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# German Simmentals show significant imprinting variances for 10 traits of carcass composition

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## Introduction

Genomic imprinting is the parent-of-origin specific gene expression and is determined by epigenetic modification of genes. Important is that the actual gene sequence remains unchanged, only the gene transcription is altered. It is an epigenetic marking of genes, which are differentially expressed from the maternally and paternally inherited alleles. The different expression means that during development the parental genomes are functionally non-equivalent. A special type of imprinting, known as partial imprinting, influences allele expression in one or several of the following ways: it is possible that an allele is understated in its expression. A second possibility is the cell-type-specific allele expression. In another variation the expression is changed in different phases of life.

The first studies about imprinting in farm animals were done by De Vries et al. (1994), who analysed imprinting variance components in pigs. Engellandt and Tier (2001) found significant effects of paternal gametes in two fatness traits of finishing bulls. Essl and Voith (2002) estimated the variance of imprinting effects on dairy- and fitness-related traits of cattle. Over more QTL and imprinting analysis exist, which reported about imprinted alleles. Therefore we are interested in the relative portion of the additive genetic variance induced by imprinted genes.

## Material and Method

In two abattoirs slaughter data were collected from 66,500 German (dual-purpose) Simmental fattening bulls. In both slaughter houses, VIA-devices (VBS2000 from E + V Technology GmbH, 16515 Oranienburg, Germany) had been installed during the data-collection period from 1998 – 2004. Altogether 26 traits were analysed. The used pedigree only included the ancestors of the slaughtered animals. 365,000 ancestors were extracted from a comprehensive Simmental pedigree and it resulted in a pedigree depth of up to 17 generations.

A linear mixed model with the following effects was fitted to each trait and variance components were estimated via REML:

$$Y_{ijklmnop} = SM_i + GT_j + KN_k + b_1 x + b_2 x^2 + b_3 x^3 + s_l + d_m + y_n + m_o + e_{ijklmnop}$$

$Y_{ijklmnop}$ = traits

$SM_i$ = interaction of farm and slaughterhouse (fix) (i = 1, ..., 6044)

$GT_j$ = birth type (fix) (j = 1, 2, 3)

$KN_k$ = calve number (fix) (k = 1, 2, 3)

$b_1$ = linear regression of the slaughter age

$b_2$ = quadratic regression of the slaughter age

$b_3$ = cubic regression of the slaughter age

$s_l$ = additive genetic effect as sire (random) (l = 1, ..., 356880)

$d_m$ = additive genetic effect as dam (random) (m = 1, ..., 356880)

$y_n$ = y-chromosomal inheritance (random) (n = 1, …, 57)

$m_o$ = mitochondrial inheritance (random) (o = 1, …, 34622)

$e_{ijklmnop}$ = random residuals

With the additive genetic effect as sire and as dam, we estimated two breeding values of each animal, using the following animal model: $y = X\beta + Z_s a_s + Z_d a_d + e$

The structure of the mixed model equations are as follows:

$$\begin{bmatrix} X'W^{-1}X & X'W^{-1}Z_s & X'W^{-1}Z_d \\ Z'_s W^{-1}X & Z'_s W^{-1}Z_s + A^{-1}\alpha_1 & Z'_s W^{-1}Z_d + A^{-1}\alpha_2 \\ Z'_d W^{-1}X & Z'_d W^{-1}Z_s + A^{-1}\alpha_2 & Z'_d W^{-1}Z_d + A^{-1}\alpha_3 \end{bmatrix} \begin{bmatrix} \beta \\ a_s \\ a_d \end{bmatrix} = \begin{bmatrix} X'W^{-1}y \\ Z'_s W^{-1}y \\ Z'_d W^{-1}y \end{bmatrix}$$

y = vector of observations

X = design matrix for fixed effects

β = vector of fixed effects

A = additive genetic relationship matrix of parents only

$Z_s$, $Z_d$ = design matrix for random effects

$a_s$ = vector for breeding values with paternal expression pattern

$a_d$ = vector for breeding values with maternal expression pattern

W is a diagonal matrix with appropriate weights in order to account for the inbreeding coefficients of the parents: $w_i = \left[ \frac{1/2\sigma_s^2(1-F_{si}) + 1/2\sigma_d^2(1-F_{di}) + \sigma_e^2}{1/2\sigma_s^2 + 1/2\sigma_d^2 + \sigma_e^2} \right]^{-1}$

The variance of the random effects is:

$$Var \begin{bmatrix} a_s \\ a_d \\ e \end{bmatrix} = \begin{bmatrix} 1/2\, A\sigma_s^2 & 1/2\, A\sigma_{sd} & 0 \\ 1/2\, A\sigma_{sd} & 1/2\, A\sigma_d^2 & 0 \\ 0 & 0 & W\sigma_e^{2^*} \end{bmatrix}$$

Where $\sigma_s^2$ and $\sigma_d^2$ are the gametic variances for the genetic effects as sire and as dam, respectively, and $\sigma_{sd}$ is its covariance.

If imprinting is present, then S* is positive definite:

$$S^* = \begin{bmatrix} \sigma_s^2 & \sigma_{sd} \\ \sigma_{sd} & \sigma_d^2 \end{bmatrix}$$

and in the absence of imprinting S* is not positive definite:

$$S^* = \begin{bmatrix} \sigma_g^2 & \sigma_g^2 \\ \sigma_g^2 & \sigma_g^2 \end{bmatrix}$$

A REML likelihood ratio test with two degrees of freedom was used to test the hypothesis of imprinting against the absence of imprinting. For significantly imprinted traits the imprinting variance ($\sigma_i^2$) can be calculated as $\sigma_i^2 = \sigma_s^2 + \sigma_d^2 - 2\sigma_{sd}$ and the total additive genetic variance is $\sigma_a^2 = \sigma_s^2 + \sigma_d^2$.

Parental contributions to the imprinting variance can be calculated as $\sigma_m^2 - \sigma_{sd}$ (maternal) and $\sigma_s^2 - \sigma_{sd}$ (paternal).

## Results and Discussion

For 10 traits we found significant imprinting variances. Up to 25 % of the total additive genetic variance is due to imprinting in these traits. The parental contribution to the imprinting variance was different for traits, ranging from no maternal contribution to the imprinting variance (fat class) to nearly no paternal contribution to the imprinting variance (round).

With the model we estimate two breeding values per animal. Thus, we can calculate the differences between these values for each animal together with corresponding standard error, as summarized in the Figure.

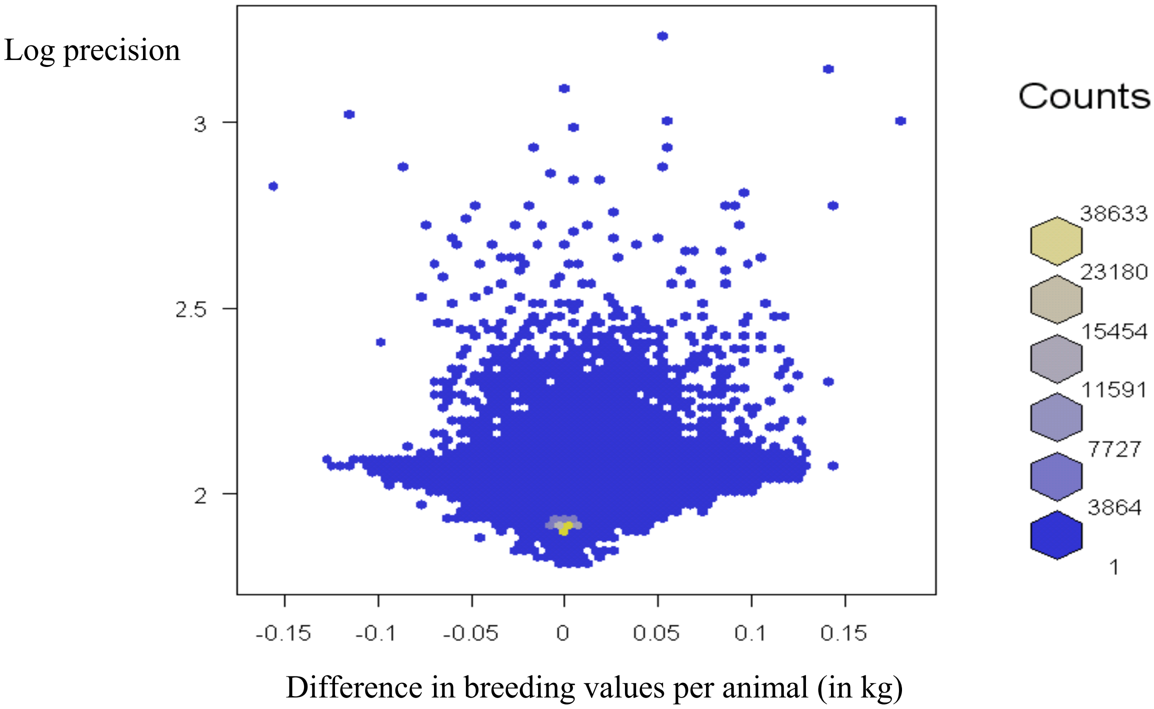

**Figure: Imprinting effects: difference in breeding values per animal (x-axis) in a clustered scatter plot for a section trait of the round. The precision (y-axis) is calculated with the reciprocal of the standard error.**

The imprinting effect for each animal is exactly this difference between the breeding values per animal. The figure shows, that the differences in the breeding values reach up to 150 g, which 35% of one genetic standard deviation. Thus the influence of imprinting genes in this trait is not small.

The questions arise, whether the separation of imprinting variance from other variances is possible. It is no problem for e.g. the y-chromosomal effect or mitochondrial effect, because they have different correlation structures. But it is impossible, when maternal effects are present. The maternal effect and the maternal imprinting variance have the common correlation structure. Therefore, the estimates of the imprinting variance can be interpreted as upper bounds and may be contaminated by a maternal variance contribution. But even in this case the model with two breeding values per animal remains applicable.

## Literature

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