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Prevalence and genetic parameters for cryptorchidism in Swedish-born Icelandic horses

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ABSTRACT

Cryptorchidism is a defect in males where one or both testes fail to descend normally into the scrotum. If both testes are retained, the stallion will be infertile. There are few studies of the prevalence of the condition in horses based on field data, and heritability estimates for equine cryptorchidism are lacking. The objective of this study was to provide data on prevalence and heritability of cryptorchidism in Icelandic horses born in Sweden. A questionnaire was sent to breeders, asking whether or not stallions born at the farm had both testes present in the scrotum at the age of 1, 6 and 12 months. Fixed effects were analysed using logistic regression and genetic parameters were estimated using linear animal models. Information about 595 yearling stallions born 1997–2011 was used in the analyses. Close to 9% of the yearlings did not have both testes in the scrotum. Probability of cryptorchidism in yearlings was significantly influenced by farm and time period of birth. Heritability estimates for cryptorchidism ranged from 0.12 to 0.32 (SE 0.08–0.12) on the observable scale, and from 0.35 to 0.96 (SE 0.24–0.40) when transformed to the underlying continuous scale. The results support that equine cryptorchidism is heritable. Further studies with more individuals included would be needed to confirm this.

1. Introduction

Cryptorchidism is a congenital defect in males where one (unilateral) or both (bilateral) of the testes fail to descend normally into the scrotum. The retained testis can be located in the abdominal cavity, the inguinal canal or subcutaneously at the inguinal ring (Amann and Veeramachaneni, 2007). The defect occurs in several species, including horse, cattle, sheep, pig, cat, dog, rabbit and human. The prevalence in horses has been suggested to be 2–8% in a review by Amann and Veeramachaneni (2007). This figure seem however to be based on few, older, studies mainly focused on type of defects rather than on prevalence.

A more complicated and costly surgery is needed for castration of cryptorchid stallions. Bilateral cryptorchidism cause sterility in stallions (Mueller and Parks, 1999). The condition is suggested to be influenced by genetic, epigenetic and environmental factors, including endogenous hormonal state of the pregnant mare (Hayes, 1986; Leipold et al., 1986; Amann and Veeramachaneni, 2006). In humans, a recently developed cryptorchidism gene atlas included 217 candidate loci of interest for further studies (Cannistraci et al., 2013), but the genetic causes of the defect are still unclear. In dogs and pigs, moderate to high heritabilities (0.23–0.75) of the liability of cryptorchidism have been estimated (Mikami and Fredeen, 1979; Nielen et al., 2001; Dolf et al., 2010). To our knowledge, there are no published heritability estimates of cryptorchidism in horses, and the genetic background of different forms of the condition remains to be clarified (Stout, 2014).

In a large study of data from veterinary university teaching hospitals, ponies as a group, and some specific breeds, were overrepresented for cryptorchidism (Hayes, 1986). A recent study of veterinary medicine hospital data on castrations during 2000–2009 in Sweden indicated that cryptorchidism was relatively common in the Icelandic horse breed. In that study 43% of the castrated Icelandic horse stallions were cryptorchid (Engsäll, 2011). However, most Icelandic horses are castrated in their home environment, and complicated cases are probably clearly overrepresented among castrations at veterinary hospitals. In an Icelandic study on castration reports (Tryggvadóttir, 1991), the prevalence of cryptorchidism in young stallions was estimated to

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4.5%.

The aim of this study was to estimate the prevalence of cryptorchidism in Icelandic horses born in Sweden, based on field data, and to estimate genetic parameters for the defect.

2. Materials and methods

2.1. Data collection

A questionnaire was sent by mail to 80 Icelandic horse breeders that were chosen because they had registered at least 5 colts since 1990. Information from the international Icelandic horse database WorldFengur (Lorange, 2011) was used to identify all stallions born at the different farms during 1990–2011. The breeders were asked to tick boxes with 'yes', 'no' or 'unknown' to answer whether or not each of their male foals had both his testes in the scrotum at the age of 1, 6 and 12 months. The status at 1 month of age was chosen as normal testis descent take place before two weeks of age according to Bergin et al. (1970). We were also interested in the status at later ages as the Icelandic horse is said to be late-maturing. We chose the age of 12 months as yearlings often remain in their breeder's farm and are rarely castrated. The status at 6 months was included as an intermediate measure.

If the horse did not have both testes in the scrotum at 12 months, the breeder was asked to fill in a second part of the questionnaire. In this second part we asked if the stallion was castrated and if so, about time and method of castration, which testis (or whether both testes) were retained and if the retained testis was located in the abdomen or in the inguinal canal. We also asked if there were other abnormalities found, such as torsion or deviation in size or texture of the testes. Some breeders spontaneously provided additional information about their horses in this part of the questionnaire.

Out of 80 questionnaires sent, four were returned because of incorrect addresses. Among the remaining, the percentage of answer was 57%. We also informed about the study in the main magazine of the Icelandic horse organisation and on their homepage, and two additional breeders sent in information. In total 45 breeders that had bred 864 male foals answered the first part of the questionnaire. The more detailed second part of the questionnaire was answered for 47 horses with retained testes at 12 months of age.

2.2. Data description

Only five of the 47 yearlings for which the second part of the questionnaire was available were bilateral cryptorchid. Some breeders did not know the location of the retained testis (right or left; inguinal or abdominal). Among those with information provided, the left testis was retained in 12 horses, and the right in 9 horses. The testis was located in the inguinal canal in 24 horses and in the abdomen for 5 horses. In all cases of abdominal cryptorchidism, the right testis was found in the abdomen. In one of these horses, castrated in a veterinary medicine hospital at the age of four, a tumour had developed in the retained testis. In 5 of the 47 horses, the inguinal testis/testes descended between the ages of 2–3.5 years according to the breeders' replies.

Of the 47 horses, 39 were castrated, most commonly at 2–4 years of age. The most common remark at castration was a larger than normal size difference between the testes.

2.3. Data editing

Three foals that died when young were excluded from the study. For three other horses the breeders reported that both testes had been in the scrotum at younger ages, but that one testis was later retracted so that the horses could not be castrated using standard procedures. These horses were also removed from further analyses, leaving 858 horses in the data. Some of the breeders did not fill in the status of the horse at all ages (1, 6 and 12 months) in the main questionnaire. When the horse did not have both testes in the scrotum at 12 months of age, and information was missing about the status at 1 and 6 months, we assumed that the horse did not have both testes in the scrotum at younger ages. In case the testes were descended already at 1 month but no further information was given, we assumed that the testes remained in the scrotum at older ages. When both testes were in the scrotum at the age of 12 months but no information was given about the status at younger ages, we assumed the status to be "unknown" at 1 and 6 months of age.

Several breeders commented that they did not examine the testes in young foals, and some did not examine their stallions' testes until it was time for castration at 1–2, 1–3, or in one case up to 5 years of age. Those breeders provided information on stallions with retained testes at the time of castration. For cases where the information was very difficult to interpret, we contacted the breeders for clarifications.

Few of the breeders active today were breeding Icelandic horses in the early 1990s, and there were few observations per year in the earlier part of the data. Only one horse in the data born before 1997 was reported not to have both testes in the scrotum, and we decided to analyse data on horses born during the last 15 years in the data (1997–2011) only. This had very small effects on the estimated prevalences. The number of stallions born 1997–2011 with both testes in the scrotum, with at least one retained testis, or with missing information is shown for the different ages in Table 1.

2.4. Trait definitions

We defined different traits at 1 (CRYPT1), 6 (CRYPT6) and 12 (CRYPT12) months of age. The horses were scored as unaffected (0) if both testes were in the scrotum, and as cryptorchid (1) if at least one testis was retained at that age. In addition, we defined a trait, CRYPT12+, where we also included information from breeders about the testes' descent of horses up to 5 years of age. For this trait we excluded horses born 2011 so that all horses in the study would be at least 2 years of age at the time of data collection. For each of the traits we defined, we excluded horses with unknown status before analysing the data further.

The number of horses with information on any of the traits CRYPT1, CRYPT6, CRYPT12 and CRYPT12+ per breeding farm varied between 1 and 65. Some sires had offspring in one farm only, but all farms with more than one horse in the data had used more

Table 1
Distribution of horses according to reported testis descent status, with absolute and relative figures.

Age	No. of horses			Frequency of cryptorchid	
	Unaffected	Cryptorchid	Unknown	Of all with known status	Of all including unknown status
1 month	65	87	601	0.57	0.12
6 months	221	83	449	0.27	0.11
12 months	539	56	158	0.09	0.07
≥ 12 months[a]	601	50	102	0.08	0.07

[a] Including also horses that were older than 12 months when the breeder examined them.