GENETIC ASPECTS OF DISEASES AND LESIONS IN HORSES

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SUMMARY

A review of culling reasons in different Swedish horse populations and four case studies proving genetic variation in different diseases are presented. Locomotor problems are shown to be the most common cause of culling adult horses. However, many problems also occur at a young age. Stillbirths are shown to vary among and within breeds. Shetland ponies have a higher stillbirth rate than other breeds, especially in the "Mini" type. An abnormality of the extremity bones due to a recessive gene is also found within this breed. Osteochondrosis appears in about 11% of Standardbred trotter yearlings and the heritability is estimated at 0.1 on the visible and 0.2-0.3 on the underlying scale. The impact of osteochondrosis on performance is significant only when multiple lesions or clinical signs of synovial effusion are present. A fourth case, roaring, demonstrates the needs for progeny testing as this defect may be either of clearly genetic or of non-genetic origin

It is concluded that systematic recording and analyses of diagnosed diseases in horses are essential to monitor the health status of horse populations and for application of preventive measures, including selection, to improve the health of horses.
Keywords: horse, genetic defects, stillbirths, osteochondrosis, roaring, culling

INTRODUCTION

Irrespective of the use of horses, either it is for sport purposes in different disciplines or for forestry or ranching work, a sound constitution is a prerequisite for successful performance. When the horse has been trained to perform as a race horse, with a normal peak at 3-5 years of age, or as a show jumper or dressage horse, with top performance at 10-15 years of age, large investments have been made to reach these levels. It is therefore quite essential that the horse has such constitutional qualities that it can stay sound and utilize its potential to perform. That assumes good rearing and management of the horses at all stages of life, but also genetically sound horses to start with. Animal welfare aspects further supports the importance of healthy horses.

Failures to develop a high performance level are of many different origins, the most common being lack of talent. However, losses for various reasons and impaired performance due to defects, contagious diseases, inappropriate rearing, feeding, training and management broadly reflect different areas where problems may occur.

Although environmental factors dominate also genetic factors contribute to the health status of the horse. As the horse has been a close companion of man since its domestication many observations about characteristics of the horse have been noted and empirical conclusions have been drawn about the inheritance of certain characteristics. One striking example is roaring (*laryngeal hemiplegia*), a defect mentioned for its hereditary nature in the literature already in the 16th century.

The importance of considering soundness in horse breeding compared to selection practices in other species was discussed by Glodek (1979). While specialized selection for single traits may well be the case in swine and poultry for muscular growth, which has proven to cause damages to the skeleton, similar side-effects are unlikely to occur among horses, where functional traits are of primary interest. However, when selection for breeding is practised among young horses, not yet exposed to tests of physical strength, problems may occur also in horse populations. A weak point is also, especially in the racing sport, that mares with poor constitution quit racing and are used for breeding by less professional breeders.

In defining the breeding objectives for different horse breeds performance and constitutional traits are equally important. While performance traits are rather straight-forwardly defined and analysed, deficiencies in constitution may have many causes. These need to be quantified as regards incidence and relative importance. Obviously reasonably accurate methods for diagnosis and the mode of their inheritance are also components essential to consider in order to establish effective selection policies.

Literature contains considerable information on defects in the horse. Robinson (1989) lists 47 genetic defects. However, in many cases the proofs of inheritance are weak or could not be quantified due to limited information.

In the following different types of studies on impaired functions of the horse are illustrated with results from Swedish investigations. They comprise stillbirths in general and in Shetland ponies in particular, deformed extremity bones in the same breed, osteochondrosis in Standardbred trotters and a case study on roaring in Warmblood riding horses. The purpose is to illustrate the diversity and importance of problems and mode of inheritance pointing at possible ways to consider soundness in horse breeding. To start with, a review is given on the most common reasons of death or culling of horses.

CAUSES OF DEATH AND CULLING OF HORSES

A number of studies have pointed at deficiencies of the locomotor, respiratory, cardiovascular and alimentary systems to be the main causes of culling or death (e.g. Baker and Ellis, 1981; Clausen et al., 1990; Lindner and Offeney, 1992). However, results are quite dependent on the type of data being available. Most studies are based on insurance statistics or results from post-mortem examinations at animal clinics. One must remember that insurance statistics only consider cases that an insurance for the time being covers, while hospital data only refers to cases that were treated and examined at clinics.

Table 1. Horses culled (%) by 1990 for different reasons (Wallin et al. 1997)

Culling reason	Performance tested Warmblood horses born 1968-82 (n=503/1847)	Riding school horses born 1970-75 (n=208/344)	Coldblood horses born 1970-75 (n=97/204)
Locomotor problems	60.1	60.6	21.6
Accidents	9.1	2.4	3.1
Respiratory diseases	8.9	8.2	2.1
Digestive diseases	5.6	4.8	6.2
Diseases of nervous system	5.4	2.9	22.7
Circulatory diseases	2.6	2.4	0.0
Other diseases	4.1	4.8	17.5
Unknown	4.2	13.9	26.8

In a recent Swedish study three different kinds of data were used (Wallin et al., 1997). The first set was a retrospective study based on a questionnaire to owners of horses that at 4 years of age had participated in a performance test (Riding Horse Quality Test), comprising 503 dead out of 1847 participating horses. Results in Table 1 show the dominant role of diseases of the musculoskeletal system. The second set consisted of 344 horses mainly used at riding schools, of which 208 were dead. Again, more than half of these were culled due to locomotor problems. The third group consisted of 204 horses, mainly of the Swedish Ardenner and North-Swedish breeds. Of these 111 had died. Here the culling reasons were more spread, but less well documented. The most common identified reason was, though, deficiencies in temperament.

Although this study was based on censored data the general conclusion was that the vast majority of problems that cause impaired functions of the horse, and eventually its culling, belong to the locomotor system. Problems were largely diagnosed as degenerative joint disease. Navicular disease was reported to be the most common single diagnosis, 6.8 and 9.1%, respectively, among the Warmblood horses. It must, however, be noted that the baseline data for the studies presented were collected on generally healthy horses at 3-4 years of age. Consequently reasons for losses of foals and young, still growing horses, were not covered.

CASE STUDIES

Stillbirths. In all animal production systems liveability at birth is important. In horse breeding the relative cost to produce a foal is quite high compared to most other species. Thus, it is important that the pregnant mare also gives birth to a healthy foal. The Swedish Horse Board, Jockey Club and Trotting Association annually check the outcome of all coverings by reports from the breeders. Stillbirth rates in 14 breeds were summarized for the years 1985-91 by Gusén (1993). Stillbirths included foals dying in the first 9 days. The figures varied between 1.1 and 4.8% and were rather similar to, or lower than, what is found in cattle. However, the completeness of reporting may be questioned according to a study by Shaub et al. (1996) on Shetland ponies. This breed surprisingly showed the highest stillbirth rate among the pony breeds.

In the latter study a questionnaire was sent to the owners of all Shetland mares covered in 1993 for a follow-up of the foaling results in 1994. Response was received for 1507 mares (83%). The ponies were classified according to wither height as "Mini" (≤86 cm) or "Normal" (>86 cm). The overall stillbirth rate was 8.3%, with 11.8% dead at first and 7.3% at later parities. When mating "Normal" horses with each other the stillbirth rate was 6.6%, while a corresponding rate of 14.0% was found among the "Mini" horses. Two important conclusions could be drawn. Apparently the stillbirth rate was much higher in 1994 than had been reported earlier. Whether it really was higher or the reporting procedure was more accurate still remains to be investigated. Whatever the reason the loss rate among Shetland ponies at birth is of such a magnitude that it should be further analysed as regards a more accurate reporting method to be adopted and for a potential genetic variation among stallions to consider in selection. As regards the "Mini" horses obviously these carry less viable foals. The data set of 93 foalings so far analysed was too small to draw any definite conclusions about the causes. However, the generally high death rate is alarming and questions the ethics of breeding extreme pleasure horses at the cost of an increased stillbirth rate.

Abnormality of the appendicular skeleton in Shetland ponies. Cases of Shetland ponies with fully developed *ulna* and *fibula* has been noted since long in the UK and The Netherlands (Speed, 1958; Hermans, 1970). Hermans (1970) proved through test matings that the malformation most likely was caused by an autosomal recessive gene. Mating affected animals with each other resulted in only affected foals (n=9), while the same dams (n=5) all gave normal foals when mated to a normal stallion. This malformation becomes more serious as a result of growth, and it is usually necessary to cull affected animals as they develop abnormal movements and lameness due to the short legs and incorrect angles between the various bones. Several cases of foals with fully developed *ulna* and *fibula* have in recent years been diagnosed in Shetland ponies also in Sweden (Thyreen, 1994).

In the Swedish study 8 cases were identified with a full pedigree. Five of these had the same sire. Two had the same dam and the grand-dam also had an affected progeny. The Swedish data fully supports the theory of a simple, recessive, autosomal inheritance. The study indicated a gene frequency of 18% in the population of Swedish Shetland ponies.

The Swedish animal breeding laws seriously consider animal welfare aspects. Thus, animals which transmit genetic defects that affects the well-being of animals must be rejected from breeding. As a consequence Shetland stallions that are found to produce foals with fully developed *ulna* and *fibula* are culled from breeding. However, the identity of an affected foal must be verified by blood-typing and the malformation also needs to be diagnosed radiographically before any breeding decisions are made.

Osteochondrosis in standardbred trotters. One of the most common skeletal disorders developing in growing horses is osteochondrosis. Nutritional as well as genetic and bio-mechanical factors are suggested to contribute to the development of this disorder.

A number of studies in different species have proved that hereditary factors contribute to the prevalence of osteochondrosis (Lundeheim, 1987; Olsson, 1976). The lesions studied may involve different joints or sites and therefore are not necessarily governed by the same genes. A

few studies involving the genetics of equine osteochondrosis have also been performed. Considerable differences between progeny groups as regards radiographic findings of osteochondrosis, and heritabilities of the order 0.1-0.3, have been found (Gröndahl and Dolvik, 1993; Schougaard et al., 1990). It seems also to be generally accepted that breed differences exist. Ponies are seldom found with osteochondrosis, whereas 10 to 15% of Standardbred trotters and Warmblood riding horses, when radiographed, show the disorder. There is no evidence of a general difference in the occurrence of osteochondrosis between trotters and riding horses, although the sites of the lesions may vary somewhat between the breeds (Sandgren et al., 1993; Winter et al, 1996).

Extensive studies to investigate the prevalence of osteochondros, its heritability and importance for performance in Swedish Standardbred trotters have been conducted by Sandgren et al. (1993), Philipsson et al.(1993) and Brendow et al. (1997). The progeny (n=793) of 24 Standardbred trotter stallions were radiographed for findings of oestochondroiss (OC) in the tarsocrural (hock) joint and palmar/plantar osteochondral fragments (POF) in the metacarpo- and meta-tarsophalangeal (fetlock) joints. Clinical examinations for synovial effusion were also made. Twenty of the stallions were drawn at random from the population of Swedish Standardbred trotter stallions approved for breeding, and 4 stallions were selected for their own OC and POF status. Results are summarized in Table 2.

Hock joint OC was diagnosed in 10.5% and fetlock joint POF in 21.5% of the offsprings of the randomly selected stallions. The frequency of OC and POF varied among the progeny groups between 0-24% and 11-44%, respectively. Three times as many of the progeny (26.4%) of the stallions themselves diagnosed with OC were found with the same defect compared to the progeny of the stallions free from this defect (9.6%). A corresponding, but less pronounced, difference was found for POF. Stallions diagnosed with OC had a somewhat lower than average frequency of POF among their progeny. Similarly POF-stallions had slightly fewer than average number of progeny with OC. The predisposing factors of OC and POF therefore seem to be inherited rather independently of each other.

For estimation of heritabilities the findings were analysed as all-or-none traits and as sire-offspring regressions. The heritability was estimated to be 0.08-0.09 on the visible scale for both traits, corresponding to 0.27 for OC and to 0.17 for POF on the underlying quantitative scale. Sire-offspring regressions yielded heritabilities of 0.34 and 0.23 for OC and POF, respectively.

Although the predisposition for OC and POF seems partly to be of genetic origin their importance for performance and longevity needs to be known in order to correctly assess the value of ostechondrosis in a selection programme. Therefore these 793 horses were followed up as regards their racing careers. A total of 505 or 63.7% of the horses started at least once at the age of 3-5 years. No effect at all could be shown for any single radiologically found defect on the ability to start, or on the accumulated number of starts, % placings or earnings. Horses with multiple defects had slightly lower number of starts and performance index compared to the other horses. Radiographically diagnosed OC horses, also showing clinical signs of synovial effusion of the hock joint, had a significantly lower percentage racing progeny compared to OC-horses free from clinical signs, 59 vs 79%.

Table 2. Incidence of radiographed hock joint OC and plantar fetlock joint fragments (POF) in progeny groups of standardbred trotters (Philipsson et al., 1993)

Stallion*	No. of offspring examined	Offspring with hock joint OC		Offspring with fetlock joint POF	
		No.	%	No.	%
Randomly chosen sires					
1	34	2	5.8	10	29.4
2	34	5	14.7	5	14.7
3	34	5	14.7	5	14.7
4	39	4	10.3	8	20.5
5	37	3	8.1	5	13.5
6	35	2	5.7	8	22.9
7	33	4	12.1	9	27.3
8	37	4	10.8	6	16.2
9	33	7	21.2	8	24.2
10	32	2	6.2	9	28.1
11 (POF)	32	0	0.0	13	40.6
12	34	0	0.0	4	11.8
13	33	5	15.1	8	24.2
14	34	7	20.6	15	44.1
15	29	2	6.9	5	17.2
16	29	7	24.1	8	27.6
17	36	1	2.8	4	11.1
18 (OC+POF)	29	6	20.7	5	17.2
19	35	4	11.4	6	17.1
20	35	1	2.9	4	11.4
Total	**674**	**71**	**10.5**	**145**	**21.5**
Selected sires					
21 (POF)	29	3	10.3	13	44.8
22 (OC+POF)	30	5	16.7	7	23.3
23 (POF)	32	0	0.0	10	31.3
24 (OC)	28	12	42.9	3	10.7
Total	**119**	**20**	**16.8**	**33**	**27.7**

*Status of stallion indicated within brackets.

Roaring. Although this disorder is well documented since long and that it has a genetic origin, the etiology is not fully clear (Cahill and Goulden, 1987; Poncet et al., 1989). Tall horses with a long neck are usually more predisposed to this defect of the larynx, where an atrophied muscle may cause a noise due to some difficulties at breathing when performing. Although affected stallions generally have an increased frequency of roaring progeny there may also be other factors, e.g. infections, that cause the defect to develop.

In two retrospective Swedish studies the journals of the major animal hospitals were utilized to investigate whether two Swedish Warmblood stallions transmitted the defect to their progeny (Graaf, 1986; Stenlund, 1994). Both stallions were successful in international sport, but were surgically treated when roaring became manifest several years after approval for breeding. In the first case 107 horses were diagnosed roarers and 21 of these were by the suspect stallion. In the second study another 12 roaring progeny of this stallion were diagnosed, giving a total of at least 13% of the progeny being operated. The stallion definitely carried genes for the defect and was rejected from breeding.

In the second case a total of 106 Warmblood horses were diagnosed roarers. However, none of the 140 offsprings, at the time of the study being 4-8 years old, of the suspect stallion were found diagnosed as roarer. Consequently this stallion was reapproved for breeding.

DISCUSSION

As a strong and healthy horse is a prerequisite for performance it is obvious that preventive measures to avoid problems are extremely important. One of these measures is breeding for sound horses. The four cases presented are just examples of the variety of problems that exist and that so far only little has been done to systematically improve the health status of horses by selection. In fact, it is surprising how little attention has been paid to systematic recording and analysis of information on various defects and lesions. Many studies are just the result of special investigations in order to clarify whether a stallion transmits a problem or not.

To start with, the *first case* indicated the importance of liveability at birth and the genetic variation that may exist among as well as between breeds. It also pointed at the quality of data and reporting system used to be accurate. The *second and fourth cases,* demonstrating effects due to single or only few genes, indicated the needs of a systematic recording of defects diagnosed at animal clinics and an integration of the data from the different clinics so that the general incidence levels can be monitored as well as possible carriers of genetic defects can be detected. The studies also pointed at the importance of uniquely and accurately identified horses and their pedigrees to accomodate for reliable genetic analyses and selection decisions.

The studies on *osteochondrosis*, as an example of a developmental disorder with a multifactorial background, raise a number of questions regarding diagnosis of the defects as well as the inheritance and importance of different expressions of the lesions. Obviously, many horses do perform very well despite the defects. Several very successful stallions in international racing have been found with OC or POF. However, these horses may not have shown any clinical but radiographic signs of the disorders as young horses.

A horse population under such a strong selection for performance as the Standardbred trotters would naturally keep the most affected horses out of breeding, while horses with less pronounced lesions, as they perform equally well as defect free horses, may be selected for breeding if no other measures for selection against the lesion are taken. This assumes, though, that horses are not competing medicated and that surgeries to eliminate the defect are considered when selecting breeding stock. One important question to be answered is whether radiographed OC horses with and without other clinical signs are just different phenotypic expressions of the same genetic trait

or not. In breeds with the main selection pressure applied at a young age, before any real performance has been made, no natural selection against the defect will take place. The only way of improving the defect status would then be to consider the defect when initially selecting individuals for breeding. The requirement to assess the influence of the defect on performance and longevity and on risks at sales of horses remains high in order to apply the right weighting of traits for selection.

In the cases of defects caused by single or only few genes DNA-technologies may in future become effective tools in detecting defect animals or carriers at an early stage of life. The HYPP syndrome (*Hyperkalemic periodic paralysis*) of Quarter horses is an example where this technique has already been successfully applied (Rudolph, et al., 1992). Yet, accurate diagnosis and recording of defects will be required in order to continuously monitor health status of the populations and for analysis of family data in relation to possible candidate genes when investigating various defects.

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