and Data Analysis* 44, 571-586.
- Habier D, Götz K-U and Dempfle L 2007. Estimation of genetic parameters on test stations using purebred and crossbred progeny of sires of the Bavarian Piétrain. *Livestock Science* 107, 142-151.
- Hermesch S, Luxford BG and Graser H-U 2000. Genetic parameters for lean meat yield, meat quality, reproduction and feed efficiency traits for Australian pigs, 2. Genetic relationships between production, carcass and meat quality traits. *Livestock Production Science* 65, 249-259.
- Hill, WG and Keightley, PD 1988. Interaction between molecular and quantitative genetics. pp. 41-55 in *Advances in Animal Breeding*, edited by Korver, S, van der Steen,

- HAM, van Arendonk, JAM, Bakker, H, Brascamp, EW and Dommerholt, J. Proceedings of the World Symposium in Honour of Professor Politiek, RD, Wageningen, The Netherlands, 11-14 September 1988. PUDOC, Wageningen, The Netherlands.
- Hovenier R, Kanis E, van Asseldonk T and Westerink NG 1992. Genetic parameters of pig meat quality traits in a halothane negative population. *Livestock Production Science* 32, 309-321.
- Jeon J-T, Carlborg Ö, Törnsten A, Giuffra E, Amarger V, Chardon P, Andersson-Eklund L, Andersson K, Hansson I, Lundström K and Andersson L 1999. A paternally expressed QTL affecting skeletal and cardiac muscle mass in pigs maps to the *IGF2* locus. *Nature Genetics* 21, 157-158.
- Kadarmideen HN, Schwörer D, Ilahi H, Malek M and Hofer A 2004. Genetics of osteochondral disease and its relationship with meat quality and quantity, growth, and feed conversion traits in pigs. *Journal of Animal Science* 82, 3118-3127.
- Knapp P, Willam A and Sölkner J 1997. Genetic parameters for lean meat content and meat quality traits in different pig breeds. *Livestock Production Science* 52, 69-73.
- Lo LL, McLaren DG, McKeith FK, Fernando RL and Novakofski J 1992. Genetic analyses of growth, real-time ultrasound, carcass, and pork quality traits in Duroc and Landrace pigs: II. Heritabilities and correlations. *Journal of Animal Science* 70, 2387-2396.
- Neugebauer N, Räder I, Schild HJ, Zimmer D and Reinsch N. Evidence for parent-of-origin effects on genetic variability for beef traits. *Journal of Animal Science* 88, 523-532.
- Nezer C, Moreau L, Brouwers B, Coppieters W, Detilleux J, Hanset R, Karim L, Kvasz A, Leroy P and Georges M 1999. An imprinted QTL with major effect on muscle mass and fat deposition maps to the *IGF2* locus in pigs. *Nature Genetics* 21, 155-156.
- Quaas, RL, Pollak, EJ 1980. Mixed model methodology for farm and ranch beef cattle testing programs. *Journal of Animal Science* 51, 1277-1287.

- Reinsch N, Engellandt T, Schild HJ and Kalm E 1999. Lack of evidence for bovine Y-chromosomal variation in beef traits. A Bayesian analysis of Simmental data. *Journal of Animal Breeding and Genetics* 116, 437-445.
- Self SG and Liang K 1987. Asymptotic properties of maximum likelihood estimators and likelihood ratio tests under nonstandard conditions. *Journal of the American Statistical Association* 82, 605-610.
- Stella A, Stalder KJ, Saxton AM and Boettcher PJ 2003. Estimation of variances for genetic effects on litter size in Yorkshire and Landrace swine. *Journal of Animal Science* 81, 2171-2178.
- Tholen E, Jüngst H, Schulze-Langenhorst C and Schellander K 2005. Genetic foundation of meat quality traits of station tested slaughter pigs in North Rhine-Westphalia (Germany). *Archiv für Tierzucht (Special Issue)* 48, 123-130.
- Van Laere A-S, Nguyen M, Braunschweig M, Nezer C, Colette C, Moreau L, Archibald AL, Haley CS, Buys N, Tally M, Andersson G, Georges M and Andersson L 2003. A regulatory mutation in *IGF2* causes a major QTL effect on muscle growth in the pig. *Nature* 425, 823-836.
- Welham SJ, Cullis BR, Gogel BJ, Gilmour AR and Thompson R 2004. Prediction in linear mixed models. *Australian and New Zealand Journal of Statistics* 46, 325-347.

Chapter 3

On-station heritabilities and parent-of-origin effects for beef traits in
German Simmental

Submitted

On-station heritabilities and parent-of-origin effects for beef traits in German Simmental

N. Neugebauer¹, J. Kögel², U. Geuder², S. Kämmerer³, N. Reinsch¹

1) Forschungsinstitut für die Biologie landwirtschaftlicher Nutztiere, FBN, Forschungsbereich Genetik und Biometrie, Wilhelm-Stahl-Allee 2, 18196 Dummerstorf, Germany

2) Bayerische Landesanstalt für Landwirtschaft, Institut für Tierzucht, Prof.-Dürrwaechter-Platz 1, 85586 Poing-Grub, Germany

3) Bayerische Landesanstalt für Landwirtschaft, Abteilung Qualitätssicherung und Untersuchungswesen, Prof.-Zorn-Str. 20c, 85586 Poing-Grub, Germany

Abstract

In order to assess heritabilities as well as the relative importance of genomic imprinting for genetic variation of traits for growth, carcass composition, meat quality and fatty acid profiles of beef a data set with observations from 3,368 Simmental fattening bulls was analysed. These animals were the sons of 632 sires, which were progeny-tested in the years 2000 to 2007 on two test stations in Bavaria. A set of 46 different traits was investigated the number of observations per trait was between 2,107 and 3,368. Estimated heritabilities of growth and carcass quality traits were around 50 % and most of the meat quality traits showed heritabilities between 30 % and 40 %. Heritability estimates for fatty acid profiles were considerably uniform at about 40 %. Genomic imprinting significantly contributed to the genetic variance of four traits (muscularity score, loss, E-U-R-O-P-beefiness class and the polyunsaturated fatty acids) with corresponding imprinting variances that accounted for 5, 5.5, 7 and 11 per cent of the total additive-genetic variation.

Introduction

Genetic evaluation for beef traits in German Simmental mainly relies on extensive field data from slaughterhouses. Until 2007 additional progeny testing was carried out on two Bavarian test stations (Westerschondorf and Schwarzenau). Potential benefits of a station test are higher heritabilities and also higher genetic gain due to a better standardised environment, provided the latter matches usual production conditions in order to avoid genotype-environment interactions. A further advantage is a broader spectrum of traits, especially such, which are difficult to record under field conditions.

In a dataset with results from the two mentioned test stations from the Bavarian State Research Center of Agriculture, where young Simmental bulls were fattened, the genetic analysis is interesting because it provides a rich set of traits especially related to meat and carcass quality. Traits of special interest are the contents of several fatty acids, meat colour and some more common traits like EUROP-beefiness score, net daily gain, muscularity score and fatness score. Estimates of genetic parameters for the content of fatty acids in beef are rare in the literature. Malau-Aduliand, Siebert, Deland, Bottema, Pitchford & Edriss (1998 and 2000) analysed over 300 crossbreds of Hereford dams with Angus, Belgian Blue, Hereford, Jersey, Limousin, South Devon or Wagyu sires. The data were recorded after weaning and 90 days in the feedlot, from the slaughtered animals adipose tissue was analysed for fatty acid profiles. They reported heritabilities for the sum of saturated fatty acids (SFA) between 15% (weaning) and 23% (slaughter), for the sum of monounsaturated fatty acids (MUFA) from 16% (weaning) to 57% (slaughter) and for the polyunsaturated fatty acids (PUFA) they found heritabilities between 0% (weaning) and 15% (slaughter). Pitchford, Deland, Siebert, Malau-Aduliand, & Bottema (2002) analysed data from the same project, but with more animals and calculated heritabilities for single fatty acids between 14% (stearic acid) and 21% (palmitic acid), for MUFA they found 27%.

In a recent study (Neugebauer, Räder, Schild, Zimmer & Reinsch, 2009), genomic imprinting was found to contribute significantly to the additive genetic variation of mainly carcass-composition traits in Simmental. These results were found in a comprehensive analysis of slaughterhouse data, which were recorded by a video-image-analysis device, from more than 65.000 Simmental young bulls and can be viewed as a confirmation of an earlier study of German Gelbvieh data from a conducted field test (Engellandt & Tier, 2002). Under genomic imprinting genes from the male (female) parent are not expressed at all or expressed at a lower level (partial imprinting), while the copy from the female (male) parent is fully expressed. Genomic imprinting is caused by epigenetic modification and will be newly established in each generation during gametogenesis depending on the sex of the parent. The studies from Engellandt and Tier (2002) and Neugebauer et al. (2009) differ in their statistical approach: while the first analysis only allowed for extra paternal genetic variation, the Simmental-study accounted for both paternal and maternal imprinting simultaneously.

The aim in analysing the aforementioned Simmental station-data was thus two-fold: First all traits were analysed assuming standard (*Mendelian*) additive inheritance and genetic parameters were derived. Second each trait was again examined by applying a statistical model allowing for genomic imprinting, both maternal and paternal simultaneously, followed by a statistical test for a significant imprinting variance component. Therefore this article contributes to the quantitative genetics of traits related to meat and carcass quality, growth and, especially, the fatty acid profile in beef.

Material and Methods

Animals and traits

In two test stations (Westerschondorf and Schwarzenau) German Simmental bulls were fattened for the purpose of progeny testing. Animals were bought as calves on auctions at differ-

ent places in Bavaria. After a rearing and adaptation period (until about 150 days of age) young bulls were fed semi ad libitum maize silage, 2 kg of protein concentrate (60% soybean meal, 5% minerals and 35% of a mixture of barley and wheat) and 0.5 kg (at the beginning of fattening) to 2.5 kg (at the end of fattening) coarsely cracked maize meal. This type of ration is standard in specialised fattening farms in southern Germany. The fattening period ended at an age of 450 days, with an average final weight of about 600 kg (Kögel, 1999). Feed, final weight, group size (8) and floor design (slatted floor) were selected according to common production conditions in the field in order to minimize possible genotype-environment interactions.

Between 2000 and 2007 3,368 Simmental fattening bulls were slaughtered and up to 46 traits per animal were recorded. Table 1 to Table 3 (page: 86, 87, 89) provide an overview of the number of observations per trait (range from 2,107 to 3,368), the raw means and standard deviations. Six traits (the first ones in Table 1) are related to growth and twelve to carcass quality (all others in Table 1). Meat quality is covered by twelve traits (Table 2) and the fatty acid pattern in beef (Table 3) by the content of eleven single fatty acids and three combined traits (SFA, MUFA, PUFA). Carcass traits were recorded according to DLG (Deutsche Landwirtschafts-Gesellschaft, German agricultural association) guidelines (Scheper and Scholz, 1985).

Meat quality traits and fatty acid profiles were analysed in *M. longissimus dorsi*. Meat colour, meat reflectance, pH 45 min post mortem and 24 h post mortem were measured at the 9th rib of the left carcass side. Meat colour and reflectance were determined at ten positions using a Minolta Spectrophotometer 508 i, (Minolta Camera, Milton Keynes, UK). For intramuscular fat content and fatty acid analysis a sample of the 9th rib was taken 24 h after slaughter and analysed using NIR spectroscopy. The FOSS 6500 NIR spectrometer used a calibration for IMF content based on data that were measured according to § 64 LFGB (2006). Calibrations for fatty acids had been developed using capillary gas chromatography as reference method.

For this purpose lipids had been extracted from muscle with chloroform/methanol (Folch, Lees & Sloane Stanley, 1957). Methyl esters had been prepared using TMSH (DGF, 2007). Fatty acid methyl esters had been separated on an HP 6890 Network GC system (Agilent, Germany) which had been equipped with a flame ionisation detector and a capillary column (Supelco Fused Silica SP 2380, 100 m, Sigma-Aldrich, Germany). Helium at 3,4 bar had been used as the carrier gas, resulting in a flow of 1,95 ml/min at 60°C. Temperature conditions had been as follows: the initial oven temperature had been 60°C, held for 1 min, subsequently increased to 120°C at a rate of 8°C min⁻¹ and then to 242°C at 1.5°C min⁻¹. The injector had been set at 250°C, and the detector temperature had been 260°C. Fatty acids had been identified and quantified using appropriate standards (Sigma-Aldrich, Germany; Larodan, Sweden). For the analysis of the loss during storage a steak from the 10th rib was stored for 13 days at 4°C. Cooking loss was determined after grilling the steak until the core temperature reached 65°C. Shear force measurements was carried out with an Instron 4301 material testing machine (Instron, Germany).

The pedigree file was extracted from a comprehensive Simmental pedigree and consisted of all parents of the slaughtered animals and their ancestors (53,403 animals). As a measure of pedigree completeness the proportion of known great-grandparents of the parent-generation was calculated and resulted in an average value of 51.3%.

Effects in the model

Genetics analysis of all traits was based on the following linear model, which was adopted from the standard genetic evaluation for Simmental on-station progeny test results

$$y_{ijklmnop} = SD_i + GT_j + LA_k + bx + as_l + ad_m + m_n + yc_o + e_{ijklmnop} \quad (1)$$

where y is the vector of observations, SD_i is the fixed effect of slaughter day, GT_j is the fixed effect of birth types (single-, twin- and multiple births), LA_k is the fixed effect of lacta-

tion number of the dam, subdivided into first, second and all later lactation, b is a linear regression on slaughter age, as_l is the random additive genetic effect as sire, ad_m the random additive genetic effect as dam, m_n is the random mitochondrial effect and yc_o the random y-chromosomal effect, $e_{ijklmnop}$ is the random residual. The assumption for the estimation of heritabilities was, that as_l (random additive genetic effect as sire) and ad_m (random additive genetic effect as dam) were equal with a correlation of one (see below). In this case all variances of the genetic effects are equal

$$\text{Var} \begin{bmatrix} a_s \\ a_d \end{bmatrix} = A \otimes S, \quad \text{where } S = \begin{bmatrix} \sigma_g^2 & \sigma_g^2 \\ \sigma_g^2 & \sigma_g^2 \end{bmatrix} \quad (2)$$

where a_s and a_d are the vectors, which included the elements of the genetic effects as_l and ad_m . This model represent a non-imprinting model (*Mendelian* model), the corresponding imprinting model will be presented in the following paragraph.

Maternal inheritance was considered by tracing back in the pedigree until the very first female founder was identified. This female got a number and was assigned to each observation with the same female founder as the corresponding mitochondrial effect. The assumption was that the variance of mitochondrial effects was $I\sigma_{mi}^2$ where I is an identity matrix with dimension MI , and MI is the number of distinct founder mitochondria in the pedigree. The same system was used for the y-chromosomal inheritance and by tracing back the paternal inheritance. The variance of the y-chromosomal effects was $I\sigma_{yc}^2$, where I is an identity matrix with dimension YC , and YC is the number of distinct founder y-chromosomes in the pedigree.

The ASReml program was used for variance component estimation (Gilmour, Cullis, Welham, Gogel, & Thompson, 2004); approximative standard errors for heritabilities and other functions of variance components were derived by applying the delta method.

Principle of imprinting analysis

The imprinting model consisted of fixed effects as described before and of two genetic random effects per animal: one for the genetic effect as sire, i.e. half of the breeding value as sire and one for the genetic effect as dam, i.e. half of the breeding value as dam (Reinsch & Guiard, in preparation)

$$Y = X\beta + Z_s a_s + Z_d a_d + e \quad (3)$$

where Y is the vector of observations, X is the design matrix for fixed effects with the corresponding vector β , Z_s and Z_d are the design matrices for random genetic effects with the vectors a_s and a_d for half of the breeding values as sire and as dam and e is the vector of random residuals. The covariance of the random effects includes the additive genetic relationship matrix A and the corresponding gametic variances correspond to the breeding value as sire and as dam

$$\text{Var} \begin{bmatrix} a_s \\ a_d \\ e \end{bmatrix} = \begin{bmatrix} \frac{1}{2} A\sigma_s^2 & \frac{1}{2} A\sigma_{sd} & 0 \\ \frac{1}{2} A\sigma_{sd} & \frac{1}{2} A\sigma_d^2 & 0 \\ 0 & 0 & W\sigma_R^2 \end{bmatrix} \quad (4)$$

The structure of the mixed model equations is

$$\begin{bmatrix} X'W^{-1}X & X'W^{-1}Z_s & X'W^{-1}Z_d \\ Z_s'W^{-1}X & Z_s'W^{-1}Z_s + A^{-1}\alpha_1 & Z_s'W^{-1}Z_d + A^{-1}\alpha_2 \\ Z_d'W^{-1}X & Z_d'W^{-1}Z_s + A^{-1}\alpha_2 & Z_d'W^{-1}Z_d + A^{-1}\alpha_3 \end{bmatrix} \begin{bmatrix} \beta \\ a_s \\ a_d \end{bmatrix} = \begin{bmatrix} X'W^{-1}y \\ Z_s'W^{-1}y \\ Z_d'W^{-1}y \end{bmatrix} \quad (5)$$

The assumption for the estimation of imprinting analysis was, that as_l (random additive genetic effect as sire) and ad_m (random additive genetic effect as dam) were not equal with different variances and a non-perfect correlation

$$\text{Var} \begin{bmatrix} a_s \\ a_d \end{bmatrix} = A \otimes S, \quad \text{where } S = \begin{bmatrix} \sigma_s^2 & \sigma_{sd} \\ \sigma_{sd} & \sigma_d^2 \end{bmatrix} \quad (6)$$

The variance components for the genetic effects are $\frac{1}{2}\sigma_s^2$ for the paternal one, $\frac{1}{2}\sigma_d^2$ for the maternal one and $\frac{1}{2}\sigma_{sd}$ for their covariance expressed in terms of gametic variances. The weighting factors W (diagonal matrix) with elements

$$w_i = \left[\frac{\frac{1}{2}\sigma_s^2(1-F_{si}) + \frac{1}{2}\sigma_d^2(1-F_{di}) + \sigma_e^2}{\frac{1}{2}\sigma_s^2 + \frac{1}{2}\sigma_d^2 + \sigma_e^2} \right]^{-1} \quad (7)$$

account appropriately for potential inbreeding of parents, by regulating the error variance of each observation due to variation in the Mendelian sampling term according to the inbreeding coefficients of both parents (special kind of reduced model, Quaas & Pollak, 1980).

The estimated imprinting variance can be calculated as the sum of both corresponding genetic variances per animal minus twice the covariance

$$\sigma_i^2 = \sigma_s^2 + \sigma_d^2 - 2\sigma_{sd}. \quad (8)$$

The imprinting variance σ_i^2 is a proportion of the total additive genetic variance ($\sigma_a^2 = \sigma_s^2 + \sigma_d^2$), for each trait the imprinting variance can be divided into a paternal contribution $\sigma_s^2 - \sigma_{sd}$ and a maternal contribution $\sigma_d^2 - \sigma_{sd}$. In the imprinting model one part of the total additive genetic variance will be formed by the imprinting variance, the other part is the Mendelian variance $\sigma_M^2 = \sigma_a^2 - \sigma_i^2 = 2\sigma_{sd}$ which describes the “unimprinted” component of the additive genetic variance (not to be confused with the *Mendelian* sampling variance).

Testing hypotheses on the imprinting variance

The null hypothesis assumes no imprinting ($\sigma_i^2 = 0$) in the data set and is presented by the *Mendelian* model, where all variances of the genetic variance are equal (formula no. (2)), in this case the matrix S is not positive definite. For the alternative hypothesis the variances of both genetic effects are different (formula no. (5)), i.e. the genetic covariance matrix is positive definite.

For each trait in the data set variance components for the Mendelian model and the imprinting model were estimated. A restricted likelihood ratio test (RLRT) with two degrees of freedom was used to test the hypothesis of imprinting against the absence of imprinting. The χ^2 distribution of $RLRT$ is $RLRT \sim p\chi_1^2 + (1-p)\chi_2^2$, so in this case the correct χ^2 distribution is between the distribution of degrees of freedom (df)=1 and the distribution of df=2. In principle, it is possible to estimate p with a parametric bootstrap method to find the correct degrees of freedom of the χ^2 distribution (Crainiceanu & Ruppert, 2004). In our application this approach is, however, prohibitively demanding in terms of computing time. Therefore we decided to treat $p = 0$ and used a χ^2 distribution with DF=2, resulting in a conservative test.

Results and Discussion

Estimated heritabilities together with their standard errors from the Mendelian model without genomic imprinting are listed in Table 1 to Table 3 (page: 86, 87, 89). The estimated heritabilities of traits related to growth and carcass quality ranged around 50% except average daily gain during the last test period with 18% (Table 1). Engellandt, Reinsch, Schild, & Kalm (1999b) reported a heritability of 12% of net gain for German Simmental and again the same estimate was found in a unconducted field test of German Gelbvieh finishing bulls (Engellandt, Reinsch, Schild, & Kalm, 1999a), for the conducted field test the same authors reported a heritability of 34% of net gain. Neugebauer et al. (2009) found heritability for net BW gain of 28% in German Simmental. The carcass traits expressed as proportion show relatively high estimated heritabilities with 48-74%, also the area of *M. longissimus dorsi* with 64% (Table 1). Estimates of heritabilities for Simmental fattening bulls from Neugebauer et al. (2009) are on the same level as the results from this analysis for beefiness score (31%) and meat percentage (27%), comparable results for pistola cut/slaughter weight (17%), killing out percentage (22%) and carcass weight (26%) are on smaller levels but the fat score showed a

Table 1: Overview over traits related to growth and carcass quality: number of observations (n), raw means (\bar{x}) and standard deviations (sd, in brackets), estimated genetic standard deviations ($\hat{\sigma}_g$) and heritabilities (\hat{h}^2) from a Mendelian model together with their standard errors (se, in brackets).

trait (unit)	n	raw mean (sd)	genetic std dev	heritability (se)
Initial weight (kg)	3368	87.55 (11.82)	5.29	0.58 (0.04)
Carcass weight (kg)	3368	345.14 (31.35)	13.21	0.51 (0.04)
Net BW gain (g/d)	3356	765.38 (68.49)	29.02	0.50 (0.04)
Average daily gain in lifetime (g/d)	3356	1292.52 (111.16)	49.57	0.53 (0.04)
Average daily gain during test (g/d)	3368	1406.58 (128.89)	59.27	0.57 (0.04)
Average daily gain during the last test period (g/d)	3368	1315.64 (312.64)	86.25	0.18 (0.03)
Kidney fat (%)	3368	2.85 (0.75)	0.22	0.49 (0.02)
Slaughterhouse waste (%)	3367	5.91 (0.99)	0.46	0.56 (0.04)
Pistola cut/slaughter weight (%)	3368	42.79 (1.23)	0.49	0.48 (0.04)
Killing out percentage (%)	3368	57.69 (1.4)	0.74	0.74 (0.04)
Area of <i>M. longissimus dorsi</i> (cm ²)	3368	49.47 (6.58)	3.18	0.64 (0.04)
Lean meat proportion (%)	3344	67.85 (1.78)	0.74	0.42 (0.04)
Bone proportion (%)	3366	14.48 (0.51)	0.19	0.32 (0.03)
Tendon fat proportion (%)	3344	17.92 (2.15)	0.79	0.32 (0.03)
Meat percentage (%)	3344	82.77 (0.99)	0.36	0.32 (0.03)
E-U-R-O-P-beefiness class (15 classes) ^a	3366	3.25 (0.45)	0.18	0.37 (0.03)
Fat score (1-15 scores) ^a	3366	2.91 (0.52)	0.11	0.09 (0.02)
Muscularity score (1-15 scores) ^b	3366	5.28 (0.93)	0.34	0.32 (0.03)

a) EU carcass grading system

b) Scoring of life animals

higher heritability as in this analysis. Other possible comparisons are the results from Reinsch et al. (1999) because they analysed Simmental young bulls from beef progeny field tests. The design of the test is different to our data set (field data from slaughterhouses) but the animals

are from the same population. Their threshold-model heritability for E-U-R-O-P (EU carcass grading system, with five classes) was 20%. Our result for E-U-R-O-P (15 classes) heritability was 37% (Table 1, page: 86). The smaller heritabilities of the other analysis might have been caused a greater residual variation under production conditions and a coarser measurement of the trait (five versus 15 classes). Crews, Pollak, Weaver, Quaas & Lipsey (2003) estimated a 48% heritability for carcass weight in American Simmental bulls. Our result of carcass weight is on the same level (51%). Engellandt et al. (1999b) reported a heritability of 61% of kidney fat for German Gelbvieh from a conducted field test. Our result of kidney fat is somewhat lower (49%).

Table 2: Overview over traits related to meat quality: number of observations (n), raw means (\bar{x}) and standard deviations (sd , in brackets), estimated genetic standard deviations ($\hat{\sigma}_g$) and heritabilities (\hat{h}^2) from a Mendelian model together with their standard errors (se , in brackets).

trait (unit)	n	raw mean (sd)	genetic std dev	heritability (se)
Loss (%) ^a	3368	6.1 (0.83)	0.26	0.45 (0.04)
Storage loss (%)	3291	2.9 (1.44)	0.00	0.00 (0.02)
Cooking loss (%)	3361	28.68 (3.15)	0.67	0.12 (0.03)
pH 45 minutes <i>post mortem</i>	3355	6.92 (0.15)	0.00	0.00 (0.00)
pH 24 hours <i>post mortem</i>	3347	5.56 (0.13)	0.02	0.10 (0.03)
pH 12 days <i>post mortem</i>	3363	5.57 (0.13)	0.02	0.07 (0.03)
Colour L	2180	37.4 (2.2)	0.77	0.38 (0.04)
Colour a	2180	12.5 (1.64)	0.42	0.18 (0.03)
Colour b	2180	3.83 (1.46)	0.40	0.19 (0.04)
Intramuscular fat	3335	2.35 (0.88)	0.37	0.50 (0.04)
Average shear force (N)	3352	36.7 (11.41)	3.48	0.34 (0.03)
Average energy of shear force (mJ)	3368	0.33 (0.11)	0.03	0.36 (0.04)

a) Loss=warm carcass weight – cold carcass weight

Meat quality traits show high heritabilities: e.g. loss (45%) and intramuscular fat (50%) (Table 2, page: 87). Most of these traits have heritabilities between 30 and 40%. In the literature few comparable heritability estimates exist for our traits because definition of cuts varies considerably across countries and a lot of the analysed traits are very rare. The best possible comparison is with the results of Kögel, Lindner & Schuster (2006), who analysed an earlier version of the data set, which was recorded under a different design. They reported heritabilities for colour L, colour a and colour b (55%, 33%, 29%), storage loss and cooking loss (61% and 16%), intramuscular fat (63%) and shear force (45%) and shear force energy (45%). For all traits the results were higher as our estimates with differences between 0.04 and 0.17. For storage loss the difference is extraordinarily large with 61%.

The heritabilities of fatty acid profiles are considerably uniform around 40%, the ratio of fatty acids 18:0 to 18:1 is somewhat higher (49%), but the ratios of the fatty acids 14:0 to 14:1 and 16:0 to 16:1 have lower estimates with 21-22% (Table 3, page: 89). Animal model heritabilities of fatty acid composition in crossbred cattle (Hereford and other breeds) were estimated by Malau-Aduliand et al. (1998 and 2000) and by Pitchford et al. (2002). The analysed traits were proportions of single fatty acids relative to the total of all fatty acids (%; Malau-Aduliand et al.; 1998 and Pitchford et al.; 2002) and, in one case (Malau-Aduliand et al.; 2000), relative to the fatty acids extracted from the phospholipid fraction of beef. The heritability for PUFA was estimated at 15%, for SFA the results ranged from 22-27%, for MUFA from 14-57%, C 16:0 from 13-21%, C 18:0 from 12-14%, C 18:1n-9 from 5-17%. Heritability estimates of C 14:0, C 16:1n-9 and IMF were calculated with 18%, 16% and 18% (Pitchford et al. (2002). A comparison with our results suffers from the difference in trait definition [relative proportions versus absolute fatty acid content (mg/100g)], but the pattern of high and low estimates is nevertheless similar.

Table 3: Overview over traits related to fatty acid profiles and the ratios of fatty acids: number of observations (n), raw means (\bar{x}) and standard deviations (sd , in brackets), estimated genetic standard deviations ($\hat{\sigma}_g$) and heritabilities (\hat{h}^2) from a Mendelian model together with their standard errors (se , in brackets).

trait (unit)	n	raw mean (sd)	genetic std dev	heritability (se)
C 14:0 (mg/100g meat)	2110	49.74 (23.64)	8.88	0.38 (0.04)
C 14:1 cis -9 (mg/100g meat)	2107	8.64 (5.37)	1.87	0.35 (0.04)
C 16:0 (mg/100g meat)	2110	524.71 (216.23)	85.74	0.43 (0.04)
C 16:1 cis -9 (mg/100g meat)	2110	53.84 (25.92)	9.49	0.37 (0.04)
C 18:0 (mg/100g meat)	2110	380.1 (126.5)	47.72	0.40 (0.04)
C 18:1 $trans$ (mg/100g meat)	2110	43.19 (13.55)	5.32	0.41 (0.04)
C 18:1 cis -9 (mg/100g meat)	2110	732.36 (320.7)	128.67	0.44 (0.05)
C 18:1 cis -11 (mg/100g meat)	2110	37.32 (14.28)	5.74	0.45 (0.05)
C 18:3 n -3 (mg/100g meat)	2110	7.3 (1.09)	0.32	0.29 (0.04)
C 22:4 (mg/100g meat)	2110	9.15 (0.94)	0.36	0.41 (0.05)
CLA c9 t11 (mg/100g meat)	2110	9.1 (3.29)	1.12	0.33 (0.04)
SFA (mg/100g meat) ^a	2110	987.54 (375.68)	144.36	0.40 (0.04)
MUFA (mg/100g meat) ^b	2110	876.25 (383.41)	152.71	0.43 (0.05)
PUFA (mg/100g meat) ^c	2110	174.91 (11.07)	3.66	0.34 (0.04)
Ratio of C14:1 to C14:0	2107	0.16 (0.03)	0.009	0.22 (0.04)
Ratio of C16:1 to C16:0	2110	0.1 (0.01)	0.003	0.21 (0.04)
Ratio of C18:1 to C18:0	2110	1.88 (0.3)	0.131	0.49 (0.05)

a) The sum of saturated fatty acids (SFA) was calculated as: C10:0 + C12:0 + C14:0 + C15:0 + C16:0 + C17:0 + C18:0 + C20:0 + C22:0 + C24:0.

b) The sum of monounsaturated fatty acids (MUFA) was calculated as: C14:1 + C16:1 + C18:1 $trans$ + C18:1 cis 9 + C18:1 cis 11 + C20:1 + C22:1 + C24:1.

c) The sum of polyunsaturated fatty acids (PUFA) is the sum of n-6 and n-3 FA.

The sample size of the data is clearly too small to allow a reliable estimation of genetic correlation (Koots and Gibson; 1994). Therefore, as a convenient surrogate for a comprehensive multi-trait analysis pair-wise correlations between estimated breeding values (from the Men-

delian model) of different traits were computed to analyse especially the genetic correlation between fatty acid related traits and other slaughter traits, since there are almost no results in the literature. Particularly the correlation between the sum of saturated fatty acids (SFA) and intramuscular fat of *longissimus* muscle showed a close connection. The high correlation of 0.74 points out that both the *de novo* fatty acid synthesis and the triacylglycerol synthesis are in connection with the deposition of intramuscular fat (Nürnberg, 2009).

For four traits (shown in Table 4, page: 92) the imprinting variances were significantly different from zero, with error probabilities ranging from 0% to 4.97%. Imprinting variances σ_i^2 of these traits were estimated to account between 5% and 11% of the respective total additive genetic variance (Table 4). This ratio was highest for the sum of polyunsaturated fatty acids (10.95%). The authors are unaware of any comparable estimate in the literature. The other three traits with a significant imprinting variance are related to carcass composition, which is in good agreement both with the Gelbvieh study of Engellandt and Tier (2002) and a recent analysis of video-recorded slaughterhouse-data from Simmental young bulls showing significant imprinting variance for ten traits, among them E-U-R-O-P-beefiness score with a relative imprinting variance of 10.75% (Neugebauer et al., 2009). The estimated imprinting variances are a result of the incomplete genetic correlation between both genetic effects (Table 4) and the differences between their variances. For the significant traits these correlations between the breeding values as sire and as dam ranged from 0.00 for PUFA, 0.74 for loss, 0.85 for E-U-R-O-P to 0.98 for muscularity score. Their standard errors, 0.54 for PUFA and 0.92 for loss, are very high. This indicates that the imprinting variances are not based on the incomplete genetic correlation only, but also on the differences between the variances.

The imprinting-model heritabilities for the four significant traits are presented in Table 4, together with their approximative standard errors. Estimates ranged from 20% to 34% and were lower than their corresponding Mendelian model counterparts for loss, E-U-R-O-P -grade and muscularity score with corresponding reductions of 35%, 27% and 37.5% of the Mendelian

estimates. The heritability estimate for PUFA from the imprinting model was however not reduced at all. Engellandt and Tier (2002) and Neugebauer et al., 2009 found similar reductions of heritabilities between the Mendelian and the imprinting model between 1% and 4% (Simmental data). This is in good agreement with the results of this study.

Variation of y-chromosomal inheritance turned out to be almost completely absent for all traits, which is in good accordance with Reinsch et al. (1999), Engellandt and Tier (2002) and Neugebauer et al. (2009). Estimates for the relative proportion of the mitochondrial variance were higher in some cases, but were not significant. This is in good agreement with Neugebauer et al. (2009).

Conclusion

Estimated heritabilities of growth and carcass quality traits ranged around 50%. Most of meat quality traits showed heritabilities between 30 and 40% and the heritabilities of fatty acid composition were considerable uniform around 40%. Significant genetic imprinting variance was observed for four traits (muscularity score, loss, E-U-R-O-P-beefiness class and PUFA), where between 5% and 11% of the total additive genetic variance was controlled by imprinted loci.

Acknowledgement

We thank K. Nürnberg for her help and ideas with regard to fatty acid traits and I. Räder in preparing the data set. The research was partially funded by the Fugato-plus QuaLIPID project of the German Federal Ministry of Education and Research.

Table 4: The four traits with significant imprinting variances, with their genetic standard deviation, relative imprinting variance, heritability (imprinting model), genetic correlation between breeding values as sire and as dam. The standard errors (se) for estimates in brackets.

trait	genetic standard deviation	relative imprinting variance ¹ (%)	heritability	correlation	RLRT ²	p-value ³
Loss	0.313	5.50 (0.90)	0.29 (0.05)	0.74 (0.92)	10.42	0.005
E-U-R-O-P-beefiness class	0.681	6.99 (0.00)	0.27 (0.01)	0.85 (0.00)	14.56	0.000
Muscularity score	0.391	5.03 (0.00)	0.20 (0.00)	0.98 (0.00)	10.12	0.006
PUFA	5.432	10.95 (1.06)	0.34 (0.08)	0.0 (0.54)	5.99	0.050

- 1) The imprinting variance is expressed relative to the total additive genetic variance
- 2) $2(\text{LogL}(\text{imprinting model}) - \text{LogL}(\text{Mendelian model}))$
- 3) Error probabilities

Literature

- Crainiceanu, C. M., & Ruppert, D. (2004). Likelihood ratio tests in linear mixed models with one variance component. *J. R. Statist. Soc. B* 66(1), 165-185.
- Crews Jr., D. H., Pollak, E. J., Weaver, R. L., Quaas, R. L., & Lipsey, R. J. (2003). Genetic parameters for carcass traits and their live animal indicators in Simmental cattle. *J. Anim. Sci.* 81, 1427-1433.
- DGF: Deutsche Gesellschaft für Fettwissenschaften e. V. (Hrsg.): Methode C-VI 11e (98) Fettsäuremethylester (TMSH-Methode). DGF-Einheitmethoden. Deutsche Einheitmethoden zur Untersuchung von Fetten, Fettprodukten, Tensiden und verwandten Stoffen. Wiss. Verlagsgesellschaft mbH, Stuttgart, 2. Auflage einschl. 12. Aktualisierungslieferung, 2007.
- Engelland, T., & Tier, B. (2002). Genetic variances due to imprinted genes in cattle. *J. Anim. Breed. Genet.* 119 (3), 154-165.
- Engelland, T., Reinsch, N., Schild, H. J., & Kalm, E. (1999a). Progeny test for beef traits in German Simmental – organization, genetic parameters and breeding values. *J. Anim. Breed. Genet.* 116, 47-60.
- Engelland, T., Reinsch, N., Schild, H. J., & Kalm, E. (1999b). Genetic parameters from two different field testing schemes for beef traits of German Gelbvieh finishing bulls. *Livest. Prod. Sci.* 60, 219-228.
- Falconer, D. S., & Mackay, T. F. C. (1996). *Introduction to Quantitative Genetics*. Ed. 4. Longman Group, Essex, UK, 132-133 and 315-316.
- Folch, J., Lees, M. Sloane Stanley, G. H., 1957: A simple method for the isolation and purification of the total lipids from animal tissues. *J. Biol. Chem.* 226, 497-509.

- Gilmour, A. R., Cullis, B. R., Welham, S. J., Gogel, B. J., & Thompson, R. (2004). An efficient computing strategy for prediction in mixed linear models. *Computational Statistics and Data Analysis* 44, 571-586.
- Kögel, J. (1999). Zuchtwertschätzung bei der Nachkommenprüfung in Station beim Rind. *Schule und Beratung (SUB)*, 04-05/99, IV-18.
- Kögel, J., Lindner, P., & Schuster, M. (2006). Rindfleischqualität – Genetische Parameter und Zuchtwertschätzung. *Schule und Beratung (SUB)* 6, (www.stmlf.bybn.de/sub/).
- Koots, K. R., & Gibson, J. P. (1994). How precise are genetic correlation estimates? In *Proceedings of the 5th World Congress on Genetics Applied to Livest. Prod.* 18, 353-356.
- LFGB: Bestimmung des Gesamtfettgehaltes in Fleisch und Fleischerzeugnissen L 06.00-6. § 64 Amtliche Sammlung von Untersuchungsverfahren. Lebensmittel- und Futtermittelgesetzbuch. Neugefasst durch Bek. v. 26.04.2006 (BGBl. I S. 945), zuletzt geändert durch Art. 12 des Gesetzes v. 26.02.2008 (BGBl. I S. 215).
- Malau-Aduliand, A. E. O., Edriss, M. A., Siebert, B. D., Bottema, C. D. K., & Pitchford, W. S. (2000). Breed differences and genetic parameters for melting point, marbling score and fatty acid composition of lot-fed cattle. *J. Anim. Physiol. a. Anim. Nutr.* 83, 95–105.
- Malau-Aduliand, A. E. O., Siebert, B. D., Deland, M. P. B., Bottema, C. D. K., & Pitchford, W. S. (1998). Heritabilities of triacylglycerol fatty acids from the adipose tissue of beef cattle at weaning and slaughter. In *Proceedings of the 6th World Congress on Genetics Applied to Livest. Prod.*, (Vol. 25), 181.
- Neugebauer, N., Räder, I., Schild, H. J., Zimmer, D., & Reinsch, N. (2009). Evidence for parent-of-origin effects on genetic variability for beef traits. *J. Anim. Sci.* 88, 523-532.

- Nürnberg, K. (2009). Optimising the nutritional profile of beef. In: *Improving the sensory and nutritional quality of fresh meat*. Woodhead Publishing Limited, Cambridge England, Hrsg. CRC Press LLV, 321-341.
- Pitchford, W. S., Deland, M. P. B., Siebert, B. D., Malau-Aduliand, A. E. O., & Bottema, C. D. K. (2002). Genetic variation in fatness and fatty acid composition of crossbred cattle. *J. Anim. Sci.* 80, 2825-2832.
- Quaas, R. L., & Pollak, E. J. (1980). Mixed model methodology for farm and ranch beef cattle testing programs. *J. Anim. Sci.* 51, 1277-1287.
- Reinsch, N., & Guiard, V. Estimable genetic covariance components under mixed Mendelian and imprinted inheritance (in preparation).
- Reinsch, N., Engellandt, T., Schild, H. J., & Kalm, E. (1999). Lack of evidence for bovine Y-chromosomal variation in beef traits. A Bayesian analysis of Simmental data. *J. Anim. Breed. Genet.* 116, 437-445.
- Scheper, J., & Scholz, W. (1985). *DLG-Schnittführung für die Zerlegung der Schlachtkörper von Rind, Kalb, Schwein und Schaf*. DLG Verlag, Frankfurt/M.

Chapter 4

Parent-of-origin modulates direct and maternal genetic effects for
birth weight and gestation length in German Holsteins

Parent-of-origin modulates direct and maternal genetic effects for birth weight and gestation length in German Holsteins

N. Neugebauer*, E. Stamer[†], W. Junge[†], E. Kalm[†], N. Reinsch*

*) Forschungsinstitut für die Biologie landwirtschaftlicher Nutztiere, FBN, Forschungsbereich Genetik und Biometrie, Wilhelm-Stahl-Allee 2, 18196 Dummerstorf, Germany

†) Institut für Tierzucht und Tierhaltung, Christian-Albrechts-Universität zu Kiel, Olshausenstraße 40, 24118 Kiel, Germany

Abstract

The importance of genomic imprinting for traits undergoing the impact of maternal genetic effects was studied by applying gametic models. Thereby the full model covered situations where an imprinting variance occurs both for the direct and the maternal trait. A German Holstein dataset with around 8000 first parity calvings comprised observations for birth weight of calves and gestation length. Though convergence was not obtained for all models required for hypothesis testing it could be concluded that respective proportions of 20% and 12% of the direct genetic variance are due to imprinted loci. Direct heritabilities (68% and 72%) were considerably higher compared to their maternal counterparts (11% and 17%). For gestation length even the maternal trait showed a significant imprinting variance. Despite of computational difficulties the results demonstrate that a separation of Mendelian and imprinting components of the additive genetic variance is in principle possible when maternal genetic effects are present.

Introduction

In the presence of genomic imprinting the expression of the copy of a gene inherited from one parent is at a significantly lower level than for the copy from the other parent. For the statistical treatment of genomic imprinting there exist different statistical models. De Vries et al. (1994) fitted an animal model with either an additional paternal or maternal gametic effect, as initially proposed by Gibson et al. (1988) and Schaeffer et al. (1989). Later Tier and Sölkner (1993) and Engellandt and Tier (2001) applied the same methodology. Essl and Voith (2002) analysed each trait twice, the first time genetic effects were fitted for sires and the second time for dams. Results on imprinting were derived by a comparison of the respective variance components. Stella et al. (2003) analysed the impact of genomic imprinting on litter size of pigs with a gametic model, assuming no maternal effects. Reinsch and Guiard (in preparation) proposed problem solving models with two random genetic effects for each animal. Reduced versions of this model were employed for the analyses of slaughter traits in cattle and pigs (Neugebauer et al., 2009a, b, c). In German Simmental genomic imprinting was found to contribute significantly to the additive genetic variation of mainly carcass-composition traits. This analysis did not consider maternal genetic effects because generally they are assumed of minor importance for slaughter traits. If maternal genetic effects exist, the partitioning of genetic variance components causes more problems. As a consequence our estimates for the imprinting variances of the slaughter traits may, in principle, be contaminated by fractions of maternal genetic variances (Neugebauer et al., 2009b). For traits like birth weight or gestation length it is well-known that maternal effects play a role. Maternal effects in mammals have been studied extensively and the importance especially for the early development has long been recognized (e.g. Willham, 1972; Meyer, 1992; Roehe, 1993). In presence of genetic maternal effects the genotype of the mother has an influence on the phenotype of the offspring. Maternal effects may bias genetic components. The simultaneous consideration of imprinting and maternal effects is difficult and thus the objective of this study was to combine the analy-

sis of genomic imprinting and maternal effects to separate imprinting variance and maternal variance. We used an extended version of the suggested gametic model of Reinsch and Guiard. It enables analysts to estimate the imprinting variance of the direct and the maternal effect. Thus this study is the first comprehensive analysis of direct and maternal effects with considering possible imprinting effects. We tested the theory with a reproduction trait data set because maternal effects particularly occur in life of the female and lags by one generation (Willham, 1980).

Material and Methods

Principle of analysis

In general the model of analysis comprises fixed effects and, among possible additional random effects, two genetic effects, the direct effect and the maternal effect, per animal. Basically the model in matrix notation analogous to Willham (1972) is

$$y = X\beta + Z_{\text{dir}} a_{\text{dir}} + Z_{\text{mat}} a_{\text{mat}} + e. \quad (1)$$

In order to allow for genomic imprinting we describe the direct effect with the gametic effect as sire and gametic effect as dam. The same method is done with the genetic maternal effect, the paternal and maternal gametes of the mother explain the maternal effect of the progeny.

Thus the model in matrix notation is

$$y = X\beta + Z_{d_s} g_{d_s} + Z_{d_d} g_{d_d} + Z_{m_s} g_{m_s} + Z_{m_d} g_{m_d} + e \quad (2)$$

Assuming the covariance of random effects as

$$\text{Var} \begin{bmatrix} g_{d_s} \\ g_{d_d} \\ g_{m_s} \\ g_{m_d} \\ e \end{bmatrix} = \begin{bmatrix} G\sigma_{d_s}^2 & G\sigma_{d_{sd}} & G\sigma_{d_s m_s} & G\sigma_{d_s m_d} & 0 \\ G\sigma_{d_{sd}} & G\sigma_{d_d}^2 & G\sigma_{d_d m_s} & G\sigma_{d_d m_d} & 0 \\ G\sigma_{d_s m_s} & G\sigma_{d_d m_s} & G\sigma_{m_s}^2 & G\sigma_{m_{sd}} & 0 \\ G\sigma_{d_s m_d} & G\sigma_{d_d m_d} & G\sigma_{m_{sd}} & G\sigma_{m_d}^2 & 0 \\ \hline 0 & 0 & 0 & 0 & I\sigma_e^2 \end{bmatrix} = \begin{bmatrix} S \otimes G & 0 \\ \hline 0 & I\sigma_e^2 \end{bmatrix} \quad (3)$$

results in the following mixed model equations (4) on page 101, where y is the vector of observations, X is the design matrix for fixed effects with the corresponding vector β ; Z_{d_s} and Z_{d_d} are the design matrices for direct random gametic effects with the vectors g_{d_s} (paternal expression pattern) and g_{d_d} (maternal expression pattern) for parental gametic effects; Z_{m_s} and Z_{m_d} are the design matrices for maternal random gametic effects with the vectors g_{m_s} (paternal expression pattern) and g_{m_d} (maternal expression pattern) for parental gametic effects of the dam and e is the vector of random residuals. In the mixed model equation the variance components are represent by

$$\begin{bmatrix} \alpha_1 & \alpha_2 & \alpha_3 & \alpha_4 \\ \alpha_2 & \alpha_5 & \alpha_6 & \alpha_7 \\ \alpha_3 & \alpha_6 & \alpha_8 & \alpha_9 \\ \alpha_4 & \alpha_7 & \alpha_9 & \alpha_{10} \end{bmatrix}^{-1} = \frac{1}{\sigma_e^2} \begin{bmatrix} \sigma_{d_s}^2 & \sigma_{d_{sd}} & \sigma_{d_s m_s} & \sigma_{d_s m_d} \\ \sigma_{d_{sd}} & \sigma_{d_d}^2 & \sigma_{d_d m_s} & \sigma_{d_d m_d} \\ \sigma_{d_s m_s} & \sigma_{d_d m_s} & \sigma_{m_s}^2 & \sigma_{m_{sd}} \\ \sigma_{d_s m_d} & \sigma_{d_d m_d} & \sigma_{m_{sd}} & \sigma_{m_d}^2 \end{bmatrix} = \frac{1}{\sigma_e^2} S \quad (5)$$

The matrix G is the usual gametic relationship matrix (Schaeffer et al., 1989) and the variances in the diagonal of S are the gametic variances as sire ($\sigma_{d_s}^2$) and as dam ($\sigma_{d_d}^2$) for the maternal effect, the off-diagonal elements are the corresponding covariances.

$$\begin{bmatrix}
\mathbf{X}'\mathbf{X} & \mathbf{X}'\mathbf{Z}_{d_s} & \mathbf{X}'\mathbf{Z}_{d_d} & \mathbf{X}'\mathbf{Z}_{m_s} & \mathbf{X}'\mathbf{Z}_{m_d} \\
\mathbf{Z}_{d_s}'\mathbf{X} & \mathbf{Z}_{d_s}'\mathbf{Z}_{d_s} + \mathbf{G}^{-1}\alpha_1 & \mathbf{Z}_{d_s}'\mathbf{Z}_{d_d} + \mathbf{G}^{-1}\alpha_2 & \mathbf{Z}_{d_s}'\mathbf{Z}_{m_s} + \mathbf{G}^{-1}\alpha_3 & \mathbf{Z}_{d_s}'\mathbf{Z}_{m_d} + \mathbf{G}^{-1}\alpha_4 \\
\mathbf{Z}_{d_d}'\mathbf{X} & \mathbf{Z}_{d_d}'\mathbf{Z}_{d_s} + \mathbf{G}^{-1}\alpha_2 & \mathbf{Z}_{d_d}'\mathbf{Z}_{d_d} + \mathbf{G}^{-1}\alpha_5 & \mathbf{Z}_{d_d}'\mathbf{Z}_{m_s} + \mathbf{G}^{-1}\alpha_6 & \mathbf{Z}_{d_d}'\mathbf{Z}_{m_d} + \mathbf{G}^{-1}\alpha_7 \\
\mathbf{Z}_{m_s}'\mathbf{X} & \mathbf{Z}_{m_s}'\mathbf{Z}_{d_s} + \mathbf{G}^{-1}\alpha_3 & \mathbf{Z}_{m_s}'\mathbf{Z}_{d_d} + \mathbf{G}^{-1}\alpha_6 & \mathbf{Z}_{m_s}'\mathbf{Z}_{m_s} + \mathbf{G}^{-1}\alpha_8 & \mathbf{Z}_{m_s}'\mathbf{Z}_{m_d} + \mathbf{G}^{-1}\alpha_9 \\
\mathbf{Z}_{m_d}'\mathbf{X} & \mathbf{Z}_{m_d}'\mathbf{Z}_{d_s} + \mathbf{G}^{-1}\alpha_4 & \mathbf{Z}_{m_d}'\mathbf{Z}_{d_d} + \mathbf{G}^{-1}\alpha_7 & \mathbf{Z}_{m_d}'\mathbf{Z}_{m_s} + \mathbf{G}^{-1}\alpha_9 & \mathbf{Z}_{m_d}'\mathbf{Z}_{m_d} + \mathbf{G}^{-1}\alpha_{10}
\end{bmatrix}
\begin{bmatrix}
\hat{\beta} \\
\hat{\vartheta}_{d_s} \\
\hat{\vartheta}_{d_d} \\
\hat{\vartheta}_{m_s} \\
\hat{\vartheta}_{m_d}
\end{bmatrix}
=
\begin{bmatrix}
\mathbf{X}'\mathbf{y} \\
\mathbf{Z}_{d_s}'\mathbf{y} \\
\mathbf{Z}_{d_d}'\mathbf{y} \\
\mathbf{Z}_{m_s}'\mathbf{y} \\
\mathbf{Z}_{m_d}'\mathbf{y}
\end{bmatrix}
\quad (4)$$

A generalisation of the phenotypic variance formula from Thompson (1976) was used for the calculation of the phenotypic variance

$$\sigma_p^2 = \sigma_{d_s}^2 + \sigma_{d_d}^2 + \sigma_{m_s}^2 + \sigma_{m_d}^2 + \sigma_{d_d m_s} + \sigma_{d_d m_d} + \sigma_e^2 \quad (6)$$

The imprinting variance of the direct effect $\sigma_{i_d}^2$ can be expressed as the variance of the difference of the direct gametic effects as sire and as dam

$$\sigma_{i_d}^2 = \sigma_{d_s}^2 + \sigma_{d_d}^2 - 2\sigma_{d_{sd}} \quad (7)$$

The imprinting variance $\sigma_{i_d}^2$ is a part of the additive genetic variance $\sigma_{a_d}^2$ for the direct trait.

The imprinting variance of the maternal effect $\sigma_{i_m}^2$ can be calculated in the same way

$$\sigma_{i_m}^2 = \sigma_{m_s}^2 + \sigma_{m_d}^2 - 2\sigma_{m_{sd}} \quad (8)$$

Again the imprinting variance $\sigma_{i_m}^2$ is part of the additive genetic variance $\sigma_{a_m}^2$ for the maternal trait. The other parts of both additive genetic variances are called Mendelian variance ($\sigma_{M_d}^2$ and $\sigma_{M_m}^2$), which describe the “unimprinted” components of the additive genetic variance (not to be confused with the *Mendelian* sampling variance).

Additionally, each imprinting variance can be divided into a paternal contribution $\sigma_s^2 - \sigma_{sd}$ and a maternal contribution $\sigma_d^2 - \sigma_{sd}$.

Hypothesis testing

For testing hypotheses concerning the gametic covariance components, which are summarised in the matrix S , different versions S of were used. For the *Mendelian* model (no imprinting for the direct and the maternal trait) S was set to

$$S_{00} = \begin{bmatrix} \sigma_d^2 & \sigma_d^2 & \sigma_{dm} & \sigma_{dm} \\ \sigma_d^2 & \sigma_d^2 & \sigma_{dm} & \sigma_{dm} \\ \sigma_{dm} & \sigma_{dm} & \sigma_m^2 & \sigma_m^2 \\ \sigma_{dm} & \sigma_{dm} & \sigma_m^2 & \sigma_m^2 \end{bmatrix} \quad (9)$$

Model two allowed for imprinting of the direct trait, but not for the maternal, therefore

$$S_{10} = \begin{bmatrix} \sigma_{d_s}^2 & \sigma_{d_{sd}} & \sigma_{d_s m} & \sigma_{d_s m} \\ \sigma_{d_{sd}} & \sigma_{d_d}^2 & \sigma_{d_d m} & \sigma_{d_d m} \\ \sigma_{d_s m} & \sigma_{d_d m} & \sigma_m^2 & \sigma_m^2 \\ \sigma_{d_s m} & \sigma_{d_d m} & \sigma_m^2 & \sigma_m^2 \end{bmatrix} \quad (10)$$

Similarly, model three assumed absence of imprinting for the direct trait but presence of imprinting for the maternal trait, thus

$$S_{01} = \begin{bmatrix} \sigma_d^2 & \sigma_d^2 & \sigma_{dm_s} & \sigma_{dm_s} \\ \sigma_d^2 & \sigma_d^2 & \sigma_{dm_d} & \sigma_{dm_d} \\ \sigma_{dm_s} & \sigma_{dm_d} & \sigma_{m_s}^2 & \sigma_{m_{sd}} \\ \sigma_{dm_s} & \sigma_{dm_d} & \sigma_{m_{sd}} & \sigma_{m_d}^2 \end{bmatrix} \quad (11)$$

and model four represents the full model, where genomic imprinting may be present both for the direct and the maternal trait, i.e.,

$$S_{11} = \begin{bmatrix} \sigma_{d_s}^2 & \sigma_{d_{sd}} & \sigma_{d_s m_s} & \sigma_{d_s m_d} \\ \sigma_{d_{sd}} & \sigma_{d_d}^2 & \sigma_{d_d m_s} & \sigma_{d_d m_d} \\ \sigma_{d_s m_s} & \sigma_{d_d m_s} & \sigma_{m_s}^2 & \sigma_{m_{sd}} \\ \sigma_{d_s m_d} & \sigma_{d_d m_d} & \sigma_{m_{sd}} & \sigma_{m_d}^2 \end{bmatrix} \quad (12)$$

For each trait four models - three imprinting models (S_{01} , S_{10} and S_{11}) and one non-imprinting model (S_{00}) - were applied with the aim to calculate the REML log likelihood for each one. Model four (S_{11}) was fitted with the help of the REMLF90 program (Miształ et al.,

2002), the resulting variance components were taken as starting values for ASReml, which was used in order to calculate the REML log likelihood ratio test (RLRT) with appropriate degrees of freedom to test the hypothesis of no imprinting. Since convergence was not obtained in all desired cases simplified models where the correlated maternal effect was replaced by an uncorrelated maternal grandsire effect were additionally fitted to the data. In order to test for genomic imprinting these models were again applied in two versions, the first with $\sigma_{d_s}^2$ and $\sigma_{d_d}^2$ different (i.e. with imprinting) and the second representing the null hypothesis with both variance components equal.

Animals and traits

The analysed data were collected between 1998 and 2007/2008 on three dairy farms in Northern Germany. A part of the data set was already used by Junge et al. (2003) for analysis of calf losses. For analysis only observations from the first lactations without twin births were used. Observations between 265 and 295 days were accepted as plausible values for gestation length, following recommendation of Philipsson et al. (1979). This resulted in 8,375 observations for birth weight and 8,116 observations for gestation length, 51.76 % of the calves were males. In this data set calves descended from 303 bulls. The calves' dams were progeny of 473 different bulls, whereby seven of these bulls appeared as sire in both generations. The means of gestation length were 278.9 days (standard deviation 4.76) and 41.2 kg (standard deviation 4.79) for the birth weight. The pedigree file included 40,024 animals. In 31 cases calves and their mothers had the same sires. As a measure of pedigree completeness the proportion of known great-grandparents of the parent-generation was calculated and resulted in an average value of 71%.

Effects in the model

Fixed effects in the model (13) were adopted from Junge et al. (2003)

$$y_{ijklmno} = HYS_i + S_j + b_1x + b_2x^2 + b_3x^3 + dp_k + dm_l + mp_m + mm_n + e_{ijklmno} \quad (13)$$

Where y is an observation, HYS_i is the fixed effect of the barn year season (in months) interaction, S_j is the fixed effect of sex, b_1, b_2, b_3 are linear, quadratic and cubic regressions on calving age (x), dp_k is the random direct gametic effect as sire, dm_l the random direct gametic effect as dam, mp_m and mm_n are the random maternal gametic effects as sire and as dam and $e_{ijklmnop}$ is the random residual.

The REMLF90 program (Miształ et al. 2002) was used for estimation of variance components of the gametic direct and maternal effect imprinting model (model 4, S_{11}). 320,490 equations had to be solved and other programs ran into convergence problems. The ASReml program (Gilmour *et al.*, 2004) was used for the estimation of all models (starting values for model 4 came from the results of REMLF90). Approximative standard errors for heritabilities and other functions of variance components were derived by applying the delta method.

Results and Discussion

Genetic parameters

Table 1 shows the results of the Mendelian model (model 1, S_{00}). The heritability from the *Mendelian* model is 33% for birth weight and 49% for gestation length for the direct effect. These results are in good agreement with results from the literature (Philipsson et al., 1979; Trus and Wilton, 1988). Other analyses showed larger heritabilities (0.50-0.63) for the direct effect (Groen et al., 1998; Junge et al., 2003; Eriksson et al., 2004; Stamer et al., 2004.). The heritability estimates for the maternal effect are 0.05 (birth weight) and 0.07 (gestation length)

are again in good accordance with the literature (Philipsson, 1976; Junge et al., 2003; Stamer et al., 2004).

Table 1: Overview of both analyzed traits: number of observation (n), raw mean and standard deviation. The direct and maternal heritabilities in the last columns are from the Mendelian model (assuming no imprinting, model 1).

Trait (unit)	n	Mean	Std dev	$h_d^{2\ 1}$	$h_m^{2\ 1}$	r_{dm}
Birth weight (kg)	8,375	41.13	4.83	0.33	0.05	0.58
Gestation length (days)	8,116	278.88	4.76	0.49	0.07	0.01

The estimates (REMLF90) of the gametic direct and maternal effects imprinting model (model 4) are shown in Table 2.

Table 2: Estimates of the gametic direct and maternal effects imprinting model. Variances in the diagonal, covariances above and correlations below the diagonal.

	Birth weight				Gestation length			
	As sire (d)	As dam (d)	As sire (m)	As dam (m)	As sire (d)	As dam (d)	As sire (m)	As dam (m)
As sire (d)	5.914	8.384	-0.969	-0.114	12.65	10.63	-0.015	-1.494
As dam (d)	0.733	22.150	-4.402	-1.785	0.616	23.52	-2.646	-5.866
As sire (m)	-0.036	-0.839	1.244	0.854	-0.003	-0.433	1.589	1.630
As dam (m)	-0.004	-0.307	0.620	1.523	-0.246	-0.708	0.757	2.919

The results showed high differences between gametic variances as sire and as dam. The gametic correlations between the direct and maternal effects are all negative. This is similarly found in the literature, where commonly negative direct and maternal genetic correlations for birth weight and calving traits were reported (Philipsson et al., 1979; Meijering, 1984; Groen et al., 1997; Bennett and Gregory, 2001; Eriksson et al., 2004). The positive correlations be-

tween the direct and maternal trait from the Mendelian model (Table 1) became negative if each of the genetic variance components were divided in two gametic variance components (Table 2). The latter results are in better agreement with the literature than the correlations from the Mendelian model. Residual variances were 8.6 kg² and 5.2 days².

For the estimates shown above REMLF90 indicated convergence with respect to the standard convergence criterion. However, when these estimates were used as starting values in ASReml, the iterative estimation procedure went on and no final convergence could be obtained. This was obviously correct since the REML log likelihood for the REMLF90 estimates, which were obtained with ASReml, were somewhat lower than those from the reduced models (S_{01} , S_{10} and S_{00}), a fact that again indicates lack of convergence.

Convergence was also not obtained for model 2 (S_{10}). Therefore hypotheses concerning the imprinting variance for the direct trait could not be performed. Such tests were however possible for the maternal trait (S_{01} against S_{00}) and resulted in a significant (p-value: 0.014) maternal imprinting variance for gestation length. The imprinting variance $\sigma_{i_m}^2$ was estimated with 48% relative to the maternal additive genetic variance $\sigma_{a_m}^2$, which itself is relatively small.

Estimates (and their standard error in brackets) for the genetic covariance parameters from the simplified models, where maternal effects were modelled as uncorrelated maternal grandsire effects were

$$\begin{bmatrix} 6.064_{(0.97)} & 8.686_{(1.03)} & 0 \\ 8.686_{(1.03)} & 15.690_{(1.43)} & 0 \\ 0 & 0 & 0.3E-06_{(0.0)} \end{bmatrix} \text{ and } \begin{bmatrix} 10.306_{(1.44)} & 10.161_{(1.21)} & 0 \\ 10.161_{(1.21)} & 12.676_{(1.36)} & 0 \\ 0 & 0 & 0.292_{(0.40)} \end{bmatrix}$$

for birth weight and gestation length. Comparisons with their Mendelian counterparts gave RLRT-values of 28.00 and 6.56 and corresponding p-values of 8.32E-07 and 0.038. In con-

clusion for both traits the hypothesis tests from the simplified models let us reject the null hypothesis of no imprinting for the direct trait. Note that the estimates for the direct gametic variances have the same tendencies as the full-model estimates obtained with REMLF90. Direct genetic variances were 21.75 kg^2 and 22.98 days^2 with corresponding direct heritabilities of 68% and 72%. The estimated heritabilities of the maternal trait are 11% for birth weight and 17% for gestation length. Against our expectations heritabilities from the imprinting models were larger in comparison with the heritabilities of the Mendelian model (1) (Table 1). Engellandt and Tier (2002) and Neugebauer et al. (2009) noticed the contrary, i.e. heritabilities of the traits from imprinting models dropped down. Estimates for the direct imprinting variances in absolute numbers were 4.38 kg^2 and 2.66 days^2 , translating into relative imprinting variances of 20% and 12%. Despite of numerical difficulties it could be demonstrated that, at least, in principle, imprinting variances can be estimated for traits which are affected by maternal effects. The relative imprinting variance of 12% and 20% is in the upper range of estimates found for slaughter traits in cattle and pigs (Neugebauer et al., 2009a, b, c). Further analyses are needed, given the non-convergence of some of the sub-models required for particular hypothesis tests.

Conclusion

Parts of the additive genetic variance in cattle reproduction traits are influenced by direct and maternal effects. It is possible to derive the imprinting variance as the variance of the difference between both breeding values per animal for the direct effect. In a reproduction data set on German Holsteins, significant genetic imprinting variance was observed for birth weight and gestation length. Proportions of 12% and 20% of the direct additive genetic variance were controlled by imprinted loci, 48% of the maternal additive genetic variance were influenced by imprinted loci in gestation length.

Literature

- Bennett, G. L., Gregory, K. E., 2001: Genetic (co)variances for calving difficulty score in composite and parental populations of beef cattle: I. Calving difficulty score, birth weight, weaning weight, and postweaning gain. *J. Anim. Sci.* 79: 45-51.
- De Vries, A. G., Kerr, R., Tier, B., Long, T., Meuwissen T. H. E., 1994: Gametic imprinting effects on rate and composition of pig growth. *Theor. Appl. Genet.* 88: 1037-1042.
- Engelland, T., Tier, B., 2002: Genetic variances due to imprinted genes in cattle. *J. Anim. Breed. Genet.* 119 (3): 154-165.
- Eriksson, S., Näsholm, A., Johansson, K., Philipsson, J., 2004: Genetic relationships between calving and carcass traits for Charolais and Hereford cattle in Sweden. *J. Anim. Sci.* 82: 2269-2276.
- Essl, A., Voith, K., 2002: Genomic imprinting effects on dairy- and fitness-related traits in cattle. *J. Anim. Breed. Genet.* 119 (3): 182-189.
- Gibson, J. P., Kennedy, B. W., Schaeffer, L. R., Southwood, O. I., 1988: Gametic models for estimation of autosomally inherited effects that are expressed only when received from either male or female parent (abstract). *J. Dairy Sci.* 71 (suppl 1): 143.
- Gilmour, A. R., Cullis, B. R., Welham, S. J., Gogel, B. J. and Thompson, R., 2004: An efficient computing strategy for prediction in mixed linear models. *Computational Statistics and Data Analysis* 44, 571-586.
- Gilmour, A.R., Gogel, B.J., Cullis, B.R., and Thompson, R. 2006 ASReml User Guide Release 2.0 VSN International Ltd, Hemel Hempstead, HP1 1ES, UK
- Groen, A. F., van Aabel, J. P. J. M., Hulzebosch, A. A., 1998: Calving performance in dairy cattle – influence of maturity of dam on the correlation between direct and indirect effects. *Proceedings of the 6th World Congress on Genetics Applied to Livest. Prod.*, (Vol. 23), 387-390.

- Junge, W., Stamer, E., Reinsch, N., Kalm, E., 2003: Züchterische Möglichkeiten zur Senkung von Kälberverlusten. *Züchtungskunde* 75: 479-488.
- Meijering, A., 1984: Dystocia and stillbirth in cattle – a review of causes, relations and implications. *Livest. Prod. Sci.* 11: 143-177.
- Meyer, K., 1992: Bias and sampling covariances of estimates of variance components due to maternal effects. *Genet. Sel. Evol.* 24: 487-509.
- Misztal, I., Tsuruta, S., Strabel, T., Auvray, B., Druet, T., Lee, D. H., 2002: BLUPF90 and related programs (BGF90). Proceedings of the 7th World Congress on Genetics Applied to Livest. Prod., Montpellier, France. CD-ROM communication 28:07.
- Neugebauer, N., Räder, I., Schild, H. J., Zimmer, D., Reinsch, N., 2009a: Evidence for parent-of-origin effects on genetic variability for beef traits. *J. Anim. Sci.* 88: 523-532.
- Neugebauer, N., Luther, H., Reinsch, N., 2009b: Parent-of-origin effects cause genetic variation in pig performance traits. (accepted for publication in *Animal*).
- Neugebauer, N., Kögel, J., Geuder, U., Kämmerer, S., Reinsch, N., 2009c: On-station heritabilities and parent-of-origin effects for beef traits in German Simmental. (submitted).
- Philipsson, J., 1976: Studies on calving difficulty, stillbirth and associated factors in Swedish cattle breeds, III. Genetic parameters. *Acta. Agric. Scand.* 26: 211-220.
- Philipsson, J., Foulley, J. L., Lederer, J., Liboriussen, T., Osinga, A., 1979: Sire evaluation standards and breeding strategies for limiting dystocia and stillbirth. *Livest. Prod. Sci.* 6: 111-127.
- Reinsch, N. and Guiard, V.: Estimable genetic covariance components under mixed Mendelian and imprinted inheritance (in preparation).
- Roehe, R., Kennedy, B. W., 1993: The influence of maternal effects on accuracy of evaluation of litter size in swine. *J. Anim. Sci.* 71: 2353-2364.

- Schaeffer, L. R., Kennedy, B. W., Gibson, J. P., 1989: The inverse of the gametic relationship matrix. *J. Dairy Sci.* 72(5): 1266-1272.
- Stamer, E., Hafez, S., Junge, W., Kalm, E., 2004: Genetische Parameter für das Geburts- und Absetzgewicht von schwarzbunten weiblichen Kälbern. *Züchtungskunde* 76 (3): 188-195.
- Stella, A., Stalder, K. J., Saxton A. M., Boettcher, P. J., 2003: Estimation of variances for gametic effects on litter size in Yorkshire and Landrace swine. *J. Anim. Sci.* 81: 2171-2178.
- Thompson, R., 1976: The estimation of maternal genetic variances. *Biometrics* 32: 903-917.
- Tier, B. and Sölkner, J., 1993: Analysing gametic variation with an animal model. *Theor. Appl. Genet.* 85: 868-872.
- Trus, D., Wilton, W., 1988: Genetic parameters for maternal traits in beef cattle. *Can. J. Anim. Sci.* 68: 119-128.
- Willham, R. L., 1972: The role of maternal effects in animal breeding: III. Biometrical aspects of maternal effects in animals. *J. Anim. Sci.* 35: 1288-1293.
- Willham, R. L., 1980: Problems in estimating maternal effects. *J. Anim. Sci.* 35: 1288-1293. *Livestock Production Science* 7: 405-418.

General discussion

The intent of this study was to investigate the importance of genomic imprinting for genetic variation in livestock. We used the reduced imprinting model (Reinsch and Guiard, in preparation) to estimate genetic variation in slaughter traits of cattle and pigs caused by imprinting. Additionally, the estimation of imprinting variances in the presence of maternal genetic effects with gametic models was focused.

The general model included two random genetic effects for each animal, which allow for the simultaneous estimation of paternal and maternal imprinting as well as of any combination of full and partial imprinting. The reduced imprinting model, which requires the additive genetic relationship matrix, is especially useful for the evaluation of slaughter traits. In this case maternal genetic effects, whose variance cannot be separated in this model are expected to have no or only a small influence. The separation of imprinting variances of the direct trait and maternal variances is difficult (cf. introduction). The maternal effect cannot be simply described with an additional parameter, because the design matrices for the direct effect as dam and the genetic maternal effect are identical. As a consequence, the estimates of the imprinting variances without the separation of maternal effects may be partly contaminated by the maternal genetic variance. Thus the estimates of the imprinting variances can be interpreted as upper bounds of the imprinting variances and the maternal variances, if existent. Conversely, this also means that all estimates of maternal effects can be contaminated by imprinting variances.

For the estimation of imprinting variances in the presence of maternal genetic effects this model has to be extended. The reduced model cannot be implemented with an additional maternal effect. Using the reduced model for the direct effect and a reduced model for the maternal effect is also not possible because the Mendelian sampling effects are not

independent. Thus, weighting of observations with diagonal matrices is no longer possible. This problem can be solved with a gametic model for the direct and maternal effect with imprinting, which is called the gametic direct and maternal effects imprinting model. Consequently with this gametic model we estimate eight gametic effects per animal. For the prediction of the breeding values we have another possibility with the approach suggested by a reviewer (chapter two). It was proposed to fit a gametic model that explicitly account for a perfect genetic correlation ($\rho=1$) but accounts for imprinting by employing a matrix $G^* = DGD\sigma_g^2$ as covariance matrix of gametic effects, where G is the gametic relationship matrix and D is a diagonal matrix with either one or the parameter f on alternative rows. f represents the difference between the gametic variances and a ratio of $f^2 = \sigma_s^2 / \sigma_d^2$, results in $\sigma_i^2 = \sigma_s^2(1-f)^2$. The inverse of G^* can be computed from the pedigree for known f . This model leads to half the equations as the gametic direct and maternal effects imprinting model, but the approach has to be extended for a correlation different from one. The gametic direct and maternal effects imprinting model is primarily interesting for the estimation of imprinting variances in birth weight, gestation length (e.g. Willham, 1972; Willham, 1980; Meyer, 1992; Roehe, 1993). From a scientific view, it could also be interesting to use the latter model for the estimation of imprinting variances in slaughter traits, in order to separate the imprinting variance from all other components in the presence of both genomic imprinting and maternal effects.

To specify the weights (W) in the reduced model it is important to know the parents of an animal for which an observations is available. In practice this information may not be available. Weights for observations from animals with one or two unknown parents can be derived by assuming the missing parent as non-inbred and unrelated to all other parents.

If the dam is unknown we get

$$w_i = \left[\frac{\frac{1}{2}\sigma_s^2(1-F_{si}) + \left(\frac{1}{2}\sigma_d^2(1-0) + \frac{1}{2}\sigma_d^2\right) + \sigma_e^2}{\frac{1}{2}\sigma_s^2 + \frac{1}{2}\sigma_d^2 + \sigma_e^2} \right]^{-1} = \left[\frac{\frac{1}{2}\sigma_s^2(1-F_{si}) + \sigma_d^2 + \sigma_e^2}{\frac{1}{2}\sigma_s^2 + \frac{1}{2}\sigma_d^2 + \sigma_e^2} \right]^{-1} \quad (1)$$

Where $\frac{1}{2}\sigma_d^2(1-0)$ describes the variance of the Mendelian sampling effect of the maternal gamete and $\frac{1}{2}\sigma_d^2$ accounts for the variation of the additive genetic effect of the unknown dam.

Analogous with an unknown sire w_i becomes:

$$w_i = \left[\frac{\left(\frac{1}{2}\sigma_s^2(1-0) + \frac{1}{2}\sigma_s^2\right) + \frac{1}{2}\sigma_d^2(1-F_{di}) + \sigma_e^2}{\frac{1}{2}\sigma_s^2 + \frac{1}{2}\sigma_d^2 + \sigma_e^2} \right]^{-1} = \left[\frac{\sigma_s^2 + \frac{1}{2}\sigma_d^2(1-F_{di}) + \sigma_e^2}{\frac{1}{2}\sigma_s^2 + \frac{1}{2}\sigma_d^2 + \sigma_e^2} \right]^{-1} \quad (2)$$

And finally with both unknown parents we use

$$w_i = \left[\frac{\left(\frac{1}{2}\sigma_s^2(1-0) + \frac{1}{2}\sigma_s^2\right) + \left(\frac{1}{2}\sigma_d^2(1-0) + \frac{1}{2}\sigma_d^2\right) + \sigma_e^2}{\frac{1}{2}\sigma_s^2 + \frac{1}{2}\sigma_d^2 + \sigma_e^2} \right]^{-1} = \left[\frac{\sigma_s^2 + \sigma_d^2 + \sigma_e^2}{\frac{1}{2}\sigma_s^2 + \frac{1}{2}\sigma_d^2 + \sigma_e^2} \right]^{-1} \quad (3)$$

Chapter one to three present the analyses of slaughter data of German dual-purpose Simmental and Large White animals. For ten traits in the data set of Simmental fattening bulls, we found significant imprinting variances. In the data set of Large Whites we found 19 traits with significant imprinting variances and in the data set of Simmental bulls from the progeny test on station we identified four traits with significant imprinting variances. Chapter four treats the development of the gametic direct and maternal effects model with imprinting. This method was used to study the importance of genomic imprinting for the genetic variation, divided in influences of the direct and the maternal trait, of birth weight and gestation length. We found imprinting variances for the direct effect for both traits and for the maternal effect for gestation length.

For each analysis the imprinting model and the equivalent non-imprinting model (i.e. *Mendelian* model) were fitted to each trait of the data set. From the restricted log likelihood function of both models a restricted likelihood ratio test (RLRT) was calculated in order to

test the hypothesis of imprinting versus the absence of imprinting. In each analysis we carried out a conservative test to minimize the risk of falsely rejecting the null hypothesis. We assumed that the test statistic has a χ^2 distribution with two degrees of freedom under the null hypothesis. The type I error was $\alpha = 0.05$. All analyses together included 108 traits (including direct and maternal traits; chapter four). 36 of them showed a significant imprinting variance. With a type I error to $\alpha = 0.1$ there were 46 of the traits with a significant imprinting variance. A summary of all p-values of the RLRT is shown in the appendix (4.1). An overview on all imprinted traits from the different analyses shows that most of the relative imprinting variances were about around 10% of the additive genetic variance (Figure 1). The high influence of 48% is due to the maternal additive genetic variance, which itself is small, thus the effect of genomic imprinting is marginal for this trait.

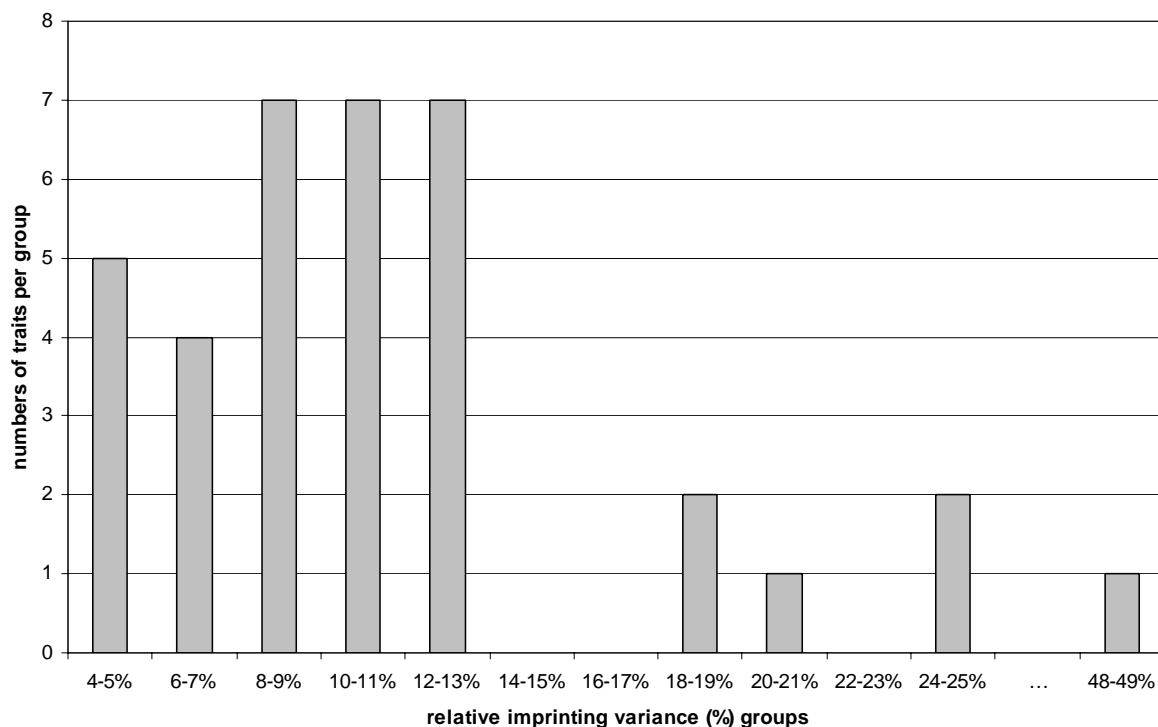


Figure 1: Distribution of the relative imprinting variance of all traits with significant results from chapter one to four. The x-axis summarised each time two percent of the relative imprinting variance in a group. The y-axis presents the number of traits per group.

Most of these traits were related to growth traits and carcass composition. The influence of genomic imprinting on the fatty acid profile in beef seems to be of minor importance. Genetic evaluation could account for such imprinting by predicting two parental breeding values per animal. This might become practically important in particular when bulls are used as terminal sires for beef calves or in cross breeding schemes for pigs: in dam lines one should use the breeding value as dam and in sire lines the breeding value as sire for a better achievement of the breeding goal. Appropriate weighting factors of the genetic effects as sire and as dam in the aggregate genotype can be derived by a special version of the discounted gene-flow method (Börner and Reinsch, 2009).

Conclusions

The reduced imprinting model is particularly useful for the analysis of slaughter traits. If maternal effects exist, the estimation of variance components has to be done with gametic models, which are more complex and computationally intensive. For the prediction of breeding values these models may be simplified. For the analysed slaughter traits in cattle and pigs we found a proportion of up to 25% of the total additive genetic variance which was attributed to be induced by imprinted loci. With the gametic direct and maternal effects model with imprinting the simultaneous estimation of imprinting variances for the direct and maternal trait is, in principle, possible. For birth weight and gestation length we noticed significant imprinting variances for the direct and maternal trait.

Literature

- Börner, V. and Reinsch, N., 2009: Gametic gene flow method accounts for genomic imprinting (accepted for publication in *J. Anim. Breed. Genet.*).
- Engellandt, T., Tier, B., 2002: Genetic variances due to imprinted genes in cattle. *J. Anim. Breed. Genet.* 119 (3): 154-165.
- Meyer, K., 1992: Bias and sampling covariances of estimates of variance components due to maternal effects. *Genet. Sel. Evol.* 24: 487-509.
- Mrode, R. A., 2005: Linear models for the prediction of animal breeding values, 2nd edition. Cromwell Press, Trowbridge, UK.
- Reinsch, N. and Guiard, V.: Estimable genetic covariance components under mixed Mendelian and imprinted inheritance (in preparation).
- Roche, R., Kennedy, B. W., 1993: The influence of maternal effects on accuracy of evaluation of litter size in swine. *J. Anim. Sci.* 71: 2353-2364.
- Willham, R. L., 1972: The role of maternal effects in animal breeding: III. Biometrical aspects of maternal effects in animals. *J. Anim. Sci.* 35: 1288-1293.
- Willham, R. L., 1980: Problems in estimating maternal effects. *J. Anim. Sci.* 35: 1288-1293. *Livest. Prod. Sci.* 7: 405-418.

Summary

A series of four detailed analyses aimed at investigating the importance of genomic imprinting for genetic variation in livestock. A common aspect of all analyses are the applied problem solving models, taking account for both paternal and maternal imprinting simultaneously and allowing for the estimation of an extra genetic variance component, the imprinting variance, which is due to imprinted loci.

The analysis of slaughter data, most of them recorded with an innovative automatic video-imaging device, revealed a significant imprinting variance for ten traits in German Simmental fattening bulls, especially for carcass composition. Between 8% and 25% of the total additive genetic variance was attributed to imprinted loci. Similar results were found in an analysis of a comprehensive data set of Large White slaughter data from the Swiss herdbook pig breeding program. Between 5% and 19% of the total additive genetic variance was significantly controlled by imprinted loci in 19 different traits. Fatty acid profiles from on-station tested male progeny of German Simmental sires appeared almost unaffected by genomic imprinting. Heritability estimates for these traits were quite uniform at about 40%. The last chapter was devoted to two traits, gestation length and birth weight in German Holstein dairy cattle, which are affected by maternal effects. In contrast to slaughter animals, where a special kind of reduced model was used, gametic models had to be fitted to the data. Though convergence was not obtained in all desired cases, it could be concluded that significant parts (12% and 20%, respectively) of the direct genetic variance are due to genomic imprinting.

Zusammenfassung

Mit Hilfe von vier ausführlichen Analysen sollte die Bedeutung der genomischen Prägung für die genetische Variation bei Nutztieren untersucht werden. Das gemeinsame Element dieser Studien sind die angewandten problemorientierten Modelle, welche die paternale und maternale Prägung simultan berücksichtigen. Diese ermöglichen die Schätzung einer zusätzlichen genetischen Komponente, der Imprintingvarianz, welche durch geprägte Genorte hervorgerufen wird.

Die erste Analyse wurde an Schlachtdaten von Mastbullen der Rasse Fleckvieh durchgeführt. In diesen Fleischleistungsdaten, welche überwiegend mittels automatischer Videobildanalyse erhoben wurden, zeigten 10 Merkmale eine signifikante Imprintingvarianz. Meist handelte es sich dabei um Schlachtkörpermerkmale, deren gesamte additiv genetische Varianz zu etwa 8% bis 25% durch geprägte Loci beeinflusst wird. Ähnliche Ergebnisse konnten in einer ausführlichen Schlachtdatenanalyse von Edelschweinen des Schweizer Herdbuchzuchtprogramms gefunden werden. Zwischen 5% und 19% der gesamten additiv genetischen Varianz von 19 dieser Merkmale wurden durch geprägte Genorte kontrolliert. Fettsäuregehalte von Rindfleisch scheinen hingegen kaum von geprägten Genen beeinflusst zu sein, wie Daten aus der Nachkommenschaftsprüfung von Fleckviehbullen auf Station zeigten. Die Heritabilitäten der Fettsäuren lagen in der Regel um 40%. Das letzte Kapitel der Arbeit widmete sich Merkmalen, die maternalen genetischen Einflüssen unterliegen, der Trächtigkeitsdauer und dem Geburtsgewicht Deutscher Holsteins. Im Gegensatz zu den Analysen von Schlachttieren, für die ein spezielles reduziertes Modell genutzt wurde, wurden bei dieser Analyse gametische Modelle verwandt. Obwohl eine Konvergenz der Schätzwerte nicht für alle gewünschten Fälle erreicht wurde, kann aus den Untersuchungen geschlussfolgert werden, dass 12% bzw. 20% der direkten genetischen Varianz der genomischen Prägung unterliegen.

Appendix

Appendix

1	Example analysis.....	122
2	Raw data analysis.....	124
2.1	Defining y-chromosomal and mitochondrial effects	124
2.2	Proportions of known ancestors.....	127
3	ASReml and Remlf90 parameter files	128
3.1	ASReml parameter file for the reduced imprinting	128
3.2	ASReml parameter file for the Mendelian version of the reduced model	129
3.3	Remlf90 parameter file for the gametic direct-maternal effects model.....	130
4	Results details	131
4.1	Detailed RLRT test statistics and corresponding p-values for all traits	131
4.2	Partial derivatives for standard errors	135
5	Calculation of precision for imprinting effects	140
5.1	Calculation of the precision of the imprinting effect estimates with ASReml	140
5.2	Approximate calculation of the precision of the imprinting effect estimates with SAS.....	143
6	English-German dictionary of trait nomenclature	146
7	Literature.....	151

1 Example analysis

The following table shows in detail all ten different matrices $Z_i G Z_j'$ defining the genetic part of the variance of observations $Var(y)$ in the example. The important point is that all these matrices are different, which is a condition for the separation of all covariance components

Table 1: The different covariance structures of the submatrices $G_{ij} = Z_i G Z_j'$, $i, j \in \{d_s, d_d, m_s, m_d\}$:

i	j	4x4 covariance structures ($Z_i G Z_j'$)
1	1	$\begin{bmatrix} 1 & 0 & 0.5 & 0 \\ 0 & 1 & 0 & 0.5625 \\ 0.5 & 0 & 1 & 0 \\ 0 & 0.5625 & 0 & 1 \end{bmatrix}$
1	2	$\begin{bmatrix} 0.125 & 0 & 0.125 & 0 \\ 0.15625 & 0.234375 & 0.15625 & 0.234375 \\ 0.125 & 0 & 0.125 & 0 \\ 0.15625 & 0.234375 & 0.15625 & 0.234375 \end{bmatrix}$
1	3	$\begin{bmatrix} 0 & 0 & 0 & 0 \\ 0.3125 & 0.15625 & 0.3125 & 0.15625 \\ 0 & 0 & 0 & 0 \\ 0.3125 & 0.15625 & 0.3125 & 0.15625 \end{bmatrix}$
1	4	$\begin{bmatrix} 0.25 & 0 & 0.25 & 0 \\ 0 & 0.3125 & 0 & 0.3125 \\ 0.25 & 0 & 0.25 & 0 \\ 0 & 0.3125 & 0 & 0.3125 \end{bmatrix}$

2	2	$\begin{bmatrix} 1 & 0.046875 & 0.5 & 0.046875 \\ 0.046875 & 1 & 0.046875 & 0.625 \\ 0.5 & 0.046875 & 1 & 0.046875 \\ 0.046875 & 0.625 & 0.046875 & 1 \end{bmatrix}$
2	3	$\begin{bmatrix} 0.5 & 0.03125 & 0.5 & 0.03125 \\ 0.09375 & 0.625 & 0.09375 & 0.625 \\ 0.5 & 0.03125 & 0.5 & 0.03125 \\ 0.09375 & 0.625 & 0.09375 & 0.625 \end{bmatrix}$
2	4	$\begin{bmatrix} 0.5 & 0.0625 & 0.5 & 0.0625 \\ 0 & 0.625 & 0 & 0.625 \\ 0.5 & 0.0625 & 0.5 & 0.0625 \\ 0 & 0.625 & 0 & 0.625 \end{bmatrix}$
3	3	$\begin{bmatrix} 0 & 0.125 & 0 & 0.125 \\ 0 & 0.25 & 0 & 0.25 \\ 0 & 0.125 & 0 & 0.125 \\ 0 & 0.25 & 0 & 0.25 \end{bmatrix}$
3	4	$\begin{bmatrix} 0.5 & 0.0625 & 0.5 & 0.0625 \\ 0 & 0.625 & 0 & 0.625 \\ 0.5 & 0.0625 & 0.5 & 0.0625 \\ 0 & 0.625 & 0 & 0.625 \end{bmatrix}$
4	4	$\begin{bmatrix} 1 & 0 & 1 & 0 \\ 0 & 1 & 0 & 1 \\ 1 & 0 & 1 & 0 \\ 0 & 1 & 0 & 1 \end{bmatrix}$

2 Raw data analysis

The analyses of all practical data sets are conducted similarly. In the first step the data sets are checked for their completeness and their correctness and prepared for the individual analysis. A SAS program written by I. Räder, which identifies the animals with the same animal identification number (ID) and a Fortran program written by G. Dietl, which is looking for wrong animal IDs (progeny and one parent or the parents have the same ID number; an animal acts as male and female; more extraction for an animal) was used. In the linear models, the y-chromosomal and mitochondrial inheritance is additionally considered to minimize their influence on the imprinting variance as a consequence of contributions of the y-chromosomal or mitochondrial variance to the covariance between paternal or maternal half sibs. For each observation parental inheritance in the pedigree was traced back until the first male/female founder was identified (2.1). The inbreeding coefficients were calculated with PEST, Version 4.0 (Groeneveld, 1998). For the univariate analyses of the different traits two models are fitted to each trait in our data set, the imprinting model (see above) and the equivalent non-imprinting model (Mendelian model).

2.1 Defining y-chromosomal and mitochondrial effects

A Fortran program (aufbereitung) was used to identify female (male) founders, from which mitochondria (y-chromosomes) of descendents were derived. For each observation maternal (paternal) inheritance was traced back in the pedigree until the first female (male) founder was identified. This female (male) founder number was then assigned to all descendents as identification of the mitochondrial (y-chromosomal) effect [gruppe(i,1)=y-chromosomal inheritance; gruppe(i,2)=mitochondrial inheritance].

```

program aufbereitung
c
c *****
c Program written by I. Räder
c *****
c

c character *(9)   tier,vater,mutter
  integer          anr,k,gr,i,j,grn,lfn,tier,vater,mutter
c character (LEN=15),dimension (51470,3) :: ped
  integer ,dimension (51470,3) :: ped
  integer, dimension(51470,3) :: gruppe

character *(12) einlesedatei
character *(12) ausgabedatei
do i=1,20
write (*,*) ' '
enddo
write (*,*) 'Wie ist der Name der Pedigreedatei ?'
read (*,'(a)') einlesedatei

write (*,*) 'Wie soll die erweiterte Pedigreed. genannt werden?'
read (*,'(a)') ausgabedatei

open (1,file=einlesedatei)
open (2,file=ausgabedatei)
c *****
c
c anr=0
c grn=0
c gr=0
50 format(3(i15,x))
do i=1,51470
read (1,*) tier,vater,mutter
ped(i,1)=tier
ped(i,2)=vater
ped(i,3)=mutter
enddo

do k=1,51470
if (ped(k,2) .ne.0000000000000000) then
do i=1,k-1
if (ped(i,1) .eq. ped(k,2)) then
grn=gruppe(i,1)
gruppe(k,1)=grn
end if
end do
end do

```



```
else
  gr=gr+1
  gruppe(k,1)=gr
end if
end do

grn=0

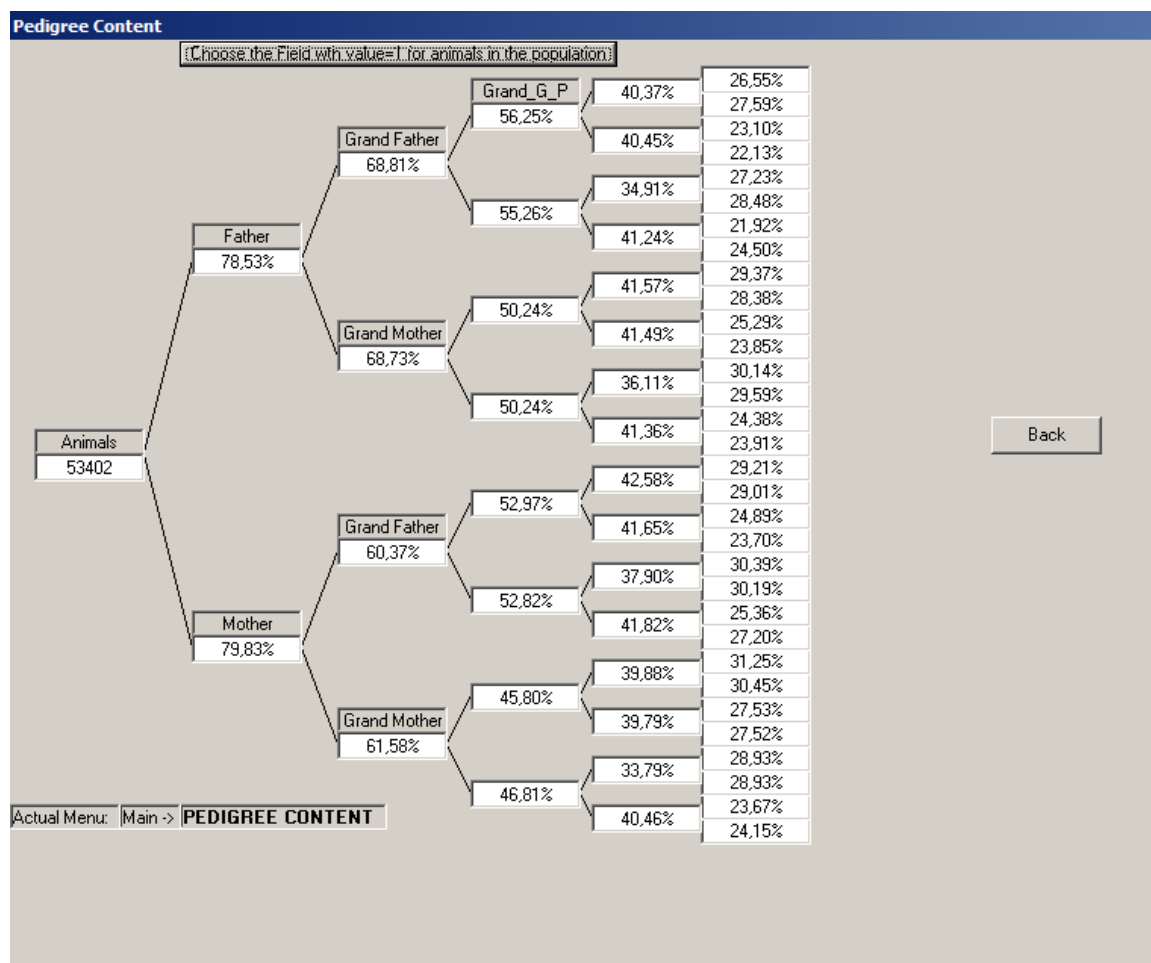
do k=1,51470
if (ped(k,3) .ne. 0000000000000000) then
do i=1,k-1
if (ped(i,1) .eq. ped(k,3)) then
  grn=gruppe(i,2)
  gruppe(k,2)=grn
end if

end do
else
  anr=anr+1
  gruppe(k,2)=anr
end if
end do

20 do i=1,51470
write (2,50) ped(i,1),ped(i,2),ped(i,3),gruppe(i,1),gruppe(i,2)
end do
write (*,*) 'Zahl der Datensätze', k

30 close(1)
close(2)
end
```

2.2 Proportions of known ancestors



This graphic shows the proportions of known ancestors in the Simmental pedigree from chapter three. Individuals designed as “animals” in the output are the parents of the Simmental young bulls, from which the traits were recorded.

3 ASReml and Remlf90 parameter files

3.1 ASReml parameter file for the reduced imprinting

Example for the ASReml parameter file for the reduced imprinting model with weights (chapter three):

```
trait

Alter      !-41.2675 !/12.7537 # mean/std dev
weight     !M0                # weighting
VATER      53403 !P           # Pedigree
MMUTTER    53403 !P
schdat     595   !I           # fixed and random effects
ychrom     37    !I
mitoch     3082 !I
gebtyp     2     !I
lakt       3     !I

#3 ===== Pedigree file
pedigreefile !MAKE

#4 ===== observation file (the first column will delete)
daten !SKIP 1 !AISING
!MAXIT 100

#5 ===== linear model ===== # with !WT for the weighting instruction
trait!WT weight ~ alter schdat gebtyp lakt !r VATER MMUTTER ychrom mitoch

#6 ===== Variance/covariance instruction
0 0 3          # R header line
VATER 2        # variance structure
2 0 US !GP
  0.0534  0.0561  0.174
VATER 0 AINV
mitoch 1
mitoch 0 IDEN  0.05
ychrom 1
ychrom 0 IDEN  0.0001
```

3.2 ASReml parameter file for the Mendelian version of the reduced model

Example for the ASReml parameter file for the Mendelian version (i.e. without imprinting) of the reduced model with weights (chapter three):

```

trait

Alter          !-41.2675 !/12.7537 # mean/std dev
weight         !M0                # weighting
VATER          53403 !P           # Pedigree
MMUTTER        53403 !P
schdat         595   !I           # fixed and random effects
ychrom         37   !I
mitoch         3082 !I
gebtyp         2    !I
lakt           3    !I

#3 ===== Pedigree file
pedigreefile  !MAKE

#4 ===== observation file (the first column will delete)
daten !SKIP 1 !AISING
!MAXIT 100

#5 ===== linear model ===== # with VATER and(MMUTTER,1) to model the sum of
both design matrices (in each column are two assignments)
trait!WT weight ~ alter schdat gebtyp lakt !r VATER and(MMUTTER, 1) ychrom mi-
toch

#6 ===== Variance/covariance instruction
0 0 3          # R header line
VATER 1       # variance structure
0.09696490
VATER 0 AINV
mitoch 1
mitoch 0 IDEN 0.05
ychrom 1
ychrom 0 IDEN 0.0001

```

3.3 Remlf90 parameter file for the gametic direct-maternal effects model

Example for the Remlf90 parameter file for the gametic direct-maternal effects imprinting model (chapter four):

```

DATAFILE
Merkmalgewicht # observation file
NUMBER_OF_TRAITS
1
NUMBER_OF_EFFECTS
9
OBSERVATION(S)
1
WEIGHT(S)

EFFECTS: POSITIONS_IN_DATAFILE NUMBER_OF_LEVELS TYPE_OF_EFFECT [EFFECT NESTED]
9 291 cross # fixed effect
10 2 cross # fixed effect
2 1 cov # linear regression
3 1 cov # quadratic regression
4 1 cov # cubic regression
5 80048 cross # random effect (direct as sire)
6 80048 cross # random effect (direct as dam)
7 80048 cross # random effect (maternal as sire)
8 80048 cross # random effect (maternal as dam)
RANDOM_RESIDUAL VALUES
8.903
RANDOM_GROUP
6 7 8 9
RANDOM_TYPE
user_file
FILE
Pedigree # pedigree file
(CO)VARIANCES
9.540 8.914 -0.6064 -0.1
8.914 18.29 -3.520 -1.455
-0.6064 -3.520 1.109 0.7995
-0.1 -1.455 0.7995 1.608

```

4 Results details

4.1 Detailed RLRT test statistics and corresponding p-values for all traits

In the following for all analysed traits (chapters 1 to 3) RLRT test statistics (for the hypothesis of a zero imprinting variance) and their p-values are given, no matter if the test was significant or not.

Table 2: Details of hypothesis tests from chapter 1.

Trait (unit)	RLRT	p-values
Beefiness score (1 to 15)	9.32	0.009
Meat percentage, %	20.5	0.000
Weight of grade 1 meat, kg	2.64	0.267
Weight of grade 2 meat, kg	0.30	0.861
Weight of grade 3 meat, kg	-	-
Weight of pistola cut, kg	5.88	0.053
Pistola cut / slaughter weight, %	0.22	0.896
Weight of strip loin, kg	6.14	0.046
Weight of strip loin, four cut, kg	4.16	0.125
Weight of strip loin + weight, kg	2.84	0.242
Round weight, kg	6.04	0.049
Hot round weight, kg	6.24	0.044
Weight of round, four cut, kg	3.86	0.145
Topside weight, kg	6.12	0.047
Silverside weight, kg	4.86	0.088
Tenderloin weight, kg	7.46	0.024

Trait (unit)	RLRT	p-values
Rose weight, kg	8.62	0.013
Shoulder weight, kg	4.04	0.133
Weight of forequarter, kg	5.14	0.077
Weight of forequarter, 3 cut, kg	-	-
Total saleable meat weight, kg	4.16	0.125
Killing out percentage, %	29.24	0.000
Fat score (1 to 15)	31.14	0.000
Carcass weight, kg	4.84	0.089
Net BW gain, g/d	5.75	0.056

Table 3: Details of hypothesis tests from chapter 2.

Trait (unit)	RLRT	p-values
Life weight at the end of the test (kg)	1.06	0.589
Average daily gain in lifetime (g/d)	16.94	0.000
Average daily gain during test (g/d)	1.82	0.402
Feed conversion (kg/kg)	8.18	0.017
Average daily feed intake (kg/d)	9.62	0.008
Total feed intake during test (kg)	8.718	0.013
Carcass weight (kg)	0.016	0.602
Carcass length (cm)	0.96	0.619
Percentage of premiums cuts (%)	8.48	0.014
Intramuscular fat content of chop (%)	12.96	0.002
Meat quality (unit)	5.22	0.074
pH 45 minutes <i>post mortem</i>	0.76	0.684
pH 30 hours <i>post mortem</i>	3.46	0.177
Meat reflectance	6.82	0.033
Back fat thickness at ham (cm)	9.66	0.008

Trait (unit)	RLRT	p-values
Back fat thickness at mid of back (cm)	2.16	0.340
Trimmed back fat (kg)	2.84	0.242
Head (kg)	0.88	0.644
Head (%)	3.40	0.182
Feet (kg)	8.14	0.017
Trimmed kidney fat (kg)	16.14	0.000
Kidney fat (%)	15.30	0.000
Pork belly (kg)	21.48	0.000
Pork belly (%)	21.84	0.000
Trimmed shoulder fat (kg)	1.44	0.487
Shoulder (kg)	8.44	0.015
Shoulder (%)	8.88	0.012
Pork chop (kg)	7.90	0.019
Pork chop (%)	10.0	0.007
Trimmed ham fat (kg)	2.03	0.363
Ham (kg)	6.80	0.033
Ham (%)	6.40	0.041
Total trimmed fat (%)	2.22	0.330

Table 4: Details of hypothesis tests from chapter 3.

Trait (unit)	RLRT	p-values
Initial weight (kg)	3.80	0.150
Carcass weight (kg)	0.80	0.670
Net weight gain (g/d)	1.20	0.549
Average daily gain in lifetime (g/d)	0.68	0.712
Average daily gain during test (g/d)	0.42	0.811

Trait (unit)	RLRT	p-values
Average daily gain during the last test period (g/d)	1.24	0.538
Kidney fat (%)	3.99	0.136
Slaughterhouse waste (%)	4.36	0.113
Pistola cut/slaughter weight (%)	2.22	0.330
Killing out percentage (%)	1.76	0.415
Area of <i>M. longissimus dorsi</i> (cm ²)	0.96	0.619
Lean meat proportion (%)	0.24	0.887
Bone proportion (%)	2.84	0.062
Tendon fat proportion (%)	1.08	0.583
Meat percentage (%)	2.50	0.081
E-U-R-O-P-beefiness class (15 classes) ^a	11.89	0.003
Fat score (1-15 scores) ^a	-0.08*	-
Muscularity score (1-15 scores) ^b	6.64	0.036
Loss (%) ^c	10.49	0.005
Storage loss (%)	-	-
Cooking loss (%)	0.80	0.670
pH 45 minutes <i>post mortem</i>	-1.10*	-
pH 24 hours <i>post mortem</i>	3.58	0.167
pH 12 days <i>post mortem</i>	3.52	0.173
Colour L	2.70	0.260
Colour a	-0.02*	-
Colour b	2.60	0.273
Intramuscular fat	0.40	0.819
Average shear force (N)	-0.08*	-
Average energy of shear force (mJ)	0.40	0.819
C 14:0 (mg/100g meat)	0.74	0.691
C 14:1 <i>cis</i> -9 (mg/100g meat)	1.06	0.589
C 16:0 (mg/100g meat)	1.80	0.407

Trait (unit)	RLRT	p-values
C 16:1 <i>cis</i> -9 (mg/100g meat)	1.30	0.522
C 18:0 (mg/100g meat)	1.10	0.577
C 18:1 <i>trans</i> (mg/100g meat)	2.30	0.317
C 18:1 <i>cis</i> -9 (mg/100g meat)	2.20	0.333
C 18:1 <i>cis</i> -11 (mg/100g meat)	2.64	0.267
C 18:3 <i>n</i> -3 (mg/100g meat)	2110	0.175
C 22:4 (mg/100g meat)	2110	0.324
CLA c9 t11 (mg/100g meat)	2.18	0.336
SFA (mg/100g meat) ^d	1.20	0.549
MUFA (mg/100g meat) ^e	2.00	0.368
PUFA (mg/100g meat) ^f	5.99	0.050
Ratio of C14:1 to C14:0	-	-
Ratio of C16:1 to C16:0	5.08	0.079
Ratio of C18:1 to C18:0	5.02	0.074

- a) EU carcass grading system
b) Scoring of live animals
c) Loss=warm carcass weight – cold carcass weight
d) The sum of saturated fatty acids (SFA) was calculated as: C10:0 + C12:0 + C14:0 + C15:0 + C16:0 + C17:0 + C18:0 + C20:0 + C22:0 + C24:0
e) The sum of monounsaturated fatty acids (MUFA) was calculated as: C14:1 + C16:1 + C18:1*trans* + C18:1*cis*9 + C18:1*cis*11 + C20:1 + C22:1 + C24:1
f) The sum of polyunsaturated fatty acids (PUFA) is the sum of n-6 and n-3 FA
- *) negative RLRT-value due to numerical inaccuracies

4.2 Partial derivatives for standard errors

In order to calculate standard errors of various functions of genetic covariance components (e.g. the relative imprinting variance or heritabilities) partial derivatives of these functions with respect to covariance parameters are needed. These partial derivatives are given here in detail:

For all calculations the matrix (V) of the Variance of Variance components (.vvp file from ASReml) was used:

$$\text{Var}(f(\theta)) = \left[\frac{\partial f(\theta)}{\partial \theta} \right] V \left[\frac{\partial f(\theta)}{\partial \theta} \right]'$$

For simplification we set:

$$\frac{1}{2}\sigma_s^2 = x_1$$

$$\frac{1}{2}\sigma_d^2 = x_2$$

$$\frac{1}{2}\sigma_{sd} = x_{12}$$

$$\sigma_w^2, \sigma_b^2, \sigma_f^2, \sigma_e^2 = x_3, x_4, x_5, x_8 \text{ (random effects of the analysis in chapter two)}$$

$$\sigma_m^2 = x_6 \text{ (random effect: mitochondrial inheritance)}$$

$$\sigma_y^2 = x_7 \text{ (random effect: y-chromosomal inheritance)}$$

Relative imprinting variance

$$\frac{\sigma_i^2}{\sigma_a^2} = \frac{2\left(\frac{1}{2}\sigma_s^2\right) + 2\left(\frac{1}{2}\sigma_d^2\right) - 4\left(\frac{1}{2}\sigma_{sd}\right)}{2\left(\frac{1}{2}\sigma_s^2\right) + 2\left(\frac{1}{2}\sigma_d^2\right)}$$

$$f^*(\theta) = 2 \left(\frac{x_1 + x_2 - 2x_{12}}{x_1 + x_2} \right)$$

$$\frac{\partial f^*(\theta)}{\partial \theta} = \left[\frac{\partial f^*(\theta)}{\partial x_1}, \frac{\partial f^*(\theta)}{\partial x_2}, \frac{\partial f^*(\theta)}{\partial x_{12}} \right] = \left[\frac{4x_{12}}{(x_1 + x_2)^2}, \frac{4x_{12}}{(x_1 + x_2)^2}, \frac{-4}{x_1 + x_2} \right]$$

Correlation between genetic effects

$$r = \frac{\sigma_{sd}}{\sqrt{\sigma_s^2 \sigma_d^2}}$$

$$r = \frac{2\left(\frac{1}{2}\sigma_{sd}\right)}{\sqrt{2\left(\frac{1}{2}\sigma_s^2\right) \cdot 2\left(\frac{1}{2}\sigma_d^2\right)}}$$

$$f^*(\theta) = \frac{2x_{12}}{\sqrt{2x_1 \cdot 2x_2}}$$

$$\frac{\partial f^*(\theta)}{\partial \theta} = \left[\frac{\partial f^*(\theta)}{\partial x_1}, \frac{\partial f^*(\theta)}{\partial x_2}, \frac{\partial f^*(\theta)}{\partial x_{12}} \right] = \left[-\frac{x_{12}x_2}{2(x_1x_2)^{\frac{3}{2}}}, -\frac{x_{12}x_1}{2(x_1x_2)^{\frac{3}{2}}}, \frac{1}{\sqrt{x_1+x_2}} \right]$$

Heritability of the compact model

$$h^2 = \frac{2\left(\frac{1}{2}\sigma_s^2\right) + 2\left(\frac{1}{2}\sigma_d^2\right)}{2\left(\frac{1}{2}\sigma_s^2\right) + 2\left(\frac{1}{2}\sigma_d^2\right) + \sigma_w^2 + \sigma_b^2 + \sigma_f^2 + \sigma_m^2 + \sigma_y^2 + \sigma_e^2}$$

$$f^*(\theta) = \frac{2(x_1) + 2(x_2)}{2(x_1) + 2(x_2) + x_3 + x_4 + x_5 + x_6 + x_7 + x_8}$$

$$\frac{\partial f^*(\theta)}{\partial \theta} = \left[\frac{\partial f^*(\theta)}{\partial x_1}, \frac{\partial f^*(\theta)}{\partial x_2}, \frac{\partial f^*(\theta)}{\partial x_3}, \frac{\partial f^*(\theta)}{\partial x_4}, \frac{\partial f^*(\theta)}{\partial x_5}, \frac{\partial f^*(\theta)}{\partial x_6}, \frac{\partial f^*(\theta)}{\partial x_7}, \frac{\partial f^*(\theta)}{\partial x_8} \right]$$

with:

$$\frac{\partial f^*(\theta)}{\partial x_1} = \frac{\partial f^*(\theta)}{\partial x_2} = \frac{2(x_3 + x_4 + x_5 + x_6 + x_7 + x_8)}{(2(x_1) + 2(x_2) + x_3 + x_4 + x_5 + x_6 + x_7 + x_8)^2}$$

and:

$$\begin{aligned} \frac{\partial f^*(\theta)}{\partial x_3} &= \frac{\partial f^*(\theta)}{\partial x_4} = \frac{\partial f^*(\theta)}{\partial x_5} = \frac{\partial f^*(\theta)}{\partial x_6} = \frac{\partial f^*(\theta)}{\partial x_7} = \frac{\partial f^*(\theta)}{\partial x_8} \\ &= \frac{-2(x_1 + x_2)}{(2(x_1) + 2(x_2) + x_3 + x_4 + x_5 + x_6 + x_7 + x_8)^2} \end{aligned}$$

Heritability of the Mendelian model

$$h^2 = \frac{2\left(\frac{1}{2}\sigma_s^2\right)}{2\left(\frac{1}{2}\sigma_s^2\right) + \sigma_w^2 + \sigma_b^2 + \sigma_f^2 + \sigma_m^2 + \sigma_y^2 + \sigma_e^2}$$

$$f^*(\theta) = \frac{2(x_1)}{2(x_1) + x_3 + x_4 + x_5 + x_6 + x_7 + x_8}$$

$$\frac{\partial f^*(\theta)}{\partial \theta} = \left[\frac{\partial f^*(\theta)}{\partial x_1}, \frac{\partial f^*(\theta)}{\partial x_3}, \frac{\partial f^*(\theta)}{\partial x_4}, \frac{\partial f^*(\theta)}{\partial x_5}, \frac{\partial f^*(\theta)}{\partial x_6}, \frac{\partial f^*(\theta)}{\partial x_7}, \frac{\partial f^*(\theta)}{\partial x_8} \right]$$

with:

$$\frac{\partial f^*(\theta)}{\partial x_1} = \frac{2(x_3 + x_4 + x_5 + x_6 + x_7 + x_8)}{(2(x_1) + x_3 + x_4 + x_5 + x_6 + x_7 + x_8)^2}$$

and:

$$\frac{\partial f^*(\theta)}{\partial x_3} = \frac{\partial f^*(\theta)}{\partial x_4} = \frac{\partial f^*(\theta)}{\partial x_5} = \frac{\partial f^*(\theta)}{\partial x_6} = \frac{\partial f^*(\theta)}{\partial x_7} = \frac{\partial f^*(\theta)}{\partial x_8} = \frac{-2x_1}{(2(x_1) + x_3 + x_4 + x_5 + x_6 + x_7 + x_8)^2}$$

mitochondrial inheritance

$$f^*(\theta) = \frac{x_6}{2(x_1) + 2(x_2) + x_3 + x_4 + x_5 + x_6 + x_7 + x_8}$$

$$\frac{\partial f^*(\theta)}{\partial \theta} = \left[\frac{\partial f^*(\theta)}{\partial x_1}, \frac{\partial f^*(\theta)}{\partial x_2}, \frac{\partial f^*(\theta)}{\partial x_3}, \frac{\partial f^*(\theta)}{\partial x_4}, \frac{\partial f^*(\theta)}{\partial x_5}, \frac{\partial f^*(\theta)}{\partial x_6}, \frac{\partial f^*(\theta)}{\partial x_7}, \frac{\partial f^*(\theta)}{\partial x_8} \right]$$

with:

$$\frac{\partial f^*(\theta)}{\partial x_1} = \frac{\partial f^*(\theta)}{\partial x_2} = \frac{-2x_6}{(2(x_1) + 2(x_2) + x_3 + x_4 + x_5 + x_6 + x_7 + x_8)^2}$$

and:

$$\frac{\partial f^*(\theta)}{\partial x_3} = \frac{\partial f^*(\theta)}{\partial x_4} = \frac{\partial f^*(\theta)}{\partial x_5} = \frac{\partial f^*(\theta)}{\partial x_7} = \frac{\partial f^*(\theta)}{\partial x_8} = \frac{-x_6}{(2(x_1) + 2(x_2) + x_3 + x_4 + x_5 + x_6 + x_7 + x_8)^2}$$

and:

$$\frac{\partial f^*(\theta)}{\partial x_6} = \frac{2(x_1) + 2(x_2) + x_3 + x_4 + x_5 + x_7 + x_8}{(2(x_1) + 2(x_2) + x_3 + x_4 + x_5 + x_6 + x_7 + x_8)^2}$$

y-chromosomale inheritance

$$f^*(\theta) = \frac{x_7}{2(x_1) + 2(x_2) + x_3 + x_4 + x_5 + x_6 + x_7 + x_8}$$

$$\frac{\partial f^*(\theta)}{\partial \theta} = \left[\frac{\partial f^*(\theta)}{\partial x_1}, \frac{\partial f^*(\theta)}{\partial x_2}, \frac{\partial f^*(\theta)}{\partial x_3}, \frac{\partial f^*(\theta)}{\partial x_4}, \frac{\partial f^*(\theta)}{\partial x_5}, \frac{\partial f^*(\theta)}{\partial x_6}, \frac{\partial f^*(\theta)}{\partial x_7}, \frac{\partial f^*(\theta)}{\partial x_8} \right]$$

with:

$$\frac{\partial f^*(\theta)}{\partial x_1} = \frac{\partial f^*(\theta)}{\partial x_2} = \frac{-2x_7}{(2(x_1) + 2(x_2) + x_3 + x_4 + x_5 + x_6 + x_7 + x_8)^2}$$

and:

$$\frac{\partial f^*(\theta)}{\partial x_3} = \frac{\partial f^*(\theta)}{\partial x_4} = \frac{\partial f^*(\theta)}{\partial x_5} = \frac{\partial f^*(\theta)}{\partial x_6} = \frac{\partial f^*(\theta)}{\partial x_8} = \frac{-x_7}{(2(x_1) + 2(x_2) + x_3 + x_4 + x_5 + x_6 + x_7 + x_8)^2}$$

and:

$$\frac{\partial f^*(\theta)}{\partial x_7} = \frac{2(x_1) + 2(x_2) + x_3 + x_4 + x_5 + x_6 + x_8}{(2(x_1) + 2(x_2) + x_3 + x_4 + x_5 + x_6 + x_7 + x_8)^2}$$

5 Calculation of precision for imprinting effects

5.1 Calculation of the precision of the imprinting effect estimates with ASReml

For the reduced imprinting model we estimated two breeding values per animal, the difference between these breeding values is the imprinting effect.

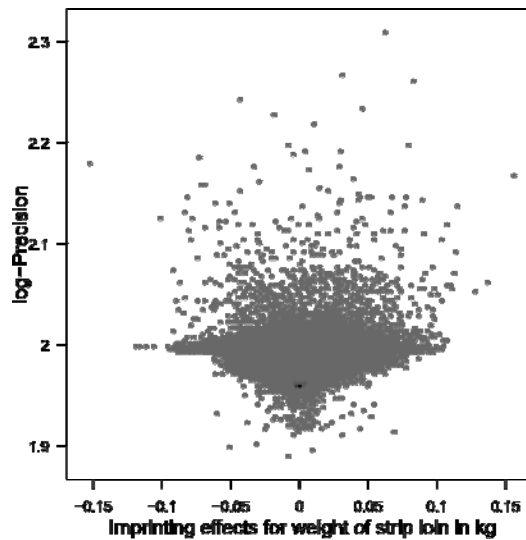


Figure 1: Example of the figures of imprinting effect (from chapter one).

For the figures (example: Figure 1) showing the variation of the imprinting effect (chapter 1 and 2) we calculated the log-precision of the imprinting effects. We used the predictions (Gilmour et al., 2004; Welham et al., 2004) of ASReml, which were formed in the final iteration. For the $Var[(\hat{a}_d - a_d) - (\hat{a}_s - a_s)]$ we needed a detour because the variance matrix for estimates is only available for equations in the dense portion.

For each animal (i) in the pedigree the statement:

```
'PREDICT father i mother i !ONLYUSE father mother'
```

In the ASReml parameter file gives the following standard error

$$SE\left[\left(\hat{a}_{d(i)} - a_{d(i)}\right) + \left(\hat{a}_{s(i)} - a_{s(i)}\right)\right] = z_{(i)} \quad (5.1)$$

and this value squared leads to

$$\begin{aligned} Var\left[\left(\hat{a}_{d(i)} - a_{d(i)}\right) + \left(\hat{a}_{s(i)} - a_{s(i)}\right)\right] &= z_{(i)}^2 \\ &= Var\left(\hat{a}_{d(i)} - a_{d(i)}\right) + Var\left(\hat{a}_{s(i)} - a_{s(i)}\right) + 2Cov\left[\left(\hat{a}_{d(i)} - a_{d(i)}\right), \left(\hat{a}_{s(i)} - a_{s(i)}\right)\right]. \end{aligned} \quad (5.2)$$

Similiary the statements ‘PREDICT mother i !ONLYUSE mother’ and ‘PREDICT father i !ONLYUSE father’ lead to $SE\left(\hat{a}_{d(i)} - a_{d(i)}\right) = x_{(i)}$ and $SE\left(\hat{a}_{s(i)} - a_{s(i)}\right) = y_{(i)}$ respectively.

Again with these values squared we yield

$$Var\left(\hat{a}_{d(i)} - a_{d(i)}\right) = x_{(i)}^2 \quad (5.3)$$

and

$$Var\left(\hat{a}_{s(i)} - a_{s(i)}\right) = y_{(i)}^2. \quad (5.4)$$

Now subtracting (5.3) and (5.4) from (5.2) gives

$$2Cov\left[\left(\hat{a}_{d(i)} - a_{d(i)}\right), \left(\hat{a}_{s(i)} - a_{s(i)}\right)\right] = z_{(i)}^2 - x_{(i)}^2 - y_{(i)}^2 \quad (5.5)$$

and lastly

$$Var\left[\left(\hat{a}_{d(i)} - a_{d(i)}\right) - \left(\hat{a}_{s(i)} - a_{s(i)}\right)\right] = x_{(i)}^2 + y_{(i)}^2 - \left(z_{(i)}^2 - x_{(i)}^2 - y_{(i)}^2\right). \quad (5.6)$$

Here is an example for the ASReml parameter file with predictions for one animal for the imprinting model (chapter two):

```

Trait
SGkg          !-82.2694 !/2.7209
VATER         40000  !P
MMUTTER      40000  !P
SEX           2      !I
STALL        374    !I
HERKUNFT     678    !I
WURF         8887   !I
BUCHT       3847   !I

#3 ===== Pedigree file
pedigreefile !MAKE

#4 ===== observation file (the first column will delete and the data will not reorder)
Traitfile !SKIP 1 !AISING !NOREORDER
!MAXIT 1 # only 1 iteration

#5 ===== linear model =====

Trait ~ SGkg SEX STALL !r VATER MMUTTER HERKUNFT WURF BUCHT

# Predict instruction for animal 11821 (the results will calculated with 9 internal deci-
mal places)

PREDICT VATER 11821 MMUTTER 11821 !ONLYUSE VATER MMUTTER !DEC 9
PREDICT VATER 11821 !ONLYUSE VATER !DEC 9
PREDICT MMUTTER 11821 !ONLYUSE MMUTTER !DEC 9

#6 ===== Variance/covariance instruction
0 0 4          # R header line
VATER 2       # variance structure
2 0 US !GU
0.212626  0.165195  0.198838
VATER 0 AINV
HERKUNFT 1
HERKUNFT 0 IDEN          0.244365691
BUCHT 1
BUCHT 0 IDEN            0.144296644
WURF 1
WURF 0 IDEN             0.40975242

```

5.2 Approximate calculation of the precision of the imprinting effect estimates with SAS

Using the genetic correlation (r ; between both breeding values per animal) leads to approximate standard errors for the imprinting effects which are much easier to compute:

$$\text{Var}\left(\hat{a}_{d(i)} - a_{d(i)}\right) + \text{Var}\left(\hat{a}_{s(i)} - a_{s(i)}\right) - \left[\overbrace{2r}^{\text{"Cov"}} \sqrt{\left(\hat{a}_{d(i)} - a_{d(i)}\right)\left(\hat{a}_{s(i)} - a_{s(i)}\right)} \right] \quad (5.7)$$

The exact correlation between estimates of genetic effects is replaced by an estimate for the correlation between true genetic effects. The following macro, written for SAS, uses the ASReML solution output (.sln) and the genetic correlation to calculate the “imprecise” precision for the imprinting effect estimates:

Zur Lösung von gemischten Modellen mit Imprinting, Schritt 1. Daten einlesen. Da Daten hintereinander ausgegeben wurden, hier aber zusammen benutzt werden müssen, werden sie separat eingelesen und dann gemergt. Es ist unter Daten der Einlesepfad, unter kWert die Korrelation der Varianzen und unter Name der des Merkmals einzutragen;

```
%macro feuerplot (Daten, kWert, Name);
  data muetterlich;
    infile &Daten;
    input Eltern $ 1-11 Tier 25-35 Wert1 45-60 Fehler1 61-71;
    if Eltern='MMUTTER';
  run;

  data vaeterlich;
    infile &Daten;
    input Eltern $ 1-11 Tier 25-35 Wert2 45-60 Fehler2 61-71;
    if Eltern='VATER';
  run;

  proc sort data=muetterlich;
    by Tier;
  run;

  proc sort data=vaeterlich;
    by Tier;
  run;
```

* Im merge-Schritt werden die zusätzlich benötigten Parameter berechnet. Aus der Fehlervarianz wird mit dem Kehrwert die Präzision nach Reinsch ermittelt; diese stellt eine vereinfachte Gangart dar, die genauen Präzisionen können aber auch ermittelt werden, dies ist nur ein sehr Rechenaufwendiger Schritt*;

```
data beide;
    merge muetterlich (in=A) vaeterlich (in=B);
    by Tier;
    if A and B;
        V1=Fehler1*Fehler1;
        V2=Fehler2*Fehler2;
        Diff=Wert1-Wert2;
        P1=log10(1/V1);
        P2=log10(1/V2);
        PDiff=log10(1/(V1+V2-2*&kWert*sqrt(V1*V2)));
        drop V1 V2;
run;
```

-- Platz schaffen, Einlesedateien werden nicht mehr benötigt--;

```
proc datasets;
    delete vaeterlich muetterlich;
run;
```

Schätzwerte gegen Präzision plotten;

Zuchtwert als Mutter;

```
options device=jpeg gsfname=abc;
proc gplot data=beide;
    filename abc 'd:\Versuch\dat'&Name'mutter.jpg';
    symbol1 F=Marker V='R';
    title 'Zuchtwert als Mutter bei '&Name;
    plot P1*Wert1;
run;
```

Zuchtwert als Vater;

```
options device=jpeg gsfname=def;
proc gplot data=beide;
    filename def 'd:\Versuch\dat'&Name'vater.jpg';
    symbol1 F=Marker V='Q';
```

```
        title 'Zuchtwert als Vater bei '&Name;
        plot P2*Wert2;
run;
*Zuchtwert als Mutter und als Vater in einem plot*;
options device=jpeg gsfname=ghi;
proc gplot data=beide;
    filename ghi 'd:\Versuch\dat'&Name'beide.jpg';
    symbol1 F=Marker V='R';
    symbol2 F=Marker V='Q';
    title 'Zuchtwerte als Mutter und als Vater bei '&Name;
    plot P2*Wert2 P1*Wert1 / overlay;
run;
*Differenz der Zuchtwerte*;
options device=jpeg gsfname=jkl;
proc gplot data=beide;
    filename jkl 'd:\Versuch\dat'&Name'diff.jpg';
    symbol1 V='Plus';
    title 'Differenz der Zuchtwerte bei '&Name;
    plot PDiff*Diff;
run;

*Ausgabe der Werte zur Grafikbearbeitung durch andere Programme (R)*;

    filename ausg 'd:\Versuch\dat'&Name'alles';
    data _null_;
    file ausg;
    set beide;
    put Wert1 P1 Wert2 P2 Diff PDiff;
run;

*-- Platz schaffen, Ausgabedatei wird nicht mehr benötigt--*;
proc datasets;
    delete beide;
run;
%mend;
%feuerplot('d:\trait.sln', '0.8034', 'trait');
```

6 English-German dictionary of trait nomenclature

Table 5: Dictionary for chapter 1 traits.

English	Deutsch
Beefiness score (1 to 15)	Fleischigkeitsklasse
Meat percentage, %	Fleischanteil
Weight of grade 1 meat, kg	Gewicht des Fleisches der Klasse 1
Weight of grade 2 meat, kg	Gewicht des Fleisches der Klasse 2
Weight of grade 3 meat, kg	Gewicht des Fleisches der Klasse 3
Weight of pistola cut, kg	Pistolengewicht
Pistola cut / slaughter weight, %	Pistolengewicht/ Schlachtgewicht
Weight of strip loin, kg	Roastbeefgewicht
Weight of strip loin, four cut, kg	Roastbeefgewicht 4er Schnitt
Weight of strip loin + weight, kg	Roastbeefgewicht + Gewicht
Round weight, kg	Keulengewicht
Hot round weight, kg	Keulengewicht warm
Weight of round, four cut, kg	Keulengewicht 4er Schnitt
Topside weight, kg	Oberschalengewicht
Silverside weight, kg	Unterschalengewicht
Tenderloin weight, kg	Filetgewicht
Rose weight, kg	Blumengewicht
Shoulder weight, kg	Buggewicht
Weight of forequarter, kg	Vorderviertelgewicht
Weight of forequarter, 3 cut, kg	Vorderviertelgewicht 3er Schnitt
Total saleable meat weight, kg	Gesamtfleischgewicht
Killing out percentage, %	Ausschlachtung
Fat score (1 to 15)	Fettklasse
Carcass weight, kg	Schlachtgewicht
Net BW gain, g/d	Nettozunahme

Table 6: Dictionary for chapter 2 traits.

English	Deutsch
Life weight at the end of the test (kg)	Lebendgewicht
Average daily gain in lifetime (g/d)	Lebenstagszunahme
Average daily gain during test (g/d)	Masttagszunahme
Feed conversion (kg/kg)	Futtermverwertung
Average daily feed intake (kg/d)	Durchschnittlicher täglicher Futtermverzehr
Total feed intake during test (kg)	Gesamter Futtermverzehr
Carcass weight (kg)	Schlachtgewicht
Carcass length (cm)	Schlachtkörperlänge
Percentage of premiums cuts (%)	Anteil wertvoller Teilstücke
Intramuscular fat content of chop (%)	Anteil intramuskuläres Fett im Kotelett
Meat quality (unit)	Fleischqualitätsnote
pH 45 minutes <i>post mortem</i>	pH-Wert 45min nach der Schlachtung
pH 30 hours <i>post mortem</i>	pH-Wert 30 h nach der Schlachtung
Meat reflectance	Fleischhelligkeit, 30 h nach der Schlachtung (andere Messtechnik als in Kapitel 3, deshalb wird eine andere Übersetzung genutzt)
Back fat thickness at ham (cm)	Rückenspeckdicke an der Kruppe
Back fat thickness at mid of back (cm)	Rückenspeckdicke in der Mitte
Trimmed back fat (kg)	Gewicht des Rückenspeckes
Head (kg)	Gewicht des Kopfes
Head (%)	Gewicht des Kopfes im Verhältnis zum Schlachtkörper
Feet (kg)	Gewicht der Füße
Trimmed kidney fat (kg)	Gewicht des Nierenfettes
Kidney fat (%)	Gewicht des Nierenfettes im Verhältnis zum Schlachtkörper
Pork belly (kg)	Gewicht des Bauches
Pork belly (%)	Gewicht des Bauches im Verhältnis zum Schlachtkörper
Trimmed shoulder fat (kg)	Gewicht des Schulterfettes
Shoulder (kg)	Gewicht der Schulter
Shoulder (%)	Gewicht der Schulter im Verhältnis zum Schlachtkörper

English	Deutsch
Pork chop (kg)	Gewicht des Koteletts
Pork chop (%)	Gewicht des Koteletts im Verhältnis zum Schlachtkörper
Trimmed ham fat (kg)	Gewicht des Schinkenfettes
Ham (kg)	Gewicht des Schinkens
Ham (%)	Gewicht des Schinkens im Verhältnis zum Schlachtkörper
Total trimmed fat (%)	Anteil des Auflagefettes

Table 7: Dictionary for chapter 3 traits.

English	Deutsch
Initial weight (kg)	Einstellungsgewicht
Carcass weight (kg)	Schlachtgewicht
Net BW gain (g/d)	Nettozunahme
Average daily gain in lifetime (g/d)	Tägliche Zunahme seit Geburt
Average daily gain during test (g/d)	Tägliche Zunahme in der Prüfung
Average daily gain during the last test period (g/d)	Tägliche Zunahme im letzten Prüfungsabschnitt
Kidney fat (%)	Gesamtnierentalg/Schlachtgewicht
Slaughterhouse waste (%)	Schlachtabschnitte nach 4. DVO/ Schlachtgewicht
Pistola cut/slaughter weight (%)	Pistolengewicht/Schlachtgewicht
Killing out percentage (%)	Ausschlachtung (Schlachtkörper warm – Schlachthofgewicht lebend)
Area of <i>M. longissimus dorsi</i> (cm ²)	Fläche des Rückenmuskels zwischen 8. und 9. Rippe / Schlachthälftenlänge
Lean meat proportion (%)	Muskelanteil (Regression)
Bone proportion (%)	Knochenanteil (Regression)

English	Deutsch
Tendon fat proportion (%)	Fettsehnenanteil
Meat percentage (%)	Fleischanteil
E-U-R-O-P-beefiness class (15 classes) ^a	E-U-R-O-P Handelklassen mit Unterklassen
Fat score (1-15 scores) ^a	Fettgewebeklassen mit Unterklassen
Muscularity score (1-15 scores) ^b	Muskelpunkte
Loss (%) ^c	Verluste (Schlachtkörper warm – Schlachtkörper kalt)
Storage loss (%)	Lagerverlust (Gewicht der Muskelscheibe zwischen 8. und 9. Rippe vor - nach der Reifung)
Cooking loss (%)	Grillverlust
pH 45 minutes <i>post mortem</i>	pH-Wert 45 min nach der Schlachtung
pH 24 hours <i>post mortem</i>	pH-Wert 24 h nach der Schlachtung
pH 12 days <i>post mortem</i>	pH-Wert nach der Reifung des Fleisches
Colour L	Helligkeit
Colour a	Rotton
Colour b	Gelbton
Intramuscular fat	Intramuskuläres Fett
Average shear force (N)	Durchschnittswert max. Scherkraft
Average energy of shear force (mJ)	Durchschnittswert der Scherenergie
C 14:0 (mg/100g meat)	Tetradecansäure
C 14:1 <i>cis</i> -9 (mg/100g meat)	Cis-9- Tetradecansäure
C 16:0 (mg/100g meat)	Hexadecansäure
C 16:1 <i>cis</i> -9 (mg/100g meat)	Cis-9- Hexadecansäure
C 18:0 (mg/100g meat)	Octadecansäure
English	Deutsch

C 18:1 <i>trans</i> (mg/100g meat)	Summe der trans-Octadecansäure-Isomere
C 18:1 <i>cis</i> -9 (mg/100g meat)	Cis-9-Octadecansäure
C 18:1 <i>cis</i> -11 (mg/100g meat)	Cis-11-Octadecansäure
C 18:3 <i>n</i> -3 (mg/100g meat)	All cis-Octadecatriensäure
C 22:4 (mg/100g meat)	All cis-7,10,13,16-Docosatetraensäure
CLA c9 t11 (mg/100g meat)	Konjugierte Linolsäuren
SFA (mg/100g meat) ^d	Gesättigte Fettsäuren
MUFA (mg/100g meat) ^e	Einfach ungesättigte Fettsäuren
PUFA (mg/100g meat) ^f	Mehrfach ungesättigte Fettsäuren
Ratio of C14:1 to C14:0	Verhältnis C14:1 zu C14:0
Ratio of C16:1 to C16:0	Verhältnis C16:1 zu C16:0
Ratio of C18:1 to C18:0	Verhältnis C18:1 zu C18:0

- a) EU carcass grading system
b) Scoring of live animals
c) Loss=warm carcass weight – cold carcass weight
d) The sum of saturated fatty acids (SFA) was calculated as: C10:0 + C12:0 + C14:0 + C15:0 + C16:0 + C17:0 + C18:0 + C20:0 + C22:0 + C24:0
e) The sum of monounsaturated fatty acids (MUFA) was calculated as: C14:1 + C16:1 + C18:1*trans* + C18:1*cis*9 + C18:1*cis*11 + C20:1 + C22:1 + C24:1
f) The sum of polyunsaturated fatty acids (PUFA) is the sum of n-6 and n-3 FA

7 Literature

Gilmour, A. R., Cullis, B. R., Welham, S. J., Gogel, B. J., Thompson, R., 2004: An efficient computing strategy for prediction in mixed linear models. *Computational Statistics and Data Analysis* 44: 571-586.

Groeneveld, E. (1998): PEST User's Manual. Institute for Animal Science and Animal Husbandry, Bundesforschungsanstalt für Landwirtschaft (FAL), Mariensee/Neustadt

Welham, S. J., Cullis, B. R., Gogel, B. J., Gilmour, A. R., Thompson, R., 2004: Prediction in linear mixed models. *Australian and New Zealand Journal of Statistics* 46: 325-347.

Danksagung

Nach Abschluss der Arbeit möchte ich allen Personen danken, die zum Gelingen beigetragen haben.

Herrn Prof. Dr. Norbert Reinsch danke ich für die Überlassung des Themas, die fachliche Unterstützung und intensive Betreuung; die jederzeit gewährten fachlichen Diskussionen, wertvollen Anregungen und Hinweise zur Anfertigung dieser Arbeit und dafür, dass er im richtigen Augenblick die Zeit angehalten hat.

Bei Herrn Prof. Dr. Georg Thaller bedanke ich mich für die Übernahme des Koreferates.

Ganz herzlich bedanke ich mich darüber hinaus bei Inge Räder, die mich tatkräftig unterstützt hat und viele Unklarheiten beseitigt hat.

Mein Dank gilt auch allen anderen Mitarbeitern des Forschungsbereiches Genetik und Biometrie des FBN Dummerstorf, für die jederzeit gewährte Hilfe und Unterstützung, sowie Karin Nürnberg und Ralf Pfuhl aus dem Forschungsbereich Muskelbiologie und Wachstum für ihre Hilfe.

Ganz besonders danke ich meiner Familie, meinen Freunden und meinen Kollegen für ihr Verständnis und ihre Geduld mit mir.

Lebenslauf

Name:	Nadine Neugebauer
Geburtsdatum:	19. Juni 1979
Geburtsort:	Hannover
Familienstand:	ledig
Staatsangehörigkeit:	deutsch
Schulbildung:	08.1985 – 07.1989 Grundschule Lehrte-Süd 08.1989 – 07.1991 Orientierungsstufe Lehrte-Süd 08.1991 – 07.1995 Realschule Lehrte 08.1995 – 07.1998 Gymnasium Lehrte Abschluss: Allgemeine Hochschulreife
Auslandspraktikum:	10.1998 – 06.1999 Milchviehwirtschaft im Betrieb O. Kristjansson, Selfoss, Island
Berufsausbildung:	07.1999 – 07.2000 Lehrbetrieb H. Niemeyer, Mardorf, Region Hannover 08.2000 – 07.2001 Lehrbetrieb NickersonLimagrain, Arpke, Region Hannover Abschluss: Gehilfenprüfung
Hochschulausbildung:	10.2001 – 11.2006 Diplomstudiengang Gartenbau, Leibniz Universität Hannover Abschluss: Diplom-Agraringenieurin
Berufliche Tätigkeit:	seit 12.2006 Wissenschaftliche Mitarbeiterin am Forschungsinstitut für die Biologie landwirtschaftlicher Nutztiere, Forschungsbereich Genetik und Biometrie, Dummerstorf