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Prevalence and inheritance of canine elbow dysplasia in German Rottweiler

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Summary

A total of 2114 scores of elbow arthrosis from the official screening programme of the German Rottweiler Breeding Association (ADRK) were analysed in respect of prevalence and genetic disposition. 45.8% showed no signs of arthritis, 40.6% were scored in Grade 1 with minor osteophytes and 13.6% were affected by arthritis of clinical relevance (Grade 2 and 3). REML estimates showed a heritability of 28% and a litter variance of 6.4%. The only significant environmental fixed effect was gender. 39.1% of the male and 51.5% of the female were free from ED which corresponds with 19.2% of the male and only 8.8% of the female in the critical ED classes of Grade 2 and 3. Differences between the years of examination could be explained by genetic gain. Month of birth as well as age at examination, in the range covered by this study, was not significant. It was stated that the effect of bodyweight should be tested before starting a breeding programme.

Zusammenfassung

Verbreitung und Erblichkeit der Ellbogendysplasie des Hundes in der Deutschen Rottweiler Population

2114 Bewertungen der Ellbogenarthrose aus dem offiziellen Screening des Allgemeinen Deutschen Rottweiler Klub (ADRK) wurden in Hinblick auf Häufigkeit und genetische Praedisposition untersucht. 45,8% der Tiere hatten keine Anzeichen von Arthrose, 40,6% wurden mit Grad 1, geringgradige Osteophythenbildung, bewertet und 13,6% waren mit Arthrosen Grad 2 und 3 von klinischer Relevanz behaftet. REML-Schätzungen zeigten eine Heritabilität von 28% und eine wurfbedingte Varianz von 6.4%. Der einzige signifikante fixe Umwelteffekt war der Einfluß des Geschlechtes. 39,1% der Rüden und 51,5% der Hündinnen waren ED-frei. Das korrespondiert damit, daß 19,2% der Rüden und nur 8,8% der Hündinnen in die kritischen ED-Klassen 2 und 3 eingestuft wurden. Unterschiede zwischen den Untersuchungsjahren konnten als genetischer Trend erklärt werden. Der Geburtsmonat sowie das Alter beim Röntgen, zumindest in dem Altersbereich, den diese Studie umfasste, waren nicht signifikant. Es wurde als wichtig angesehen, den Einfluß des Körpergewichtes näher zu prüfen, bevor ein Zuchtprogramm begonnen wird.

Introduction

Elbow dysplasia (ED) is a generic term for malformation and malfunction in the elbow joint. ED can be detected and only be graded if lesions or secondary arthrosis exist. The most common lesions in dogs are isolated processus anconeus (IPA), fragmented coronoid process (FCP) and osteochondrosis dissecans (OCD). However, even without observation of these visible defects, mild to severe osteoarthrotic changes can occur and lead to lameness and pain.

Definition of ED and literature overviews have been given by different authors, e.g. OLSSON (1974), FOX et al. (1983), FLÜCKIGER (1992), SVENSON et al. (1997), etc. A review including therapeutic aspects is given by WALDE and TELLHELM (1991). A standardized grading system, mainly based on the amount of osteophytic formation, is suggested by the

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International Elbow Working Group (IEWG). It is also the basis of grading within the German dog populations (LOEFFLER 1997).

Canine elbow dysplasia (ED) is known as a partly inherited disease in many dog breeds. Variation in the incidence level between breeds as well as between lines within breeds (families) indicate genetic variation in the risk of being affected. Thus breeding strategies can be applied to reduce lesions and secondary arthritis in the joint.

ED is mostly reported from heavy or fast-growing breeds (FLÜCKIGER 1992), of which the Rottweiler is one. The analysis of ED in national Rottweiler populations has been reported from Norway (GRONDALEN and LINGAAS 1991), Sweden (SVENSON et al. 1997) and Finland (MÄKI et al. 2000), with a relatively high frequency of affected animals of between 40 and 50%. Heritability estimates of these authors showed a clear genetic predisposition for the Rottweiler breed.

SVENSON et al. (1997) estimated different heritabilities for ED information from males ($h^2 = 0.34$) and females ($h^2 = 0.40$), using dam–daughter-and sire–son regressions. On the basis of the standard errors, the difference was not significant. Heritability estimates from Norway (GRONDALEN and LINGAAS 1991) were based on paternal half-sib-correlation ($h^2 = 0.10$) and full-sib-correlation ($h^2 = 0.29$). In Finland the estimates were derived from the estimated genetic variance using the restricted maximum likelihood procedure REML with an animal model (MÄKI et al. 1999). The estimate was $h^2 = 0.31 \pm 0.04$.

In 1997, after 1 year explorative screening, the German Rottweiler Breeding Association (ADRK) decided to introduce an obligatory testing programme for ED. The aim of this study was to characterize the ED situation in the population, to estimate genetic parameters and to discuss the consequences with respect to future breed management.

Materials and methods

The German Rottweiler population breeds about 3000 puppies a year with slight variation from year to year. About 27% of the population is X-rayed routinely for hip dysplasia (HD) and, since 1997, they are examined for HD and ED at the same time. Up to the beginning of this investigation a total of 2114 dogs had been tested. Elbow dysplasia is scored by a single scrutineer following the guidelines of the International Elbow Working Group (IEWG). ED is subdivided into four classes from unaffected (ED free) up to severe dysplasia. The grading of affected joints is proportional to the amount of osteophytes: grade 1 (mild arthrosis) covers the visible range up to 2 mm; grade 2 (moderate arthritis) from 2 to 5 mm and the most severe case, grade 3, shows more than 5 mm.

Observable lesions such as fragmented coronoid process of the ulna (FCP), osteochondrosis dissecans of the humerus (OCD) and isolated processus anconeus of the ulna (IPA) are recorded separately. The grading in this investigation was therefore based on arthrotic osteophytes only.

The data for ED and the whole pedigree data of the population were extracted from the main Rottweiler database. Complete pedigree data was provided for the 2114 animals examined. A total of 3801 dogs were included in the genetic relationship matrix.

As official registered parental results are missing because of the short time period of operation of the scheme, the genetic analysis of the population was limited to statistical parameters coming from the genetic relationship of collateral relatives. The full-sib or half-sib covariances have been estimated via variance components by analysis of variance (ANOVA) techniques. The chosen program LSMLMW (HARVEY 1987) uses Henderson's Method III for unbalanced data with nested random effects and cross-classified fixed effects. The interpretation of these covariances allows the additive genetic and maternal variance to be quantified.

The additive genetic and the maternal variance was additionally estimated using the REML algorithm (PATTERSON and THOMPSON 1971) with an animal model including

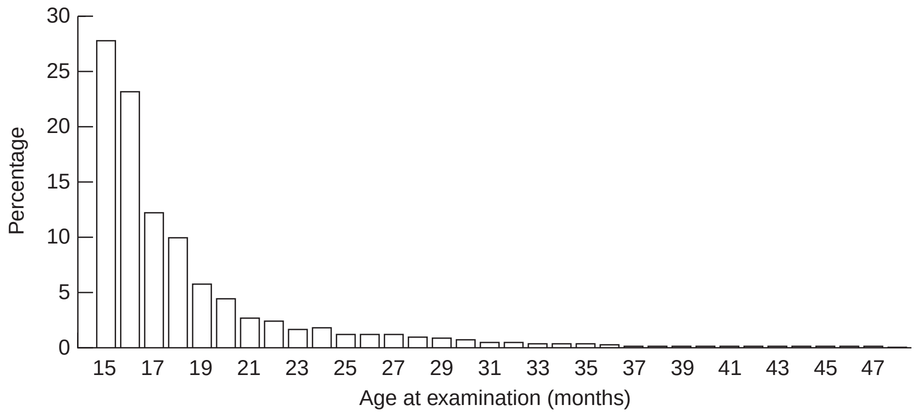

Fig. 1. Distribution of age at examination for elbow dysplasia in German Rottweiler dogs

the more complex additive genetic relationship matrix. The software packages used were MTDFREML (BOLDMAN et al. 1993) and VCE (NEUMAIER and GROENEVELD 1998).

The mixed model used in the REML variance component estimation was:

$$y_{ijklm} = \mu + a_i + m_j + S_k + MOB_l + YOE_m + b \times age_i + e_{ijklm}$$

with y = ED score; μ = population mean: a_i = additive genetic effect of animal i (random); m_j = maternal (litter) effect associated with mother j (random); S_k = effect of sex k (fix), k = 1 (male), 2 (female); MOB_l = effect of month of birth l (fix), l = 1–12 (Jan. to Dec.); YOE_m = effect of year of examination m (fix), m = 1996 to 1999; $b \times age_i$ = regression of ED. on age at examination of animal i; and e_{ijklm} = residual random effect.

The model for ANOVA had a nested design with sires, dams within sires, cross-classified with all fixed effects and with age at examination as a covariate:

$$y_{ijklmn} = \mu + s_i + d_{ij} + S_k + MOB_l + YOE_m + b \times age_n + e_{ijklmn}$$

where y = ED score; μ = population mean; s_i = effect of the sire i (random); d_{ij} = effect of the dam j within sire i (random); S_k = effect of sex k (fix), k = 1 (male), 2 (female); MOB_l = effect of month of birth l (fix), l = 1–12 (Jan to Dec); YOE_m = effect of year of examination m (fix), m = 1996–99; $b \times age_n$ = *regression of ED* on age at examination of animal$_n$; and e_{ijklmn} = residual random effect.

The use of both models should allow a comparison of the methods in respect to compatibility and a comparison with the results of other investigators. The principals and advantages of different methods are discussed by SEARLE (1989) and HOFER (1998).

Results and discussion

Descriptive statistics

In total 2114, mainly young Rottweiler dogs have been scored at the average age of 18 months with a range of 15 to 48 months. The distribution of age at examination is shown in Figure 1. Most dogs are scored at the recommended age of between 15 and 18 months, but there were also older dogs which had to be examined at a greater age in order to get a licence for breeding.

The descriptive statistics for ED in the different subgroups for month of birth, gender

Table 1. **Descriptive statistics for elbow dysplasia in Rottweiler dogs in different subgroups**

	n	ED free	ED 1	ED 2	ED 3
Month of birth					
January	116	65	40	11	0
February	179	99	51	29	0
March	236	128	83	18	7
April	198	108	70	15	5
May	273	130	107	30	6
June	171	63	83	19	6
July	231	80	114	31	6
August	191	67	89	30	5
September	167	65	72	26	4
October	154	79	60	13	2
November	107	42	52	9	4
December	91	42	38	7	4
Gender					
Males	974	381	406	148	39
Females	1140	587	453	90	10
Year of examination					
1996	293	102	148	34	9
1997	631	269	257	81	24
1998	887	434	345	96	12
1999	303	163	109	27	4
Total	2114	968	859	238	49

and year of examination are presented in Table 1. It contains an overview about seasonal differences in reproduction. There are higher numbers of puppies in early spring to summer and decreasing numbers towards winter. Males are represented significantly less than females, which corresponds to the higher demand for female than for male dogs in the breeding programme. The frequency of animals examined increased rapidly from 1996 to 1999, where it should be noted that the year 1999 is limited to data of the first 4 months.

Special attention should be paid to the significant sex effect. In total, 974 males and 1140 females were examined. Figure 2 illustrates the frequency of elbow arthritis in the whole population and the differences in the frequencies of ED between males and females. Only 39.1% of the male dogs were free of arthritis versus 51.5% of the females. This highly significant difference of 12.4% corresponds to 2% more males scored with mild arthritis in Grade 1, 7.3% in Grade 2 and 3.1% in Grade 3. However, if Grade 2 and Grade 3 are assumed to be relevant for clinical problems, males are much more on risk in this respect. This difference may partly be caused by different body weights between the sexes. Unfortunately no information was available in this investigation about the individual body weights.

Figure 3 shows the influence of the year of examination on the arthrotic changes in the elbow joint. From 1996 to 1999 the proportion of unaffected animals increased from 34.8 to 53.8%, whereas the proportion of dogs with arthrosis decreased in the same period. Explanations for this trend could be changes in the scoring system, growing pre-selection or genetic gain.

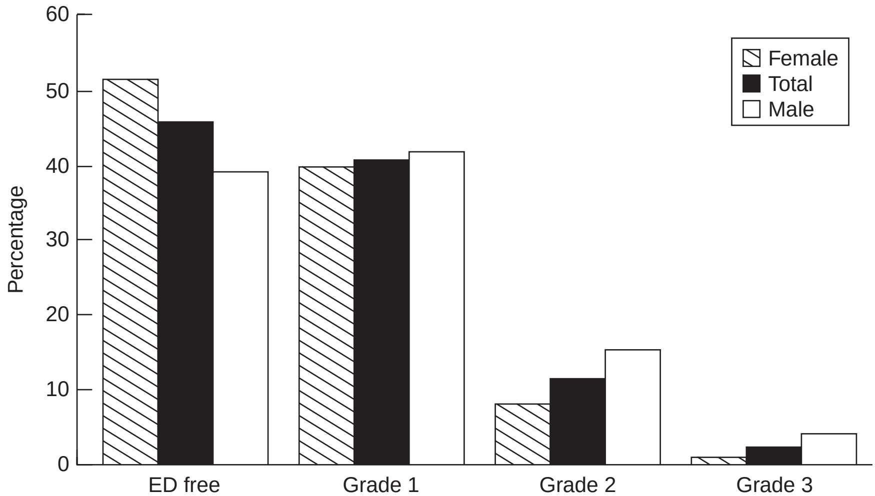

Fig. 2. Frequency of elbow arthrosis in the whole German Rottweiler population and sex distribution

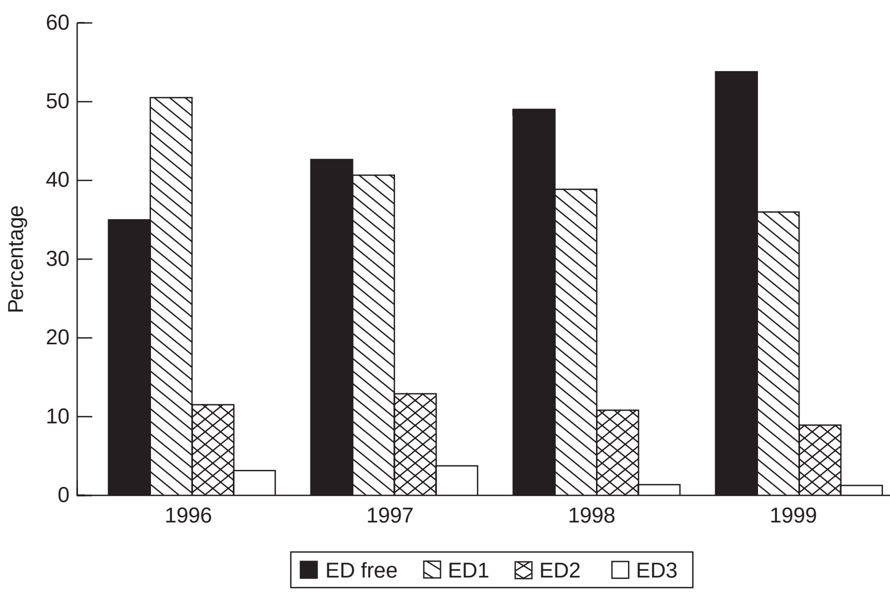

Fig. 3. Distribution of elbow displasia in Rottweiler dogs within years of examination

Multifactorial analysis of variance

The results of the hierarchical analysis of variance are shown in Table 2. The random effects 'sire' and 'dam within sire' were found to have a highly significant influence on ED. This indicates, that osteoarthrotic changes in the elbow joint are partly of genetic origin. The influences month of birth, year of examination and the regression on age at examination

Table 2. **Combined least-squares analysis of variance for elbow dysplasia in the German Rottweiler population**

Source of variation	Degrees of freedom	Mean squares	*F*-value	Probability
Sire	272	0.847	1.466	0.0000 ***
Dam within sire	829	0.578	1.373	0.0000 ***
Sex	1	17.330	41.186	0.0000 ***
Month of birth	11	0.525	1.248	0.2498 NS
Year of examination	3	0.097	0.231	0.8747 NS
Regression		0.062	0.148	0.7006 NS
Age at examination	1	0.062	0.148	0.7006 NS

NS, not significant; *** highly significant $p < 0.001$

were not found to be significant. Only gender had a highly significant influence ($p < 0.001$) on the ED score.

Environmental effects

Raw data means and least square means of all fixed effects are given in Table 3. Least square means are derived as a linear combination from REML as well as from ANOVA estimates and are, by definition, each corrected for imbalance with respect to the other effects.

The effect of month of birth showed no evident seasonal trend within year. A comparison between the two estimation procedures and the means of the raw data is illustrated in Figure 4. The difference between the results of REML and ANOVA can be explained by the fact that the REML estimation contains more information about relatives by involving the relationship matrix.

The effect of gender was the only significant environmental effect in this investigation. As already shown in Figure 2, the sex difference in the level of ED was of great importance. Means are nearly equal in all estimation procedures. The difference is about one-quarter ED score.

The year of examination indicates a degradation in the raw data, but under the multi-factorial model the year-effect was reduced to a non-significant influence. This was caused by introducing the random genetic effect. Although under the hierarchical ANOVA the variance component between sires could be confounded with the year of examination, the REML estimates with the complete relationship matrix should break down such connections; however, under REML the year of examination also diminishes to an unimportant difference. By this the trend must be interpreted as genetic progress, although the mechanism of selection, especially in the first year, cannot be easily seen. Mating decision and date of examination were about 16 months apart and in the first year the results had not been published. In dog breeding the genetic gain comes from choosing the right partner from a 'market' of male stud dogs. So the response to these decisions comes 8 weeks later, when the puppies are born and the response is observable, when the dogs are reared and X-rayed at the age of 1 year. It is possible that the communication between breeders is very efficient, without the official publication of results. Screening is only mandatory for the newly licensed dogs, but results from earlier time and from the parents may also exist. Perhaps the owners of bitches ask for the results of the males before making a mating decision. This could explain the progress from the first to the second year when there had been no official publication of results.

Although there is a great variation in the age at X-raying, the estimated linear regression

Table 3. **Level of elbow dysplasia of Rottweiler dogs in different subgroups**

	Raw data	REML	ANOVA	
	Mean	Mean (LSM)	Mean (LSM)	SE
Month of birth				
January	1.53	1.61	1.70	0.215
February	1.60	1.66	1.26	0.240
March	1.59	1.62	1.75	0.195
April	1.58	1.58	1.59	0.194
May	1.67	1.74	1.81	0.176
June	1.81	1.88	1.90	0.229
July	1.83	1.87	1.77	0.230
August	1.85	1.90	1.65	0.250
September	1.81	1.91	2.07	0.184
October	1.59	1.69	1.30	0.256
November	1.76	1.83	1.90	0.322
December	1.70	1.80	2.10	0.262
Gender				
Males	1.84	1.86	1.86	0.038
Females	1.58	1.61	1.60	0.037
Year of examination				
1996	1.82	1.87	1.78	0.098
1997	1.77	1.86	1.71	0.060
1998	1.64	1.82	1.70	0.059
1999	1.57	1.80	1.74	0.094
Age at examination				
Regression	$b = 0.0086$	$b = 0.0093$	$b = 0.0029$	0.0074

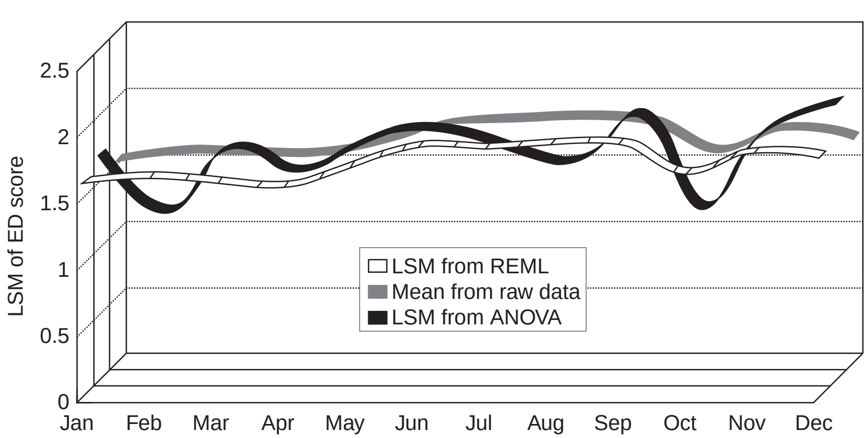

Fig. 4. Level of elbow dysplasia of Rottweiler dogs in different month of birth

Table 4. **Variance component estimation in Rottweiler dogs for elbow dysplasia from ANOVA**

Source of variation	Relative variance component			Genetic interpretation
Full-sibs	0.220			$0.5 \times \sigma^2_A + \sigma^2_{DO/EP/MAT}$
Sire		0.062		$0.25 \times \sigma^2_A$
Dam within sire		0.158		$0.25 \times \sigma^2_A + \sigma^2_{DO/EP/MAT}$
Litter			0.096	$\sigma^2_{DO/EP/MAT}$
Residual	0.780			$0.5 \times \sigma^2_A + \sigma^2_E$
Heritability		$h^2 = 0.25$		

$\sigma^2 A$ = additive genetic variance.
$\sigma^2_{DO/EP/MAT}$ = variance caused by dominance (1/4), epistasie (1/4) and maternal effects.
σ^2_E = environmental variance and residual nonadditive genetic variance.

Table 5. **Variance component estimation in Rottweiler dogs for elbow dysplasia from REML**

	Absolute variances	Relative variances
Additive genetic variance	0.1592	0.279
Variance caused by maternal effects	0.0368	0.064
Environmental variance	0.3754	0.657
Phenotypic variance	0.5714	1.000
Heritability	$h^2 = 0.28$	

indicates a low level of influence of this parameter on ED. However, this does not cover the time before the age of 15 months.

Genetic analysis

The genetic effect on ED is described by the variance components, which are found to be highly significant (Table 2). Results from ANOVA are given in Table 4. The sire component is 0.062 and represents one-quarter of the additive genetic variance. This leads to an heritability estimate of $h^2 = 0.25$. The variance component between dams within sires is also highly significant. The difference between dam and sire component is caused by effects associated with the full-sibship. This maternal component (9.6% of the total variance) covers maternal environment as well as non-additive genetic influences (e.g. one-quarter of the dominance and epistatic variance). This obvious maternal influence indicates that it is necessary to include a litter or a maternal effect in any genetic model to avoid biased estimates of heritability.

The results from the REML-procedure are given in Table 5. The results are quite similar, $h^2 = 0.28$ versus $h^2 = 0.25$. The maternal effect accounts for 6.44% of the total variance. A model without the maternal effect gave a drastic overestimation of the heritability ($h^2 = 0.38$).

Conclusions

A high frequency of elbow arthrosis (54.2%) exists in the German Rottweiler population with critical numbers of medium and severely affected dogs in terms of animals welfare.

The estimated genetic variance component of 28% indicates a genetic background strong enough to apply successful breeding strategies. However, the heritability is not high enough to select without information from relatives by simple mass selection. For breeding value estimation the model should include the random litter effect to avoid a bias caused by using information from siblings of the same litter. The REML analysis showed 6.4% of the total variance for these effects.

Males are represented more often in the higher ED classes. Further investigations must test the hypothesis that this sexual dimorphism is caused by the higher body weight of males. If that is true, breeding on the basis of ED scores might conflict with the breeding goal of Rottweilers. It will diminish expected genetic gain if there is contraselection caused by preferring antagonistic conformation traits. It also may be that the genetic variance of ED is partly caused by genetic variance of body weight. This would lead to a model with body weight as a covariate for estimation of partial genetic ED variance as well as for estimating ED breeding values. Breeding values for body mass can additionally support the breeder in assortative compensatory mating. A similar technique was applied by MEISSEN (1996) to improve racing performance in Whippets without increasing wither height.

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