A PRELIMINARY NOTE UPON THE OCCURRENCE OF INCOMPLETE DEVELOPMENT OF THE STRIATED MUSCLE FIBRE AS A CAUSE OF CERTAIN CONGENITAL DEFORMITIES OF THE EXTREMITIES.

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THE unsatisfactory nature of the current theories that are held to account for congenital distortions of the limbs, such as talipes, contractures of joints, and congenital high scapula, has for long been recognised. The subject is of interest to general surgeon and orthopœdist alike, and any well-attested observation bearing on the question deserves consideration.

It has been realised by many observers that certain congenital deformities of the limbs may be due to shortening of individual muscles or groups of muscles. Attention was frequently directed to this possibility by the older writers on congenital club-foot; and the presence of a shortened muscular or fibrous band uniting the scapula to the skull or to the vertebral column has been a constant finding in all dissections of congenital high scapula. More recently attention has been drawn to the theory that defective growth of the affected muscles might be due to failure in the ordinary sequence of development of striated muscle, incomplete development in a given muscle entailing defective growth and function. Potel,[1] who suggests this possibility, fails, however, to produce convincing evidence in favour of the theory, and no definite microscopic proof of arrest of development of striated muscle has up till now been forthcoming.

Several cases of the deformity known as "arthrogryposis multiplex congenita" have come under the writer's observation. This defect consists of the presence of bilateral club-foot and bilateral club-hand without bony malformation. The deformities are associated with decided limitation of mobility at the joints of the affected limbs. The upper, the lower, or all the limbs may be affected, but the deformity is always bilateral and symmetrical. When the lower extremities are affected, there is, in the majority of cases, a bilateral congenital dislocation at the hip-joints, which is peculiarly resistant to treatment.

Anatomical observation shows that the joints themselves are normal, and that the limitation of mobility, which is the outstanding feature, is due apparently to want of elasticity of the muscles acting over the joints, the muscles appearing wasted, inelastic, and often more or less contracted.

It is significant that an analogous condition occurs as a lethal hereditary deformity in lambs, and has been closely studied from the point of view of its Mendelian hereditary characters by Fraser Roberts.[2] Pathological investigations showed that the muscles of the affected limbs appeared atrophied and contracted to the naked eye, but when the muscles were removed by dissection, the joints and their capsules appeared normal and possessed a full range of movement. By the courtesy of Mr Millar of the Institute of Animal Genetics, portions of the affected muscles have from time to time been made available for histological examination, and it was easily shown that they were composed very largely of undifferentiated connective tissue with a considerable degree of fatty change, profusely studded with round or oval cells, each containing a central nucleus, the cytoplasm of which exhibited the staining characteristics of myoplasm. Reference to the appearance of developing striated muscle makes it clear that these cells represent primitive muscle cells such as are met with normally prior to the third month of intra-uterine life. Such an embryonic cell, which is capable of producing one striated muscle fibre, may be referred to as a "myoblast."

A few specimens of muscle have been removed by biopsy from children suffering from arthrogryposis, and, though the advanced stage met with in lambs has not been completely reproduced in the human muscles, there can be no doubt that the muscles are similarly affected. In some cases the muscle has shown large tracts of connective tissue, and there is good reason to believe that, if the muscles concerned had been subjected to examination at birth, instead of some years after, embryonic myoblastic tissue would have been present in these strands. It is not to be expected that such embryonic tissue, incapable of advancing towards perfect development and not in receipt of normal stimuli, would remain unchanged: the tendency must be towards early degeneration, and this is brought about in muscle by a fatty change in the functionless cells, which progresses rapidly after intra-uterine life. In other sections of the affected muscles, areas have been found in which

muscle fibres, varying greatly in diameter, were completely lacking in striation, either longitudinal or transverse.

The subject was advanced a stage further when a case of apparent absence of the neck muscles on both sides came under the writer's observation. Permission for a biopsy was obtained, and the lower part of one sternomastoid was excised. The muscle was found to be represented by a thin strand of pinkish tissue which on microscopic examination was seen to be composed almost wholly of embryonic myoblast cells.

The main features of the well-known deformity of congenital angling of the tibia consist of a fixed pes equinus and an anterior angling of the tibia at the junction of its lower and middle thirds. On dissection of a fœtus showing this congenital peculiarity it was found that excision of the calf muscles liberated the calcaneus, so that the equinus deformity at once disappeared, suggesting that congenital failure in the growth of the calf muscles had, by their consequent shortening, initiated the equinus deformity, and, when plantar flexion at the ankle-joint had reached its limit, had caused the cartilaginous shaft of the tibia to bend at its weakest part. Unfortunately, the microscopic findings in this case were inconclusive, as the tissue showed too much post-mortem change to allow of an adequate histological study.

Finally, a case of bilateral congenital high scapula was met with in an infant who died of inanition at the age of seven weeks. Clinical examination had shown, associated with a marked degree of high scapula on both sides, a gap in the occipital bone of the unclosed tectal plate variety. A band of muscle could be felt running from each scapula up to the occipital bone on either side of the bony gap. Dissection showed that this muscular band consisted of a portion of the trapezius muscle, the remainder of the trapezius being absent, and possibly an abnormal levator anguli scapulæ and rhomboid muscles. The upper end of the band was attached to the superior nuchal line of the occipital bone, and the conclusion seemed irresistible that the muscular bands had not only held up the scapulæ, but had also prevented the tectal plates of the occipital bone from closing to form the foramen magnum in the normal manner. Histological studies of the band showed the muscle bundles to be separated by wide swathes of connective tissue, and, scattered in fair profusion amongst this tissue, masses of primitive myoblast cells. At the same

time degenerative changes were seen to be affecting some of the formed muscle fibres at the edge of the fibrous swathes.

It is suggested that such deformities as those investigated result from the failure of certain striated muscles or parts of muscles to undergo their complete course of development, with the result that they remain less elastic, do not grow in length in proportion to the other structures in the limb, and never assume their proper contractility. These abiotrophic muscles must therefore be looked upon as constituting the primary ætiological factor in the production of these deformities.

REFERENCES.—[1] Potel, G., *Traité Pratique d'Orthopedie*, Paris, 1925, Doin. [2] Roberts, F. A. Fraser, "The Inheritance of a Lethal Muscle Contracture in the Sheep," *Journ. of Genetics*, April 1929, vol. xxi.