Inheritance of dermoid sinus in the Rhodesian ridgeback

OBJECTIVES: **To define the mode of inheritance of dermoid sinus.**

METHODS: **A chi-squared analysis was performed on data from 46 litters produced between 1990 and 2001. Data were corrected to avoid bias in the segregation ratio.**

RESULTS: **In data from 57 litters (n=492), 82 dermoid sinus positive offspring were observed. The frequency of affected offspring in the Swedish Rhodesian ridgeback population is estimated to be between 8 and 10 per cent.**

CLINICAL SIGNIFICANCE: **Bias in heredity pattern may be caused by undetected dermoid sinus type V. Improved clinical diagnosis of all dermoid sinus types is therefore crucial.**

N. H. C. SALMON HILLBERTZ

Journal of Small Animal Practice (2005)
46, 71–74

Swedish University of Agricultural Sciences, Department of Animal Breeding and Genetics, Biomedical Centre, Box 597, S-751 24, Uppsala, Sweden

INTRODUCTION

The Rhodesian ridgeback is an African dog breed, the characteristic phenotype of which is a coat that forms a dorsal ridge. The ancestry of this phenotype is suggested to be an indigenous African breed – the Hottentot hunting dog (Hare 1932, Hawley 1984). Crossbreed dogs, which originated from European breeds, were mated with the Hottentot hunting dog over a 200-year period, resulting in today's Rhodesian ridgeback breed (Mann and Stratton 1966). The Rhodesian ridgeback breed is often associated with hereditary abnormality referred to as dermoid sinus (Hathcock and others 1979, Lambrechts 1996, Cornegliani and others 2001).

Dermoid sinus is a congenital malformation caused by an incomplete separation of the skin and neural tube during embryonic development (Fatone and others 1995, Booth 1998, Cornegliani and others 2001). The abnormality is also termed a dermoid cyst or pilonidal cyst (Tshamala and Moens 2000). However, there are anatomical differences between a dermoid sinus and a dermoid cyst (Blood and Studdert 1999). A dermoid sinus is a tube-like tract lined by hair follicles and sweat and sebaceous glands (Swenson 1989, Tshamala and Moens 2000). The lumen of the sinus may also contain sebum, hair (Lambrechts 1996, Cornegliani and others 2001) and keratin debris (Scott and others 1995). The composition of the sinus is unchanged through the epidermis, dermis and lower layers of tissue. In contrast, a dermoid cyst is a closed epithelium-lined sac or capsule containing a semi-solid or liquefied substance (Goldsmith and Shofer 1992).

A dermoid sinus connected to the skin can be detected by palpation. If the sinus tube develops a secondary infection, the dog may show signs of pain during palpation (Fatone and others 1995, Lambrechts 1996). One of the characteristics of a dermoid sinus is tufts of hair protruding from single or multiple small skin openings (Hathcock and others 1979). Common anatomical locations of dermoid sinuses are the dorsal cervical, thoracic and coccygeal areas, in other words, before and after the dorsal ridge.

Dermoid sinus occurrence has been reported in American cocker spaniels (Bailey and others 2001), boerboels (Penrith and van Shouwenburg 1994), boxers (Selcer and others 1984), chow-chows (Booth 1998), English cocker spaniels (Pratt and others 2000), golden retrievers (Cornegliani and others 2001), Rhodesian ridgebacks (Hofmeyr 1963, Hathcock and others 1979, Gammie 1986), Rhodesian ridgeback crosses (Lambrechts 1996), shih tzus (Selcer and others 1984), Siberian huskies (Cornegliani and Ghibaudo 1999) and Yorkshire terriers (Fatone and others 1995).

Different types of dermoid sinus have been described that are classified according to how deeply they penetrate towards the spine (Gammie 1986), in other words their relationship to the supraspinous ligament (Tshamala and Moens 2000), and whether or not an opening in the skin exists. Four types of sinus have been described (Mann and Stratton 1966). Booth (1998) suggested a fifth type of dermoid sinus in Rhodesian ridgebacks, which was subsequently supported by Tshamala and Moens (2000) (Fig 1). The five types of dermoid sinus and the diagnostic features of each are

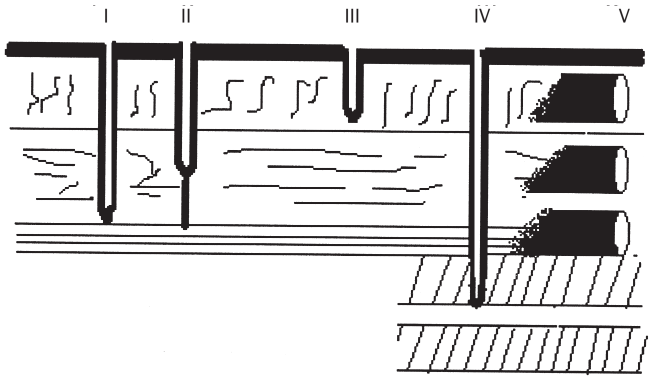

FIG 1. Five types of dermoid sinus. The classification depends on how deeply into tissue the sinus extends and whether or not a skin opening exists. Dermoid sinus type V lacks a skin opening or a definite location in a specific tissue. The diagram shows a midline longitudinal section demonstrating the different types of dermoid sinus. The original image was adapted from Mann and Stratton (1966) and has been edited by the author

Table 1. Diagnostic features of the five types of dermoid sinus

Dermoid sinus type	Features
I	Extending as far as the supraspinous ligament, and presumably nuchal ligament in the cervical area, to which it is attached. The nuchal ligament is an elastic apparatus that serves to support the head without muscular effort (Blood and Studdert 1999)
II	Not extending as far as the supraspinous ligament but connected to it by a fibrous strand
III	Not extending as far as, or connected to, the supraspinous ligament
IV	Attached to the dura mater
V	Dermoid sinus with no connection to the skin surface (Booth 1998, Tshamala and Moens 2000). No definite location in a specific tissue

listed in Table 1. The fifth type has no connection to the skin and is therefore difficult to detect via palpation of the skin. When sinus type V is located close to the spine, neurological deficiencies in the hindlimbs, behavioural changes and incontinence have been described (Booth 1998).

Mode of inheritance

Dermoid sinus in Rhodesian ridgebacks has been suggested to be inherited either as a simple autosomal recessive trait (Lord and others 1957, Mann and Stratton 1966, Angarano and Swaim 1993, Scott and others 1995), a dominant trait with incomplete penetration (Hofmeyr 1963, Unwin 1987) or a combination of two recessive genes (Mann and Stratton 1966). The first statistical trial to determine the inheritance of dermoid sinus was performed by Hare in 1932. In that study, all data were retrieved from only two sires and three dams. They produced 47 offspring. One of the sires was dermoid sinus positive. The dermoid sinus negative sire gave rise to a total of 41 offspring, and five were identified as dermoid sinus positive. The dermoid sinus affected sire gave rise to a total of six offspring, and two were dermoid sinus positive. Hare presented no opinion on the statistical results.

In 1957, Lord and others reported the mode of inheritance as being of a complex nature and not simple recessive, based upon the frequency with which dermoid sinus appeared. Hofmeyr (1963) suggested that dermoid sinus was of a dominant nature with incomplete penetration, referring to the report written by Lord and others (1957). No statistical evidence was supplied by either Lord and others (1957) or Hofmeyr (1963).

The data by Hare (1932) were re-evaluated by Mann and Stratton in 1966. They concluded that the mode of inheritance was of a simple recessive character. Stratton collected data from breeders relating to 17 sires and 21 dams. One dam was dermoid sinus positive. The matings resulted in 48 litters with a total of 376 offspring. When an analysis was performed using only the litters in which dermoid sinus had been detected (20 litters), the expected number of dermoid sinus positive offspring was 45. The actual outcome was 42 dermoid sinus positive offspring. This analysis supports the hypothesis of a simple autosomal recessive trait (Swenson 1989).

In order to better define the mode of inheritance of dermoid sinus, the work reported in the present study more extensively evaluates the incidence of dermoid sinus in the Swedish Rhodesian ridgeback population.

MATERIALS AND METHODS

Swedish Rhodesian ridgeback breeders have been reporting the occurrence of dermoid sinus to the Swedish Rhodesian Ridgeback Club since 1964. The register from the club is official and available to the public. The Swedish Rhodesian ridgeback population comprises approximately 2500 animals (Swedish Kennel Club 2000 to 2002). The breeding population during the period 1995 to 2001 was 305 animals (171 dams and 134 sires).

Data

Data were collected from the Swedish Rhodesian Ridgeback Club and refer to a total of 129 litters (1040 offspring) derived from matings between 103 sires and 106 dams over an 11-year period (1990 to 2001). The litters included in the study were those in which culling of offspring was exclusively due to dermoid sinus. The author performed the categorisation of the material. None of the parent animals included in this study were recorded as dermoid sinus positive.

The data were analysed from two different angles. One analysis was performed based on the litters where dermoid sinus positive offspring (nn) had been produced and the other analysis was performed based on the classified heterozygote parent animals. Correction of litter data was performed to avoid bias in the segregation ratio, as some parent carriers produced litters where all offspring were dermoid sinus negative (NN or Nn). The correction formula used was $q' = q/(1\text{-}p^s)$, where q is the expected frequency of nn (0·25), p is the expected frequency of NN or Nn (1-0, 25), q´ is the corrected expected frequency of nn and s is the litter size (Cavalli-Sforza and Bodmer 1971). When applying the correction formula to the litter data, it was presumed that the parent animals were phenotypically normal heterozygote carriers. Standard chi-squared analysis was performed to test the null hypothesis that data did not deviate from the expected 3:1 distribution of unaffected and affected animals.

RESULTS

Recordings of 72 litters showed no evidence of dermoid sinus positive offspring (a total of 548 individuals). Nine of 62 dams and 22 of 58 sires had produced dermoid

Table 2. Corrected data for 46 litters (from a total of 57 litters produced by classified heterozygote parent animals) in which dermoid sinus positive offspring were identified. Correction of the data was performed to avoid bias in the segregation ratio. The data covers litters produced during 1990 to 2001

Litter number	Sire	Dam	Number observed		Expected			
					Uncorrected		Corrected	
			Normal	Defect	Normal	Defect	Normal	Defect
73	Y59	X63	7	1	6	2	5·78	2·22
74	Y60	X64	8	2	7·5	2·5	7·35	2·65
75	Y60	X64	10	0	–	–	–	–
76	Y61	X65	10	0	–	–	–	–
77	Y62	X66	4	4	6	2	5·78	2·22
78	Y63	X67	7	5	9	3	8·90	3·10
79	Y63	X67	8	0	–	–	–	–
80	Y64	X68	7	1	6	2	5·78	2·22
81	Y65	X69	7	3	7·5	2·5	7·35	2·65
82	Y66	X70	9	2	8·25	2·75	8·13	2·87
83	Y67	X71	9	1	7·5	2·5	7·35	2·65
84	Y68	X9	2	0	–	–	–	–
85	Y68	X9	4	2	4·5	1·5	4·18	1·82
86	Y69	X71	9	0	–	–	–	–
87	Y70	X72	10	1	8·25	2·75	8·13	2·87
88	Y71	X73	10	1	8·25	2·75	8·13	2·87
89	Y72	X74	5	1	4·5	1·5	4·18	1·82
90	Y72	X75	2	2	3	1	2·54	1·46
91	Y73	X70	9	0	–	–	–	–
92	Y74	X73	7	1	6	2	5·78	2·22
93	Y75	X74	10	1	8·25	2·75	8·13	2·87
94	Y76	X75	11	1	9	3	8·90	3·10
95	Y77	X76	9	1	7·5	2·5	7·35	2·65
96	Y77	X76	11	2	9·75	3·25	9·67	3·33
97	Y78	X77	7	1	6	2	5·78	2·22
98	Y79	X20	2	2	3	1	2·54	1·46
99	Y80	X49	10	0	–	–	–	–
100	Y80	X78	10	0	–	–	–	–
101	Y81	X79	3	1	3	1	2·54	1·46
102	Y82	X80	6	5	8·25	2·75	8·13	2·87
103	Y83	X81	6	1	5·25	1·75	4·98	2·02
104	Y84	X78	10	2	9	3	8·90	3·10
105	Y85	X51	6	2	6	2	5·78	2·22
106	Y86	X79	7	1	6	2	5·78	2·22
107	Y87	X80	7	2	6·75	2·25	6·57	2·43
108	Y88	X81	6	1	5·25	1·75	4·98	2·02
109	Y89	X65	5	2	5·25	1·75	4·98	2·02
110	Y89	X82	3	5	6	2	5·78	2·22
111	Y90	X83	7	1	6	2	5·78	2·22
112	Y90	X84	12	1	9·75	3·25	9·67	3·33
113	Y90	X85	9	1	7·5	2·5	7·35	2·65
114	Y90	X86	6	2	6	2	5·78	2·22
115	Y91	X80	9	2	8·25	2·75	8·13	2·87
116	Y92	X73	11	0	–	–	–	–
117	Y93	X87	6	2	6	2	5·78	2·22
118	Y94	X77	5	0	–	–	–	–
119	Y95	X88	8	0	–	–	–	–
120	Y95	X58	2	1	2·25	0·75	1·70	1·30
121	Y96	X89	8	2	7·5	2·5	7·35	2·65
122	Y97	X49	8	2	7·5	2·5	7·35	2·65
123	Y98	X90	8	1	6·75	2·25	6·57	2·43
124	Y99	X91	1	1	1·5	0·5	0·86	1·14
125	Y100	X92	6	3	6·75	2·25	6·57	2·43
126	Y101	X93	6	1	5·25	1·75	4·98	2·02
127	Y102	X75	9	2	8·25	2·75	8·13	2·87
128	Y103	X94	8	2	7·5	2·5	7·35	2·65
129	Y103	X95	8	1	6·75	2·25	6·57	2·43
Sum	**45**	**44**	**410**	**82**	**300**	**100**	**290·0**	**110·0**

Normal An individual that has not been reported as dermoid sinus positive
Defect An individual that has been reported as dermoid sinus positive

sinus positive offspring in previous mating combinations. The nine dams and 22 sires were classified as heterozygote Nn. The remaining parent animals had no identified dermoid sinus positive offspring in previous combinations and were classified as homozygote NN or heterozygote Nn.

In recordings from 57 litters, 46 litters showed dermoid sinus positive offspring. All parent animals (45 sires and 44 dams) were classified as heterozygote Nn. The observed numbers of dermoid sinus positive offspring were 82, whereas 410 individuals were defined as phenotypically normal.

Chi-squared tests were performed on data from the 46 litters (Table 2) in which dermoid sinus positive offspring had been identified. This showed that χ^2 (uncorrect. expected)=43;57, $P<0{\cdot}05$, and χ^2 (correct. expected)=56;75, $P<0{\cdot}05$.

Based on reported and unreported cases, the frequency of dermoid sinus in this Rhodesian ridgeback population was assumed to range between 8 and 10 per cent.

DISCUSSION

The Swedish Rhodesian ridgeback register may be unique. The objective of the Swedish Rhodesian Ridgeback Club is to register all animals, as well as the health status of produced litters. The vast amount of information in the register allows not only classifications of parent animals (according to the hypothesis), but also enables analysis of the outcome of produced litters. It must be emphasised that the data concerning dermoid sinus appearance were reported by breeders to the Swedish Rhodesian Ridgeback Club and therefore the results entirely rely upon the accuracy of the breeders' information. Some individuals that are born with incorrect ridges, or ridgeless, are culled and normally not examined for dermoid sinus. Stillborn individuals are normally not examined either. As a consequence, a vital piece of information concerning dermoid sinus frequency is lost.

There is no information in earlier reports (Hare 1932, Mann and Stratton 1966) on whether individuals culled for reasons other than dermoid sinus have been excluded from statistical trials. To avoid bias caused by these problems, the litters included in the present study were those where dermoid sinus was the single reported reason for culling.

It is presumed that the information of litter status given by breeders to the Swedish Rhodesian Ridgeback Club was noted consecutively. From the total amount of litters included, 83 (64 per cent) of the litters showed no identified dermoid sinus positive offspring, according to breeders. The observed number of dermoid sinus positive offspring produced during 1990 to 2001 was 82. Subsequently, an analysis was performed only on those litters where dermoid sinus positive offspring had been detected (82 individuals).

Results from the chi-squared tests performed on the litter data did not support the hypothesis of a simple autosomal recessive mode of inheritance. In other words, there were significant deviations between observed and expected numbers of dermoid sinus positive offspring. Available data instead suggested a more complex dihybrid mode of inheritance. Unfortunately, the currently available material cannot formally prove such a mode of inheritance. There are two possible explanations for the deviations between the observed and corrected expected numbers of dermoid sinus positive offspring (Table 2). Some individuals are not identified as dermoid sinus positive until they have become older and secondary infection has occurred. These individuals are normally not included in the Swedish Rhodesian ridgeback register, since there are no frequent updates concerning the health status of produced litters. Thus, animals that are diagnosed as dermoid sinus positive later in life will influence dermoid sinus frequency. This is an explanation supported by the breeding committee of the Swedish Rhodesian Ridgeback Club.

A rather controversial explanation could be that the existence of sinus type V has remained undetected. Homozygote recessive individuals may not be identified as dermoid sinus affected. As a consequence, individuals affected by type V dermoid sinus could incorrectly be included in the data where parent animals are presumed to be homozygote NN or heterozygote Nn (litters with no apparent dermoid sinus positive individuals) instead of the data in Table 2 (litters with observed dermoid sinus positive individuals). Based on the current study, and considering the probable explanations for the deviations between observed and expected numbers of dermoid sinus positive offspring, the hypothesis of a recessive mode of inheritance cannot be formally excluded. In addition, differential penetrance of the disease appears to be consistent with the clinical phenotype of multiple types of dermoid sinus observed (Fig 1). Thus, a reasonable hypothesis is that a major disease gene, the activity of which is influenced by modifying genes, may play a role during the developmental stages of dermoid sinus. It is important that Rhodesian Ridgeback breeders are informed of the existence of the fifth sinus type (with no skin opening) in an effort to develop diagnostic procedures to detect this type of dermoid sinus.

The definition of a dermoid sinus is based on histological findings throughout this paper, and type V dermoid sinus may have escaped detection (as discussed above). If some of the animals classified as dermoid sinus negative were used for breeding, but were actually positive for the undetected dermoid sinus type V, attempts to identify mode of inheritance via pedigrees would not be completely reliable.

Acknowledgements

The author thanks all the Swedish Rhodesian ridgeback breeders who have made this study possible; Dr P. E. Sundgren, for his guidance, assistance, good ideas and friendship; Dr L. Swenson, for his support and positive influence during this study; Professor G. Andersson, for suggestions, critical reading of the manuscript, optimism and support; and Dr E. Hellmén, for help with the postmortem examinations performed on the dermoid sinus affected puppies.

References

Angarano, D. W. & Swaim, S. F. (1993) Congenital skin diseases. In: Disease Mechanisms in Small Animal Surgery. 2nd edn. Ed M. J. Bojrab. Lea & Febiger, Philadelphia. pp 178-183

Bailey, T. R., Holmaberg, D. L. & Yager, J. A. (2001) Nasal dermoid sinus in an American cocker spaniel. *Canadian Veterinary Journal* **42**, 213-215

Blood, D. C. & Studdert, V. P. (1999) Saunders Comprehensive Veterinary Dictionary. 2nd edn. W. B. Saunders, London. pp 305, 329

Booth, M. J. (1998) Atypical dermoid sinus in a chow chow dog. *Journal of the South African Veterinary Association* **69**, 102-104

Cavalli-Sforza, L. L. & Bodmer, W. F. (1971) The Genetics of Human Populations. W. H. Freeman, San Francisco

Cornegliani, L. & Ghibaudo, G. (1999) A dermoid sinus in a Siberian husky. *Veterinary Dermatology* **10**, 47-49

Cornegliani, L., Jommi, E. & Vercelli, A. (2001) Dermoid sinus in a golden retriever. *Journal of Small Animal Practice* **42**, 514-516

Fatone, G., Brunetti, A., Lamagna, F. & Potena, A. (1995) Dermoid sinus and spinal malformations in a Yorkshire terrier: diagnosis and follow-up. *Journal of Small Animal Practice* **36**, 178-180

Gammie, J. S. (1986) Dermoid sinus removal in a Rhodesian ridgeback dog. *Journal of the South African Veterinary Association* **27**, 250-251

Goldsmith, M. H. & Shofer, F. S. (1992) Cysts. In: Skin Tumors of the Dog and Cat. Pergamon, Oxford. pp 271-283

Hare, T. (1932) A congenital abnormality of hair follicles in dogs resembling trichostasis spinulosa. *Journal of Pathology and Bacteriology* **35**, 569-571

Hathcock, T. J., Clampett, G. E. & Broadstone, V. R. (1979) Dermoid sinus in a Rhodesian ridgeback. *Veterinary Medicine, Small Animal Clinician* **74**, 53-56

Hawley, T. C. (1984) The Hottentot dog. In: The Rhodesian Ridgeback. The Origin, History and Standard. 4th edn. N. G. Sendingpers, Bloemfontein. pp 11-12

Hofmeyr, C. F. B. (1963) Dermoid sinus in the ridgeback dog. *Journal of Small Animal Practice* **4**, 5-8

Lambrechts, N. (1996) Dermoid sinus in a crossbred Rhodesian ridgeback dog involving the second cervical vertebra. *Journal of the South African Veterinary Association* **67**, 155-157

Lord, L. H., Cawley, A. J. & Gilray, J. (1957) Mid-dorsal dermoid sinuses in Rhodesian ridgeback dogs – a case report. *Journal of the American Veterinary Medical Association* **131**, 515-518

Mann, G. E. & Stratton, J. (1966) Dermoid sinus in the Rhodesian ridgeback. *Journal of Small Animal Practice* **7**, 631-642

Penrith, M-L. & Van Shouwenburg, S. (1994) Dermoid sinus in a Boerboel bitch. *Journal of the South African Veterinary Association* **65**, 38-39

Pratt, J. N. J., Knottenbelt, C. M. & Welsh, E. M. (2000) Dermoid sinus at the lumbosacral junction in an English springer spaniel. *Journal of Small Animal Practice* **41**, 24-26

Scott, D. W., Miller, W. H. & Griffin, G. E. (1995) Congenital and hereditary defects. In: Small Animal Dermatology. 5th edn. Eds D. W. Scott, W. H. Miller and G. E. Griffin. W. B. Saunders, Philadelphia. pp 736-805

Selcer, E. A., Helman, R. G. & Selcer, R. R. (1984) Dermoid sinus in a shih tzu and a boxer. *Journal of the American Animal Hospital Association* **20**, 634-636

Swedish Kennel Club (2000-2002) Registration Statistics – Rhodesian ridgeback. *Journal of the Swedish Kennel Club*, **1-2**

Swenson, L. (1989) Ärftlig dermoid sinus hos Rhodesian ridgeback. *Doggy Rapport* **1**, 5

Tshamala, M. & Moens, Y. (2000) True dermoid cyst in a Rhodesian ridgeback. *Journal of Small Animal Practice* **41**, 352-353

Unwin, D. D. (1987) The dermoid sinus. In: Rhodesian Ridgeback. Ed A. Woodrow. Information Printing, Oxford. pp 155-159