GUIDELINES FROM THE HEALTHY DOG BREEDING TASK FORCE

Chondrodysplasia, chondrodystrophy and hereditary increased risk of intervertebral disc disease

In brief

What does this factsheet cover?
This factsheet deals with two genetic variants that influence the leg length of dogs. These are so-called FGF4 retrogenic insertions on chromosome 12 (chondrodystrophy) and chromosome 18 (chondrodysplasia). Both variants and the shortleggedness associated with them occur in many pedigree dogs and crossbreeds. The mutated alleles in these two genetic variants are frequently linked to the occurrence of intervertebral disc herniation. These associations are outlined below.

What are the health implications?
The allele responsible for chondrodysplasia does not lead to an increased risk of intervertebral disc herniation and is considered non-critical by the members of the task force.

The allele responsible for chondrodystrophy is associated with an increased risk of intervertebral disc herniation in some dog breeds. In other short-legged dog breeds, this association has not yet been described. In dog breeds where the chondrodystrophy allele is associated with an increased risk of intervertebral disc disease, this constitutes a genetic risk factor. This means that the risk of developing clinically manifest intervertebral disc disease is higher, but dogs do not necessarily develop the condition. Whether intervertebral disc disease actually develops depends, in addition to the genotype at the chondrodystrophy locus, on many other genetic factors and environmental factors that are, in some cases, not yet sufficiently well studied (e.g., body weight, physical activity, neuter status)..

How should this be addressed in breeding?
In the task force's view, classifying chondrodystrophy as a general 'trait causing suffering' is inappropriate. In some breeds, over 90% of individuals carry one or even two copies of the chondrodystrophy allele. A complete exclusion from breeding of carriers of this allele is not sensible in the context described above; rather, it would result in numerous popular dog breeds no longer being able to be bred. This goes beyond a sensible approach to combating defect breeding practices and is counterproductive. Targeted genomic and clinical studies on sufficiently large populations of the respective dog breeds are necessary in order to classify the significance of this allele on a scientifically and clinically sound basis. A reduction in the risk of intervertebral disc herniation can already be achieved today in breeds with a predisposition to the condition through other means, for example via breeding selection based on X-ray examinations of the spine. In addition, a gradual reduction in the chondrodystrophy allele can be aimed for.

A radiological approach to reducing the risk of intervertebral disc disease can be used specifically for dachshunds, as is already practised in various northern European countries. The number of intervertebral disc calcifications visible on X-ray has proven to be an easily usable indicator for identifying animals that are highly likely to develop a herniated disc during their lifetime. In Dachshunds, excluding dogs with five or more calcified intervertebral discs has proven effective as a criterion for breeding decisions.

The appropriate time for a radiographic examination of the spine is between 24 and 48 months of age, as the number of calcified intervertebral discs is highest at this stage in radiological studies. Against this background, a single examination within this timeframe is recommended. Furthermore, there is a need for research to identify further relevant genetic and other factors influencing the occurrence of intervertebral disc herniation and to characterise more precisely the health implication.

Introduction
There are various genetic factors that influence the leg length of dogs. Two genetic variants that have been well studied in various studies regarding their morphological effects and their significance for health in dachshunds are two retro-insertions of the FGF4 gene on chromosome 12 and chromosome 18, respectively. The insertion on chromosome 12 is associated with a phenotype known as chondrodystrophy (CDDY). The phenotype associated with the insertion on chromosome 18

is often referred to as chondrodysplasia (CDPA) (Dickinson & Bannasch, 2020; Parker et al., 2009).

Both genetic variants influence the production of fibroblast growth factor 4 (FGF4), a protein that plays a key role in regulating cellular processes such as proliferation, migration and differentiation, and have been linked to reduced limb length in various studies (Bannasch et al., 2022; Brown et al., 2017; Parker et al., 2009). According to some authors, the reduced limb length confers selective advantages on carriers of these mutations when hunting in animal burrows and in confined spaces (Bannasch et al., 2022).

With regard to their health relevance, a distinction must be made between the aforementioned traits: whilst an increased risk of intervertebral disc calcifications and diseases has been described in connection with CDDY for some dog breeds or certain varieties of dog breeds, no corresponding health effects are known to result from CDPA (Dickinson & Bannasch, 2020). For this reason, based on the current state of knowledge regarding intervertebral disc health, the CDPA trait is not considered relevant by the members of the task force.

Whilst CDPA therefore only contributes to shorter limb length, CDDY has been linked to an increased risk of premature intervertebral disc calcification and disc herniation in various dog breeds and varieties (Batcher et al., 2019). In many carriers of the CDDY allele, radiographically calcified intervertebral discs can be visualised, and dogs with an increased number of radiographically visible intervertebral disc calcifications are at higher risk of intervertebral disc disease.

However, the health implications of the CDDY allele appear to vary between different dog breeds and even between different varieties within dog breeds (Bruun et al., 2020). There are many carriers of this allele who never develop intervertebral disc disease, suggesting that there must be additional genetic and/or exogenous factors that either prevent the disease or contribute to its onset.

Furthermore, environmental factors such as the level of physical activity and neutering status also play a role in the development of intervertebral disc disease (Packer et al., 2016; Dorn & Seath, 2018).

Prevalence

The underlying genetic variants are very old; the alleles for CDPA and CDDY occur in numerous dog breeds as well as in crossbreeds (Dickinson & Bannasch, 2020; Embersics et al., 2023). One or both of the aforementioned alleles are virtually fixed, for example, in Dachshunds, small terrier breeds (e.g. Jack Russell, Cairn, Norwich or West Highland White Terriers), Beagles, Havanese, Maltese, Bavarian Mountain Scenthounds and most Spaniel breeds.

The CDDY trait is widespread in many dog breeds and also in crossbreeds: according to Wisdom Panel data, 14.0% of all purebred dogs and 11.4% of all crossbreeds carry the underlying allele (Donner et al., 2023). In some breeds, the CDDY allele is largely fixed, meaning that wild-type alleles are now rarely found in representatives of the aforementioned breeds. A total of 14 breeds show an allele frequency of > 90%, meaning that, assuming an even distribution of alleles, more than 99% of dogs of these breeds are likely to carry at least one copy of the allele.

Breeding management of the trait

According to current knowledge, the CDPA trait does not influence the risk of intervertebral disc herniation; from the Task Force's perspective, the breeding of dogs with CDPA is therefore unproblematic from the point of view of intervertebral disc health.

When considering the breeding management of CDDY, it is important to distinguish whether scientific publications describe an increased incidence of intervertebral disc disease in the population in question. This is the case, for example, with the Dachshund breed, although it remains controversial to what extent the risk increases in the presence of one or two copies of the CDDY allele, and whether, in particular, the heterozygous presence of a single copy of the CDDY allele alone should already be regarded as a relevant risk indicator (Bruun et al., 2020, Sullivan et al., 2025). In any case, however, it should be noted that intervertebral disc disease is not a (monogenic) hereditary disease and therefore cannot be classified as such.

Furthermore, an immediate ban on breeding involving all carriers of the CDDY allele would, given the high allele frequency, have irreversible consequences for the breeding population and genetic diversity in numerous dog breeds; in many breeds, this would result in almost all dogs being

excluded from breeding, even if they do not suffer from any associated health problems. Such a restriction of the existing breeding population and gene pool must be viewed in a highly critical light from various perspectives (e.g. restriction of existing genetic diversity).

Against this background, a complete exclusion from breeding of CDDY carriers solely on the basis of their CDDY genotype appears neither objectively justified nor expedient. A practically feasible and expedient alternative is an X-ray examination of the spine, as is already practised in various Northern European countries and also within the German Dachshund Club.

As scientific studies have shown that most calcifications become visible from the age of 24 months and may even partially regress thereafter, a radiological examination after the dog has reached two years of age to detect existing calcifications appears sensible and is accordingly already recommended in various publications (Jensen & Arnberg, 2001, Lappalainen, 2014, Reunanen et al., 2023).

In Denmark, a threshold of five calcified intervertebral discs has proven effective for excluding dachshunds from breeding. This threshold is based on study data which indicate that a significant increase in the risk of intervertebral disc herniation is to be expected from five calcified intervertebral discs onwards (Lappalainen et al., 2014), and breeding programmes based on this threshold have already led to a reduction in the number of calcified intervertebral discs in Dachshunds in Denmark. The members of the task force support such selection strategies.

If a reduction in the CDDY allele frequency is deemed appropriate, this should not be carried out too rapidly where the initial allele frequency is high, in order to preserve genetic diversity. In dachshunds, for example, the initial aim could be to breed for heterozygous animals, as these already have a reduced number of calcified intervertebral discs compared to homozygous animals (Sullivan et al., 2025).

The exact approach to managing the CDDY allele in different dog breeds should be determined by taking into account allele and genotype frequencies and the extent to which these are actually associated with health effects in the breed.

The use of the CDDY genetic test is advisable and can help to identify dogs with favourable genotypes that may be of interest for breeding. Particularly in dog breeds with CDDY allele frequencies >50%, no dog should be excluded from breeding solely on the basis of the CDDY genotype. In the event of selection against the CDDY allele, care must be taken to ensure that dogs with favourable CDDY genotypes are not used excessively in breeding, in order to preserve genetic diversity as much as possible.

Regardless of the results of veterinary examinations, animals that have suffered a herniated disc should under no circumstances be used for breeding. A suitable breeding programme aimed at reducing the incidence of disc disease should be in place for all breeds in which an increased prevalence of such conditions has been reported.

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GESUNDE HUNDEZUCHT

The Healthy Dog Breeding Task Force is an independent association of experts in veterinary medicine and genetics. Its aim is to compile and provide practical, real-world applications of scientific knowledge relating to animal breeding, so that this information can be utilised and put into practice by veterinarians and breeders.

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