OSTEOCHONDRODYSPLASIAS, LEG DEFORMITIES, AND DWARFISM IN THE CANINE

Part Two

(Warning: Pictures are slow to download. Please be patient.)

By Fred Lanting

There has been renewed interest in the subject of "abnormal" bone lengths, joints, angles between limbs, and related phenotypic variations from what I have called "the ancestral type". We need to establish some definitions of terms before entering into a discussion of the subject. The "ancestral" phenotype in my arbitrary definition (which, however, is in line with the views of many or most professionals in animal science) is one that comes to mind when one thinks of the Jackal, Northern Wolf, and descendants of the extinct Pale-footed Wolf (such as sighthounds). The head is neither brachycephalic (pushed-in/shortened) nor exaggeratedly long and narrow (the dolichocephalic Borzoi, etc.), the leg length is such that the total height at withers is roughly twice the distance from elbow or chest to the ground, and limbs give an impression of being straight. Typical examples of ancestral types are the German Shepherd Dog, Saluki, various Spitz breeds, and many pariah breeds such as found in every corner of the world.

"Abnormal" phenotypes (and this will rouse the ire of many people who love their dogs and think of them as being "normal") include breeds specifically bred to produce the characteristics that would be agreed on as being "faults" in the ancestral types. Think of the ("English") Bulldog, Pekingese, Corgis, Dachshunds/Teckels, and others. I have long maintained that there is a genetic defect affecting primarily the hypophysis or pituitary gland, the "master gland" that so greatly influences the functions of the others as well as developmental processes.

Some variation within normal parameters results from the tremendous plasticity of the canine genotype, but here we are more interested in the departure from those limits of normality. Whatever the combinations of defective DNA nucleotide pairs (adenine-thymine, cytosine-guanine, etc.), and which glands or organs they initiate the changes in, many of the irregularities we are discussing here manifest themselves in the cartilage that is on the ends of bones and "bone centers". Bone centers are those hard, mineralized portions of a growing bone that become enlarged (almost entirely on their long-axis surfaces) and fused together to form the eventual limb, and the cartilage between most bone centers is called a "growth plate" or physis. It gradually "calcifies" into bone tissue, thus uniting epiphysis (called a cap or head, usually) and metaphysis (shaft); it disappears during maturation. But if there is an abnormal coding of the nucleotide pairs, there is an abnormal calcification process, a "growth-plate disturbance".

If one bone in a two-bone limb segment (such as the tibia/fibula or ulna/radius combinations) has more of a disturbance than the other, or if one end of the growing bone's cartilage is disrupted during remodeling into bone tissue, there are unequal rates of growth and consequent bowing of that limb, with one part wanting to be longer than the other part does. In some dogs, disruption of normal cartilage-bone turnover at the ends can keep a single bone from growing in length, or if the disruption is laterally asymmetrical, the femur or humerus may also become slightly bowed. The pull of muscles and ligaments on different parts of such bones also has some effect on shape.

The general definition of "dysplasia" is poor or abnormal (*dys-*) shape or form (*-plasia*). Here, we are talking mostly about bone (*osteo*) and cartilage (*chondro-*), and mostly about those tissues in joints. But just as the poet says, "no man is an island", and genes that cause one thing can sometimes also cause something else. Some times it is a very obvious double influence, such as the gene that causes both deafness and white coat phenotypes in some breeds, or dwarfism and blood disorder in the Malamute. Most of the time, the influence of one gene or set of nucleotides is less obvious.

A couple more definitions would be helpful at this point, although you should realize that there are sometimes loose adherence to strict interpretation of such definitions:

- **chondrodysplasia:** any growth plate (cartilage) disturbance resulting in canine dwarfism; in human pathology, it has a different meaning: enchondromatosis, a rare disorder marked by enlarged cartilage and tumors in joints.
  - **chondrodystrophic:** semantically similar to the above, but while *–plasia* refers to changeable shape, *–trophic* refers to growth. Thus, an abnormal cartilage growth pattern.
    - **achondroplasia:** that type which results in an individual with extremities shorter than the trunk. Examples in dogs include Basset Hound, Shih-Tsu, and others mentioned elsewhere. In humans, it usually is marked by stubby hands, large head with sunken nasal bridge and, frequently, spinal column deformities.

CHONDRODYSTROPHY

Several breeds are of a body type we call chondrodystrophic, such as the Dachshund and Corgi. They have shorter legs (often bowed) and other dwarf characteristics in parts of the body. Frequently, these breeds also have a shorter vertebral arch that tends to produce a smaller vertebral canal. The vertebral body centers of ossification unite with the arch prematurely, with the same type of dystrophic bone growth pattern that causes shorter "long bones" in those breeds. See Chapter 16 in my 2004 orthopedics book for more illustrations and discussion on dwarfism. In some chondrodystrophoid breeds such as the Basset, a premature closure of the distal metaphyseal plate of the ulna (near the wrist) was thought by Herron, Grüll, Henschel, and von Hitz to cause fracture of an already closed anconeal process at the other end of that limb. Kasström and colleagues (and later, Wind) thought that this condition in certain dwarfed breeds "was the result of an abnormal pressure on the anconeal process... by the shortened ulna." This anomaly in the anconeal process is not the same as the failure to unite, and obviously has a different genetic origin. Dr. Wind, the eminent expert on elbows, has observed that many cases of elbow dysplasias include subluxations associated with dwarfism

There are many types of dwarfing related to slow endochondral bone formation (at the ends). Dwarfism can be proportionate or disproportionate, depending on the specific gene defects. Examples of the former include the pathological pituitary dwarfism of the German Shepherd Dog and the related Karelian Bear Dog. You can see GSD pituitary dwarfism in "The Total German Shepherd Dog" (www.Hoflin.com). The non-pathologic selective miniaturization seen in Shar-Pei, Bull Terriers, Australian Shepherds, Poodles, and numerous other miniature and toy breeds and varieties probably should not be included in a discussion of dwarfism. Disproportionate dwarfs include Bulldogs, Basset Hounds, Pekingese, Dachshunds, Corgis, and many more that we see only in this form. While some would object to inclusion of their favorite breed here, it is still true that these are results of genetic defects. There are also "unnatural" occurrences of disproportionate dwarfism in breeds where you might not expect it: Malamutes, GSDs, and a few other "accidentals". Of course, if one were to deliberately linebreed on these defects, a sub-population of short-legged representatives would be more common. That is what happened with a branch of the Parson Jack Russell Terrier, now known by several similar names in various show registries.

Some of these defects involve irregularities in the construction of the hypophysis (pituitary gland), as I have said; others may involve primary proteoglycans degradation that results in mucopolysaccharidoses, or other metabolic abnormalities. The various types of mucopolysaccharidoses involve enzyme deficiencies, incomplete fusion of the sacrum, incompletely developed vertebral end plates, short limbs, abnormal joint mobility, and other signs and deformities. Some osteochondrodysplasias (you now know how to break that word into segments, and what they mean) can be recognized at birth, others not until skeletal maturity approaches. The most common

seen at birth is the achondroplasias of rabbits, mice, and humans, although some (erroneously?) apply that term to conditions in dogs, too. Some achondroplasia is from sporadic mutation, and most seem to be transmitted by a dominant gene. A few authorities have claimed that achondroplasia is not seen in the canine, but Aegerter and Kirkpatrick describe it as a genetic chondroblast (cartilage cell) disturbance in the epiphysis. Betts calls it "a symptom rather than a disease" and does not hesitate to apply the word to the "normal" condition seen in the Dachshund, Beagle, Basset Hound, French Bulldog, Pekingese, and similar breeds. He properly excludes pituitary dwarfs, miniaturized but proportionate breeds, Malamute dwarfism, and the dwarfism of Labs with retinal dysplasia. Various forms of chondrodysplasia affect Cocker Spaniels and German Shorthaired Pointers.

Miniature Poodles are occasionally found with a form of osteochondrodysplasia that has sometimes but properly erroneously referred to as "achondrodysplasia". An increased collagen concentration and RNA content is often found in affected cartilage of such dwarfs, though DNA content is normal. There are differences in appearance between individuals because of modifier genes as well as environmental forces. Miniature Poodles with inherited epiphyseal chondrodysplasia are rhizomelic (it seems the shortness of their limbs comes primarily from the retardation of growth nearest the hips and shoulders) and often have ventrodorsal compression of the chest and enlarged joints. Occasionally a spontaneous mutation will produce symptoms similar to congenital spondyloepiphyseal dysplasia and "achondrodysplasia", and variations on these are seen in many breeds, including multiple epiphyseal dysplasia in Miniature Poodles and Beagles.

Asynchronous (unequal) growth of the radius and ulna, that is, when these two bones' growth rates are not coordinated, is found in non-dwarfs in a wide continuous "spectrum" of severity, often unnoticed by the average dog observer (and many a licensed dog-show judge!). Dwarfism is often an accompaniment to that asynchronous growth of the "double bones" in either front or rear limbs. Such dogs also have nearby limb segments shortened, such as the humerus or femur. The more extreme end of that range is considered by some to be "normal" in some breeds (to name some more: Corgis, Basset, Lhasa Apso, and Pekingese). However, elbow subluxation accompanied by pain has been reported in these dwarf breeds, and probably has a causal connection. While carpal valgus (turning out at the pasterns) and external rotation of the foot are "acceptable" within the descriptions of "breed type", the occasional or perhaps frequent abnormalities of the ligaments and joints in the elbow that accompany this should be avoided or treated.

OTHER SIGNS

Chondrodysplasia in the most general sense is sometimes called a syndrome, other times *part* of a syndrome, the definition of that word being a collection of interconnected symptoms. Some dogs may have many, some a few, and others no readily observable symptoms. The clinical signs may be very mild, from almost undetectable bowing or shortening of the legs, to obvious skeletal deformity and the presence of several health problems. *Chondrodysplasia Punctata* is one name applied to a syndrome of multi-systemic disorders, and is so-called because of the "dots" of calcium phosphate deposits in the softer cartilage. It reminds me of the school of art made popular by Georges Seurat called "Pointillism". This genetic-metabolic problem has various skeletal expressions. Depending on the particular variety, the mode of inheritance could be autosomal recessive or dominant, or X-chromosome-linked recessive or dominant, some with full penetrance, and some not.

Besides skeletal indications, there are eye disorders such as microphthalmia (smaller eyes than they should be), lens detachments, cataracts, glaucoma, retinal defects, and nystagmus (jerking or twitching of the eyeballs). Other occasionally reported symptoms are problems with internal organs, head and neck bone defects, partial deafness, alopecia, and luxated patellas (for more on this stifle problem, see my upcoming orthopedics book or some of the websites that carry my articles).

PREMATURE PHYSIS CLOSURE

Premature closure of growth plates happens because, in some etiology (manner), the ossification process of

endochondral cartilage is disturbed. Overfeeding and mineral supplementation are definitely contributors, but genetic susceptibility has to be taken into account, as well — probably much more. Ettinger mentions that "the most common cause of premature growth interference has been direct trauma to the growth plate area", though HOD and achondroplasia have also been reported in association with it. But he and his sources may have been giving too much credit/blame to physical or mechanical damage. The distal (furthest part) radius and ulna seem to be the most frequently involved sites for these disturbances.

Growth disturbances in the radius and ulna can be related to an outward twisting of the top of the ulna away from a good fit with the humeral condyles, enough so that subluxation or even luxation takes place. This lateral rotation may also exist independently, with no *observed* growth plate disruptions in those bones. The radius head can also dislocate, and both may occur at the same time, so there is quite a variety of changes possible, although the disorder is rare.

If the dislocations are not accompanied by (or secondary to) such asynchronous growth manifestations as seen in the ulna and/or radius, they are called congenital elbow dislocations. The premature closure of physes in ulna or radius, retained cartilage, chondrodysplasia or achondrodysplasia, and synostosis are separate problems. A condition of missing digits called ectrodactyly and another abnormality called cleft hand deformity have been seen in conjunction with congenital luxations and subluxations (also called arthrodysplasia) in the elbow.

ROOTS OF MANY OF THESE DEFECTS

Cholesterol has had a bad name among fad-diet promoters and people too lazy or busy to physically work off their calories. It is a product of the liver, necessary for the synthesis of Vitamin D as well as the assimilation of it, essential fatty acids, and Vitamins A, E, and K, but in sedentary people and those with genetic inability to metabolize it correctly, it can build up in the blood vessels and contribute to heart disease and stroke risk. At least some chondrodysplasias involve an error in the coding for biosynthesis of cholesterol. Since in the Havanese, a miniature Cuban breed, those who evidenced this dwarfism tested as having abnormal levels of several cholesterol-related sterols, a program of blood serum testing was undertaken. It was found that Havanese with normal straight legs had no such metabolic abnormality. in the body. These vitamins are needed for calcium utilization, bone development, and healthy eyes. The appearance of congenital defects, including osteochondrodysplasias, can often be blamed on inability to use these chemicals. Tracing a structural defect to its headwaters of a genetic defect expressed in a metabolic disorder along the route, is akin to finding the source of the Nile or Amazon.

PLEIOTROPIC DEFECTS

Pleiotropy is the phenomenon of having more than one phenotypic expression (often in grossly different manifestations) caused by the same gene — the same genetic defect. Alaskan Malamutes' dwarfism is a pleiotropic genetic defect that shows up as *both* dwarfism of their particular type *and* a blood disorder. It has been fairly extensively studied, and while one dog may vary in appearance considerably from the other, the disorder is a simple autosomal (not sex-linked) recessive trait with complete penetrance. Asynchronous growth of the radius and ulna (one at a different rate or completion than the other, remember) is part of the deformity in this breed. The chondrodysplasia in this breed has at times been mistaken for the Vitamin D deficiency called rickets, but only the tubular bones are affected, other than retarded ossification of the lateral tarsal (cuboid) bone. The head, spine, and other bones are not stunted or changed, and body length is normal. The gene that causes this chondrodysplasia also creates a macrocytic hypochromic anemia; the discovery of this being indicative of the way carriers may be found. A third effect of this one gene, by the way, is a different ability to bind certain trace minerals in the liver.

While on a judging assignment in Alaska in the early 1980s, I was presented with a Malamute from show lines, which had from an early age walked flat on its wrists. Because the forearm did not appear bowed I initially

thought it might have been a case of carpal luxation syndrome that I had been seeing with increasing frequency in American German Shepherd Dogs. I lost track of the owner and did not see any radiographs or blood analysis. Later, after seeing more Malamute Chondrodysplasia, I reconsidered my tentative "diagnosis". I think now that it could possibly have been both disorders occurring concurrently in the same dog, but more likely the carpal luxation was a result of the chondrodysplasia gene. Unfortunately my photographs of that dog were lost, but I later obtained pictures of other Malamutes, though without the extreme flat carpus.

A few other problems are similar, in that one gene (or gene pair, really) can cause ocular-skeletal dysplasia in Labrador Retrievers and possibly Samoyeds, for example. In this disorder, several defects in the eyeball, iris, and arteries serving the eyes are found in the same dogs that have short, thick leg bones (micromelia), prominent carpi (wrists) and elbows, and east-west stance in front. Hind legs usually are hyperextended (straight in stifle) yet still very short.

Great Pyrenees have their own style of micromelic dwarfism, too, as do a few other breeds. It is a simple recessive genetic trait, showing some similarities to Malamute dwarfism, and is marked by short curved ribs, underdeveloped rear limbs, all legs shortened, and abnormalities in the cartilage and bone of the vertebrae. Endochondral ossification disturbance can usually be seen on radiographs by 8 weeks. Often, ossification of the vertebral bodies, especially in the neck, is delayed right from the beginning, and visible on radiographs taken at 8 weeks of age. The metaphyses of the radius, ulna, and tibia are usually flared like the bell-bottom trousers of the hippies in the early 1970s or the sailors of a generation earlier. The condition does not automatically result in DJD (degenerative joint disease).

Norwegian Elkhound chondrodysplasia is similar to the other canine dwarfisms as well as to human spondylometaphyseal dysplasia; it is widespread in the breed, and may be associated with glycosuria (sugar in the blood), although in one study this was not found. Some curvature of the front legs may be noticed as early as 5 weeks of age, and all limbs are short in proportion to the body. It is also a simple recessive trait.

A disorder almost identical to the chondrodysplasia in two of the above-named breeds has recently been found in Akitas. Knowing how such reports usually lead to the identification of the same disease in other breeds (as has happened in panosteitis, GSD myelopathy, etc.), it is not very risky to predict that more will be added to this list in the future, though not at a high rate, given the very obvious nature of dwarfism and most breeders' desires to sweep it under the rug or eliminate it.

On an excellent website called Rhosyn German Shepherd Dogs, there was a good description and illustrations of dwarfism in the Havanese breed, and an ancillary discussion of dwarfism in the GSD, even though the site owner did not want to use that word for the condition. Havanese with dwarfism display ocular abnormalities, as do a few other breeds. The front legs grow crooked or bowed, and all four legs are shortened, giving the height-to-length ratio an undesirably short aspect. Havanese breeders have reported that all cases of early-onset cataracts leading to premature blindness, and nearly all "other serious health problems reported in Havanese within the past few years, have been in dogs that also exhibit the symptoms [of chondrodysplasia]". In Havanese, it also has been noted that some dogs have such subtle signs that they appear to have a straight leg on one side but not the other — asymmetric. Furthermore, that such asymmetrical dogs, if they are also diagnosed as having cataracts, will have the cataract in the eye that is on the same side as the crooked leg! The *Rhosyn* website mentioned above says, "To date, no Havanese with straight legs have been diagnosed with early onset cataracts!"

Many other dysplasias considered as a subcategory under osteochondrodysplasias result from disturbed ossification along the periphery (outer edge) of the growth plates in various bones such as the ribs, vertebral processes, skull, and elsewhere. Certain dwarfism characteristics have been made part of the breeds' show standards and are not much covered here, but even some breeds that are not normal skeletally, anyway, such as Dachshunds and French Bulldogs, sometimes are even more afflicted with chondrodysplasia than their

artificially-considered "normal" compatriots. In many cases, dwarfisms with partial penetrance or expression may go unrecognized, with the breeder considering the mildly affected pup to be simply a "runt".

Recently, some cases of chondrodysplasia/chondrodystrophy of the sort found in the Corgi have been reported in German Shepherd Dogs in Australia. The German Shepherd Dog Council of Australia website, http://www.gsdcouncilaustralia.org.au , has some more details on the investigation there. Some of the bitches whelping such dwarfs are daughters of popular showdogs such as Hammer v Waterkant, Lindendale Strike Force, Leitungen Prince Rowdy, and the highly-respected German export Iwan v Lechtal. Cases have been reported all across the country: in Canberra, South Australia, New South Wales and Western Australia. One cryptic comment from Downunder was, "The common denominator in all cases has been the Stud Dog." I found this dog is Aimsway Abacus, a son of German import Balou v Eppelin and a local-bred bitch, Rakishka Ali; Abacus linebreeding is: Eiko-Vasall Kirschental (5-5). Most people are reluctant to `fess up to genetic problems for fear of losing face — or stud fees. Fortunately, there is a move for openness in Australia concerning this appearance of achondroplastic dwarf GSDs, even though at this time it appears the incidence is considerably less than the incidence of pituitary (proportionate) dwarfism in the GSD that I reported on several years ago. Some examples of the latter are shown in my GSD book.

Illustrations either accompany this article or are available.

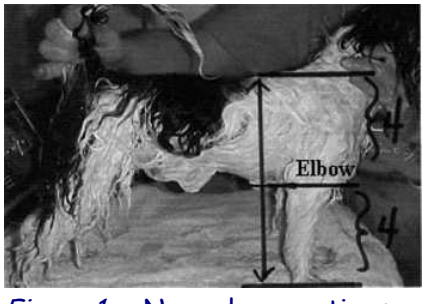
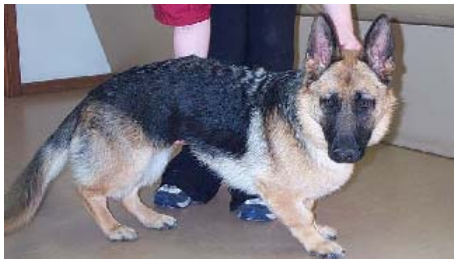


Figure 1. Normal proportions and leg length in a Havanese, with about as much chest depth as the distance from elbow to floor.

Figure 2. Chondrodysplastic dwarf "Hav" with bottom of chest well below the elbow, & with shortened limbs.

Fig. 3 Littermates Angel (chondrodysplastic dwarf) and Athello (normal leg length)

Fig. 4 "Dusty", a chondrodysplastic GSD at 12 weeks and at 9 months.



Credits: photographs graciously supplied by Rhosyn kennels in Australia.

NOTE: A well-respected AKC and Schaferhund Verein judge, Mr. Lanting has judged in more than a dozen countries, including the prestigious FCI Asian Show hosted by Japan Kennel Club, the Scottish Kennel Club, a Greyhound specialty in England, and more. National Specialties: 1994 GSD Club of America National; 1991 Tibetan Mastiff National; 1990 Shiba National; Fila Brasileiro Nationals (several times), Dogo Argentino National, Pyrenean Shepherd National. Numerous Chinese Shar Pei and Australian Shepherd specialties; regional Anatolian Shepherd specialty. Numerous GSD, Rottweiler, & Boxer specialties worldwide. He is also the author of several 'must read' books, including THE TOTAL GERMAN SHEPHERD DOG, CANINE HIP DYSPLASIA, CANINE ORTHOPEDIC PROBLEMS. A former professional all-breed handler in the US and Canada, he has lectured in over fifteen countries on Gait-and-Structure (Analytical Approach), Canine Orthopedic Disorders, and other topics, as well as being a Sr. Conf. Judges Ass'n (SCJA) Institute instructor. WV Canine College instructor & member, advisory board. His full Curriculum Vitae is very impressive and we are grateful to him for sharing that knowledge on this site. Join him in his tours to the SV shows in Germany.