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ACHONDROPLASTIC DWARFS IN CATTLE

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The steady increase in the incidence of dwarfism in the beef-cattle herds of this country in the past thirty years and since an original report on the subject in 1928 (1) has become a major cause of economic loss and a matter of grave concern to cattle-breeders. This, plus its possibilities as a source of genetic, physiological, embryological and endocrine information, makes it of intriguing interest to the biologist.

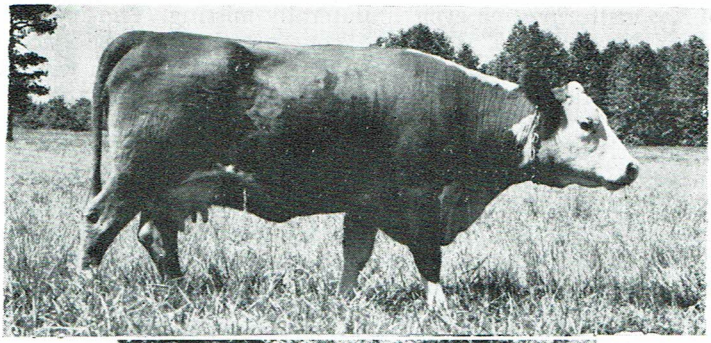
Since the first report, the condition has been called to our attention at increasingly frequent intervals, and has been the object of further studies. Within the past two years, at Tennessee Polytechnic Institute, it has been our good fortune to have a number of animals made available to us for study, and it is hoped that channels have been opened up for obtaining a larger number in the near future. The majority of our animals are of known pedigreed stock. Most of those that are included in the present study are Herefords, although the condition appears in other beef-breeds as well, notably the Aberdeen-Angus (2, 3). It occasionally appears in varying degree as an apparently isolated mutation in dairy-breeds (1, 7) as it does in many vertebrate forms, other than cattle. Most of ours' have been sacrificed and autopsied carefully, with a number of interesting findings. Our data are drawn from twelve such complete autopsies, with partial ones on three others and seven "normals" of comparable ages. Our animals range from two weeks to six months in age. Although these animals are usually sterile, a few older ones of both breeds are being fostered with the hope that a breeding-herd can be built for more detailed genetic studies. A search of the literature has been done on this condition in cattle, and on related conditions in these and other vertebrate forms, from the genetic, anatomical and physiological (especially endocrine) angles.

Our observations may be classified as follows:

1. *GROSS ANATOMY*

At one extreme is the "Dexter Monster" or "Bull Dog" type reported earlier (1, 5) with short achondroplastic legs, complete cleft palate, extremely marked prognathism, large and flabby tongue, decided under-development in the upper nasal and orbital regions, (with a consequent apparent "bulging" in the frontal region), shortening of the vertebral column, with the

tail placed high on the back, "pot-belly" and numerous variations in the internal organs. The most advanced of these cases are born dead or do not live more than a few hours. At the opposite extreme and much more frequent in occurrence, is an animal notably smaller but lacking most of the above stigmata, generally classified as a "snorter" but frequently difficult to diagnose at an early age, yet definitely a dwarf, having some of the facial features, pot-belly and unsteadiness in gait, and des-



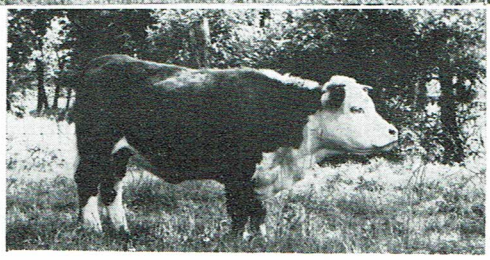


Figure 1, above Figure 2, below

1. Normal Hereford Cow — 27 months old
2. "Dwarf" Hereford Cow — of closely related breeding 27 months old
Pictures taken at identical distances.
Note difference in proportions and facial profile.

tined never to attain normal size. Measurements of the members of our present experimental herd are a good indication. One, a female, is 27 mos. old, weighs 480 pounds, is 35 inches tall at the withers (Fig. #1) and measures 54 inches at the heart-girth. Another, a male, is 7 mos. old, weight 160 pounds, is 23 inches tall, and measures 38 inches at heart-girth (Fig. #2). Both of these have typical stigmata in varying degree. Between the two extremes, one finds a wide range of variations.

Internally, at the one extreme, nearly all viscera are varyingly deformed. A very common variation is marked maldevelopment of both gonads, and frequently a complete absence of one of them, adrenals missing, or so underdeveloped and misplaced as to be impossible to find, with essentially the same true of the pancreas. The thymus is usually abnormally large, so much so as to seem almost to replace the thyroid. This mass occasionally extends upward to the submaxillary and sublingual glands, with only very little or possible no thyroid tissue to be found. This condition has been noted by others (8). Kidneys are frequently malformed or even unilaterally missing. The intestinal tract is "over-loaded" with the contents showing evidence of putrefaction and having a foul odor. This is in accord with the unpleasant odor of the feces from these animals, and the polyphagia and "pot-belly" which is constant, and which frequently is diagnostic of Dwarfism. (One wonders if the under-developed pancreas is not responsible). Polydipsia or polyuria are also usual. This is in line with findings in pituitary (chromophobe) adenoma in humans (9) and also has been attributed to malfunction of the hypothalamus. A generalized "catarrhal" condition of the respiratory tract is usual, with consequent difficulty in breathing, and gives origin to the name "snorters" by which many breeders know the less deformed animals.

The skeletal system shows an equally variable picture. In the more extreme cases, the long bones are greatly shortened, and at the epiphyses there is faulty and irregular calcification, and in some instances ,a marked overgrowth of partially-calcified cartilage. The bones of the skull, and particularly the basicranium, are markedly variable in size, shape and relationships, but in general, are greatly thickened with premature closure of the sutures, especially the spheno-occipital, and many malformations.

Dental development is precocious with teeth loose in their sockets. The skin is loose, baggy, wrinkled, but leathery. Many variations of these conditions exist in different animals.

2. *HISTOLOGY*

There are indications that the pars distalis of the pituitary is increased in total volume, although in the case first cited (1), and in one of our present series, the entire hypophysis was missing or so small and misplaced in the malformed basicranium, as not to be found. There is a decrease in number of chromophobes and basophils, with the appearance of a large number of finely granular cells of a type somewhat between the two normal cell types. Acidophils are extremely rare, occasionally missing entirely.

In those animals where thyroid tissue is located, the follicles show a vacuolated appearance at the periphery and many un-

differentiated acinus cells are present. The thymus is microscopically normal.

The adrenals are usually lobulated, so that valid study of their architecture is very difficult, but one gets the strong impression of a lessened "cortex-to-medulla" ratio, with the cortical cells not grouped in the usual fairly definite fashion, and, with a washed-out appearance of their cytoplasm.

The pancreas is found usually only in small masses scattered through the peritoneal fat, and appears to have a comparatively less amount of islet tissue than is normal in cattle.

Sex-glands are greatly retarded, and frequently malformed. Even in older animals, mitoses in spermatogonia are very rarely seen, and in some cases not seen at all. Tubules are small and frequently fille dwith a syncytial fluid. Interstitial tissue is comparatively increased.

Kidneys are malformed, and seem to have more and larger sinusoids.

3. *GENETICS*

That Dwarfism or chondrodystrophy is merely a simple recessive character seems open to doubt. This may apply to a basic character, although occasionally it may mutate to a dominant (7). However, the very great number of forms in which the condition appears would seem to indicate that there is a considerable number of modifying genes, or inhibiting factors, or both. Our cases are too few in number to indicate an exact pattern of genetic behavior ,but it seems quite certain that it is an extremely complicated type of inheritance. Both qualitative and quantitative differences, as well as the lethal or sub-lethal effect occasionally seen, are probably the result of the inter-action of a number of genes. It is apparent, that once the character appears as a mutation, it is apt to appear in a wide variety of forms in subsequent generations.

4. *COMPARISON WITH FINDINGS IN THE LITERATURE*

Our studies do not differ greatly from Crew's reports, but are being carried further. He dealt with the "Dexter" breed, an off-shoot of the Kerry, a short, compact, very short-legged animal, as is also true of our modern beef-breeds. He suggested as did Stockard with dogs, that breeding such a type was, in fact, breeding *toward* achondroplasia. We concur in that suggestion.

Stockard (10) in his classical and definitive study of chondrodystrophy in dogs, found greatly varying changes in these animals, and concluded that the genetic causes were extremely complex, especially when the axial skeleton was involved. In cattle, the spinal column and skull of these dwarfs are invariably affected, and follow a pattern very similar to that found by Stockard in dogs. The endocrine changes apparently present in cattle

also seem to parallel closely with those observed by him in dogs. Stockard also found that the shorter and more compact the "type" the greater was the likelihood of mutation in the direction of anchondroplasia. This, also, is true in cattle.

In the last few years many studies have been published on achondroplasia in beef-cattle. Gregory and his co-workers (2, 11), Lush and Hazel (12, 13), Baker and his associates (4) and especially the excellent and complete study of Fransen (6) are among many who have contributed to our knowledge of this complex subject.

The anatomical and physiological findings are in general agreement, but genetic and endocrine interpretations vary considerably. There is ample evidence that the more common type of "snorter" dwarf is basically a recessive, as most studies indicate a 1:2:1 Mendelian ratio. This probably is not true, however, of the wide variety of more advanced deformities, which do not seem to behave in any regular fashion, possibly because their occurrence is so sporadic.

The condition has been attributed to the anterior lobe of the pituitary or, more frequently, to malfunctioning thyroid, occasionally to the thymus. The fact that the pituitary picture varies histologically from an essentially normal one, to cases where the gland cannot even be found, throws doubt on this mechanism. The same is true of the thyroid-thymus relationship, which varies from an apparently normal picture to an extreme where the thymus is tremendously over-grown and the thyroid is apparently missing. This wide range of difference appears in animals not remarkably different in size or appearance, or in blood-picture.

5. *TENTATIVE CONCLUSIONS*

Dwarfism in cattle has many points of similarity to achondroplasia concurrent with dystrophia adiposo-genitalis, and is closely similar to these conditions, found spontaneously in numerous other vertebrate forms. Some of the changes found here have been produced experimentally by extirpation of the pituitary. In several ways, the conditions seen are very similar to change sseen clinically in pituitary chromophobe adenomata in humans. In other ways there is little similarity.

There seems to be little doubt that achondroplastic dwarfism in cattle as in dogs, in rabbits, in sheep, in mice, in fowls and even in humans, is of genetic origin. We suggest that it is transmitted by a recessive character, influenced markedly, however, by a number of interacting determiners which influence the time and penetrance of initial impact during embryonic life, influences which both affect range an dintensity of effect, even to the extent of lethal results.

It is our opinion that the changes seen in the glands of internal secretion in these conditions, is effect, rather than cause, and that some little-understood homeostatic mechanism tends to compensate for lost or diminished hormones. True understanding will probably require extensive studies on the embryology of dwarfs and lethal vertebrate characters.

Further studies will be pursued, as suitable materials are made available to us.

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