The heritability of degenerative joint disease in the distal tarsal joints in Icelandic horses

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Abstract

The heritability of radiographic signs of degenerative joint disease (DJD) in the distal tarsal joints and hind limb lameness after flexion test of the tarsus was estimated in a population of Icelandic horses being used for riding. Furthermore, the genetic and phenotypic correlation between the radiographic signs and lameness was estimated. Of the 614 examined horses, aged 6–12 years, 420 belonged to progeny groups from 17 selected stallions and 194 were sired by unselected stallions. The heritability was analyzed by two statistical models based on the threshold liability concept, a non-linear sire model and a linear animal model. The heritability of radiographic signs of DJD in the distal tarsal joints was not found to be significantly different from zero. The \( h^2 \) estimate for lameness was higher and in the order of 0.4. A positive genetic and phenotypic correlation was identified between radiographic signs of DJD in the distal tarsal joints and lameness. The results indicate that the prevalence of DJD in the distal tarsal joints could be reduced in Icelandic riding horses by breeding selection based on flexion test of the tarsus followed by radiographic examination.

Keywords: Equine; Bone spavin; DJD; Icelandic horse; Heritability

1. Introduction

The Icelandic horse, also known as the Icelandic toelter horse, is the only horse breed in Iceland and the native population has been isolated from other horse breeds for about 11 centuries (Adalsteinsson, 1981). Based on different traits of the riding performance and the conformation, organized horse breeding has been practiced in Iceland since the beginning of this century (Hugason, 1994). Since 1986, the Best Linear Unbiased Prediction (BLUP) method has been used to evaluate the breeding...
values in an index, where all pedigree information is included (Arnason, 1984). A list of horses with the highest indexes is published annually. The registration of horses in Iceland is voluntary and includes mostly breeding horses. Consequently, the register does not represent the age, gender or genetic background of the entire population. Examination for diseases that are suspected to be inherited is, currently, not included in the breeding program.

Degenerative joint disease (DJD) of the distal tarsal joints, sometimes called bone spavin, (Barneveld, 1983; Butler et al., 1993) has been reported to be a common condition in Icelandic horses (Eksell et al., 1998). Radiographic examination is considered essential for the diagnosis (Stashak, 1987; Butler et al., 1993) but palpation of the distal tarsus and hind limb motion evaluation before and after flexion test of the tarsus have also been shown to be of importance (Axelsson et al., 1998).

The aetiology of DJD in the distal tarsal joints is not clear but has been considered to be multifactorial (Wyn-Jones, 1988). It has been related to poor conformation and horses with sickle hocks and cow hocks have been reported to be predisposed for developing bone spavin indicating an inheritable character (Rooney, 1968; Gabel, 1980; Barneveld, 1983; Stashak, 1987). In German riding horses, the heritability of bone spavin, based on a radiographic diagnosis, has been estimated to be low, $h^2 = 0.02–0.04$ on an observed scale (Winter et al., 1996).

The purpose of the present study was to estimate the heritability of radiographic signs of DJD in the distal tarsal joints and hind limb lameness in a population of Icelandic horses being used for riding in Iceland.

2. Materials and methods

2.1. Horses

By an official advertisement, horseowners in the south, west and north of Iceland were invited to submit horses, aged 6–12 years and being used for riding, to the study. Offspring from 17 selected sires that were in use for breeding in the years 1982–1989 and having a breeding index (BLUP) higher than 105, based on more than 50 offspring (Hugason, 1997) were requested. Close relationship between the sires was avoided. Additionally, horses of the same age, sired by other, unselected stallions were included in the material. Information on the pedigrees was collected from the owners and the official register of horses in Iceland and was traced as far back as possible. The paternity of the offspring from the selected stallions was tested by blood typing.

The material consisted of 614 horses, of which 420 were in the progeny groups (half-sibs) and 144 were sired by 83 other identified stallions. The paternity of 50 horses was unknown. The mean age was 7.9 years and the distribution of gender was 24 (3.9%) stallions, 403 (65.6%) geldings and 187 (30.5%) mares.

2.2. Radiographic examination

The radiographic examination included latero-5°-proximal-mediiodistal (L5Pr-MDi), dorso-35°-lateral-plantaromedial oblique (D35L-PIMO) and plantaro-45°-lateral-dorsomedial oblique (Pl45L-DMO) views of each tarsus. The diagnostic criteria for radiographic signs of DJD in the distal tarsal joints were: rarefaction of subchondral bone, narrowing or collapse of intertarsal joint spaces (Eksell, 1998a). The presence of the radiographic signs was classified as all-or-none trait, based on the consensus of two radiologists. Horses with equivocal radiographic findings, were classified as radiographically normal.

2.3. Clinical examination

Hind limb lameness was first evaluated while trotting the horse by hand on a firm surface 25–30 m straight away from the examiner and back. After which a flexion test was performed by flexion of each tarsus for 1 min before repeating the above motion evaluation. Lameness after flexion test was classified as all-or-none trait. In doubtful cases, horses were classified as free from lameness. The clinical examination was performed, and the results were recorded after the consensus of the same two clinicians throughout the study.

2.4. Statistical methods

Four dichotomous variables were formed for the genetic analyses: radiographic signs of DJD in the
distal tarsal joints (RS), lameness, RS and lameness, and, RS and/or lameness.

The statistical models for the analysis of the heritability of the binary variables were based on the threshold liability concept (Falconer, 1989). The material was analyzed with two conceptually different models. The following non-linear, sire model was fitted to the progeny data:

\[ Y_{ijkl} = \eta + g_i + m_j + s_k + w_{ijkl} \]

where \( Y_{ijkl} \) is the \( l \)-th observation (one or two) within the \( i \)-th age class (\( i = 1, ..., 7; 6-12 \) years) and within the \( j \)-th sex class on the \( k \)-th sire; \( \eta \) is the localization (fixed effect) of the threshold on the scale of the standard normal distribution; \( g_i \) is the fixed effect of the \( i \)-th age class (\( i = 1, ..., 7; 6-12 \) years); \( m_j \) is the fixed effect of the \( j \)-th sex class (male, female); \( s_k \) is the random effect of the \( k \)-th sire (\( k = 1, ..., 17 \)) = NID(0, \( \sigma_s^2 \)) and \( w_{ijkl} \) is the random residual term = NID(0, \( \sigma_w^2 \)).

The heritability is defined as the ratio of additive genetic variance to phenotypic variance, commonly expressed as: \( h^2 = \frac{\sigma^2_g}{\sigma^2_p} \). In the model described above, \( \sigma^2_g \) is assumed to equal \( 1/4 \sigma^2_x \) and \( \sigma^2_w = \sigma^2_s + 3/4 \sigma^2_x \). The heritability can therefore be estimated as \( h^2 = \frac{4 \sigma^2_g}{\sigma^2_s + \sigma^2_w} \). The non-linear threshold model equations of Gianola and Foulley (1983) were solved by the CMMAT computer programs of Misztal et al. (1989). In threshold models only the relative size of the variance components are of interest and the \( \sigma^2_v \) components may for convenience be set arbitrarily to one.

The standard errors of the estimated threshold and heritabilities were obtained by simulating 1000 samples of data with 17 progeny groups, each composed of 25 members. The binary variables, \( Y \), were formed according to the truncated value of a normally distributed underlying variable, \( x \), in relation to the estimated thresholds. The underlying variables were generated as \( x = s + w = zh/2 + z' \sqrt{(1 - 1/4h^2)} \), where \( z \) and \( z' \) are independent and normally distributed deviates with zero means and unit variance and are obtained by the Box–Muller method (Press et al., 1989). The variation in the estimates provided by the CMMAT computer program, when run on 1000 samples of data generated from the true parameters, form a reasonable basis for statistical inferences about the sampling variance of the estimates.

Alternatively to the non-linear threshold model, an animal model, using the complete additive genetic relationship between all animals was fitted to the data. Tracing the pedigrees of the 614 horses in the material as far back as they were registered resulted in 1261 horses available for the analysis. The male paths were 2.8 generations on average with maximum tracing of six generations, while the corresponding figures on the female side were 0.5 and 3, respectively. The unrecorded individuals which did not have pedigree ties with more than one other horse provided no information to the analysis and were pruned. This left 1055 horses which were informative for the analysis, of which 122 were identified sires and 92 were identified dams with progeny records. Corresponding number of grand-sires and grand-dams with progeny records were 42 and 29, respectively.

The following linear model was assumed:

\[ Y_{ijkl} = \sigma + g_i + m_j + a_k + e_{ijkl} \]

where \( Y_{ijkl} \) is the \( l \)-th observation (zero or one) on the \( k \)-th animal within the \( i \)-th age class (\( i = 1, ..., 7; 6-12 \) years) and within the \( j \)-th sex class; \( \sigma \) is the overall mean; \( g_i \) is the fixed effect of the \( i \)-th age class (\( i = 1, ..., 7; 6-12 \) years); \( m_j \) is the fixed effect of the \( j \)-th sex class (male, female); \( a_k \) is the random effect of the breeding value of the \( k \)-th animal (\( k = 1, ..., 614 \)) = N(0, \( A \sigma^2_a \)) where \( A \) is the matrix of numeric genetic relationship between 1055 animals and \( e_{ijkl} \) is the random residual term = NID(0, \( \sigma^2_e \)).

The DFREML programs of Meyer were used for estimating the heritabilities on the observable scale by locating the maximum of the restricted maximum likelihood function by a derivative free approach (Meyer, 1993).

The heritability estimates were transformed to the underlying scale by the transformation formula of Robertson: \( h^2_{\text{unf}} = h^2_{\text{obs}} p(1-p)/z^2 \), where \( z \) is the ordinate of the standardized normal distribution at the truncation point corresponding to the frequency affected, \( p \) (Dempster and Lerner, 1950).

A series of bivariate analyses was performed by the DFREML programs for studying the association between the binary variables RS, lameness and RS and lameness. A bivariate version of the animal model described above was assumed. The genetic
(r_A) and phenotypic (r_p) correlations were estimated by the standard procedure (Falconer, 1989).

3. Results

The prevalence of radiographic signs of DJD in the distal tarsal joints (RS) was 30.3%. Hind limb lameness after flexion test (lameness) was identified in 32.4% of the horses. The combination RS and lameness was found in 16.4% while RS and/or lameness were found in 46.3%. Horses with the radiographic signs had four-times higher odds of being lame than horses without them (P<0.001).

Results from the heritability analysis according to the non-linear threshold sire model are summarized in Table 1. Solutions for the fixed effects were obtained by constraining the first level of all fixed effects exceeding the threshold to zero. The estimated location of the thresholds for six-year-old males corresponds to the frequency values, p = 0.23 for RS, 0.28 for lameness, 0.17 for RS and lameness, and, 0.34 for RS and/or lameness.

The results from the analysis based on the linear animal model are shown in Table 2. A bivariate analysis of RS and lameness, according to an animal model, yielded the following estimates of genetic and phenotypic correlations: r_A = 0.70 and r_p = 0.30. Corresponding estimates between RS and RS and lameness were obtained: r_A = 0.93 and r_p = 0.66.

4. Discussion

The heritability for lameness was, in the two models, estimated to be 0.4, which is considerably higher than for RS, where the estimated h^2 was 0.1

<table>
<thead>
<tr>
<th>Variables</th>
<th>RS</th>
<th>Lameness</th>
<th>Lameness and RS</th>
<th>Lameness and/or RS</th>
</tr>
</thead>
<tbody>
<tr>
<td>Threshold</td>
<td>0.737</td>
<td>0.598</td>
<td>0.965</td>
<td>0.406</td>
</tr>
<tr>
<td>S.E. threshold</td>
<td>0.080</td>
<td>0.100</td>
<td>0.097</td>
<td>0.095</td>
</tr>
<tr>
<td>Corresponding p</td>
<td>0.230</td>
<td>0.275</td>
<td>0.167</td>
<td>0.342</td>
</tr>
<tr>
<td>Age (years)</td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>6</td>
<td>0</td>
<td>0</td>
<td>0</td>
<td>0</td>
</tr>
<tr>
<td>7</td>
<td>-0.065</td>
<td>-0.227</td>
<td>0.258</td>
<td>-0.394</td>
</tr>
<tr>
<td>8</td>
<td>-0.251</td>
<td>0.121</td>
<td>0.022</td>
<td>-0.139</td>
</tr>
<tr>
<td>9</td>
<td>-0.571</td>
<td>0.049</td>
<td>-0.074</td>
<td>-0.439</td>
</tr>
<tr>
<td>10</td>
<td>-0.432</td>
<td>-0.033</td>
<td>0.086</td>
<td>-0.460</td>
</tr>
<tr>
<td>11</td>
<td>-0.724</td>
<td>0.017</td>
<td>-0.197</td>
<td>-0.489</td>
</tr>
<tr>
<td>12</td>
<td>-0.864</td>
<td>-0.578</td>
<td>-0.835</td>
<td>-0.682</td>
</tr>
<tr>
<td>Sex</td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Male</td>
<td>0</td>
<td>0</td>
<td>0</td>
<td>0</td>
</tr>
<tr>
<td>Female</td>
<td>0.176</td>
<td>-0.174</td>
<td>0.193</td>
<td>-0.126</td>
</tr>
<tr>
<td>Sire variance α^2</td>
<td>0.023</td>
<td>0.107</td>
<td>0.076</td>
<td>0.076</td>
</tr>
<tr>
<td>Heritability</td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>h^2</td>
<td>0.088</td>
<td>0.385</td>
<td>0.284</td>
<td>0.282</td>
</tr>
<tr>
<td>S.E. h^2</td>
<td>0.109</td>
<td>0.204</td>
<td>0.187</td>
<td>0.176</td>
</tr>
</tbody>
</table>

* Solutions to the fixed effects of the threshold, age and sex, and estimates of sire variances and of heritability.
* RS = Radiographic signs of degenerative joint disease in the distal tarsal joints; lameness = hind limb lameness after flexion test of the tarsus.
Table 2
Results from the linear model analysis

<table>
<thead>
<tr>
<th>Variables</th>
<th>Lameness</th>
<th>Lameness and RS</th>
<th>Lameness and/or RS</th>
</tr>
</thead>
<tbody>
<tr>
<td>RS</td>
<td>0.221 (0.040)</td>
<td>0.266 (0.045)</td>
<td>0.138 (0.033)</td>
</tr>
<tr>
<td>Age (years)</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>6</td>
<td>0.051 (0.052)</td>
<td>0.078 (0.053)</td>
<td>-0.008 (0.042)</td>
</tr>
<tr>
<td>7</td>
<td>0.115 (0.055)</td>
<td>-0.003 (0.056)</td>
<td>0.029 (0.044)</td>
</tr>
<tr>
<td>9</td>
<td>0.190 (0.066)</td>
<td>0.073 (0.068)</td>
<td>0.049 (0.053)</td>
</tr>
<tr>
<td>10</td>
<td>0.188 (0.067)</td>
<td>0.030 (0.069)</td>
<td>0.036 (0.054)</td>
</tr>
<tr>
<td>11</td>
<td>0.277 (0.080)</td>
<td>0.059 (0.082)</td>
<td>0.118 (0.065)</td>
</tr>
<tr>
<td>12</td>
<td>0.347 (0.098)</td>
<td>0.207 (0.100)</td>
<td>0.291 (0.079)</td>
</tr>
<tr>
<td>Sex</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Male</td>
<td>0 -0.071 (0.40)</td>
<td>0.033 (0.041)</td>
<td>-0.033 (0.032)</td>
</tr>
<tr>
<td>Female</td>
<td>0</td>
<td>0</td>
<td>0</td>
</tr>
</tbody>
</table>

Heritability and variation

| $h_{20}^2$ | 0.057 | 0.246 | 0.098 | 0.156 |
| $h_{sv}^2$ | 0.099 (0.063) | 0.418 (0.129) | 0.222 (0.084) | 0.246 (0.094) |
| $\sigma^2_e$ | 0.449 | 0.466 | 0.363 | 0.493 |

* Solutions to the fixed effects of age and sex (±S.E.). Estimates of phenotypic variance and heritability on the observed and the underlying scale.

† RS = Radiographic signs of degenerative joint disease in the distal tarsal joints; lameness = hind limb lameness after flexion test of the tarsus.

and not significantly different from zero. The radiographic diagnosis is considered to be specific for DJD in the distal tarsal joints and once present, the radiographic signs persist with bone remodelling over time. The linear increase in the prevalence of RS in the age range of 6–12 years suggest that the disposition is present in the majority of Icelandic horses. If the linear increase is continuous, all the horses will show RS if they get old enough. The linear effect of age on the radiographic signs might be partly genetic. Inclusion of the fixed effects of age in the linear model might therefore cause a serious reduction in the estimated sire variance component and consequently result in underestimation of the heritability of RS. Exclusion of age from the model would on the other hand cause overestimation of the heritability, since the progeny groups are unevenly distributed across age classes. The RS variable could be analyzed by survival analysis methodology (Klein and Moeschberger, 1997) since the variables in this study represent censored data, where the lower bound of the presence of RS is registered, i.e., whether they have developed RS at the time of examination. However, the $h^2$ estimates in the present study corresponds well to figures reported for bone spavin (radiographic diagnosis) in 3–8 years (mean age = 3.9 years) German riding horses (Winter et al., 1996).

Hind limb lameness after flexion test of the tarsus is regarded as a less specific method for the diagnosis of DJD in the distal tarsal joints than radiography (Stashak, 1987). The presence of lameness connected with the disease has also been shown to vary (Stashak 1987; Butler et al., 1993). The heritability estimates for lameness need therefore to be interpreted with care. As no other specific conditions are known to affect the tarsal or stifle joints in the Icelandic horse, the $h^2$ estimates for lameness might however be a good indicator of the heritability of DJD in the distal tarsal joints. The heritability of lameness has been estimated for Standardbred trotters in Norway and found to be 0.33 on an underlying scale (Dolvik and Gaustad, 1996). Although the lameness was connected to other conditions than
DJD of the distal tarsal joints, the result is similar to what was found for lameness in the present study. The results of these two studies indicate that the disposition for lameness may be inherited more than the correlated trait (RS).

There was no linear trend in the effect of age on the frequency of lameness. It may be that horses can be lame in the early stage of the disease and the lameness can improve over time. It may also indicate that the presence of lameness is not dependent on the same factors as developing the radiographic signs.

The estimated heritability for the two combined variables (RS and lameness, and, RS and/or lameness) was found to be similar and in the order of 0.25. They were significantly different from zero according to the animal model.

The results from the two statistical models show good agreement for the heritability estimates. The animal model, utilizing the additional information about relationship ties across the population, did not alter the results obtained by the sire model. It indicates that the dams represent a random sample. The size of the data set is however limited for heritability estimates, which are susceptible to rather large sampling errors. The standard errors for the estimates are higher in the sire model as expected, since the animal model uses more observations and accounts for the entire pedigree structure in the data.

The phenotypic correlation between RS and lameness was medium high (0.3) in the present study. The estimated genetic correlation is higher (0.7) but is a subject to a large sampling error. It corresponds to an environmental correlation in the order of 0.2 and together these positive correlations supports the relationship between RS and lameness.

A positive lameness examination followed by radiographic examination demonstrating DJD in the distal tarsal joints, could be used as excluding criterion for breeding horses. The heritability for the combined variable RS and lameness is estimated to be medium and the genetic correlation of the combined variable and RS is high. For example, stallions, with RS and lameness at the age of six years, could be excluded from the breeding. The generation interval of the Icelandic horse is approximately nine years in males and 11 years in females (Hugason et al., 1985). Assuming that the estimates in Table 1 are the correct parameters and that stallions do not reproduce before before the age of six, this type of selection would move the threshold value for six-year-old males from the current value of 0.965 to 1.011 in ten years. This means a reduction from the present frequency of 0.167 to 0.156 in ten years. Selection based on progeny testing or a combined index could possibly lead to somewhat greater genetic response. According to the higher estimated heritability of lameness, a higher response in RS and even in lameness and RS might be expected from an indirect selection for lameness alone. However, since the inclusion of age effects in the linear model might have caused underestimation of the genetic variance in RS, the combined criterium should, of caution, be recommended in practice until further knowledge will be available.

Cost–benefit analysis needs to be performed for different selection programs, weighing the expected gain in relation to the cost of the disease, the diagnostic tests and the lowered genetic gain that could be expected for other important traits.

More sensitive diagnostic methods such as scintigraphy (Lamb and Koblik, 1988) and in the future methods based on genetic markers could make the selection more favorable.

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References


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