Central Core Disease:  
Histochemical and Ultrastructural Study of Muscle Biopsies of Father and Daughter

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Summary. Two cases of central core disease, father and daughter, of a family with dominant autosomal inheritance, are presented, one with bilateral congenital dislocation of the hip. Muscle biopsy was performed in both cases. Oxidative enzymes evidenced only type I fibers, most of them presenting a central core and not uncommonly more than one. On electron microscopy the cores generally appeared well demarcated from the surrounding fibrils and were characterized by lack of mitochondria and abnormalities of the Z line. Transitional aspects from normal fibers to completely unstructured cores were observed, as well as from well structured and unstructured cores. These findings are discussed in the light of the previous literature and particular attention is paid to the problem of differentiation between central core and multicore disease. The pathogenesis of the muscular alteration is also discussed in relation with the possibility of their neurogenic origin. Eventually, the histochemical and ultrastructural similarities between central cores and target fibers are focused.

Key word: Central core disease.

seits beobachtet, wie auch Übergänge von gut strukturierten und unstrukturierten „Cores“. Die Befunde werden unter Berücksichtigung der einschlägigen Literatur diskutiert. Es wird besonders eingegangen auf das Problem der Unterscheidung zwischen „Central Core“ und „Multiple Core“ und „Multiple Core Disease“. Die Pathogenese der Muskelveränderung wird im besonderen auch im Hinblick auf die mögliche neurogene Verursachung diskutiert. Es wird im weiteren auf die histochemischen und ultrastrukturellen Gemeinsamkeiten zwischen „Central Cores“ und „Target Fibers“ eingegangen.

Central core disease is a rare, benign, nonprogressive myopathy. Since the original description by Shy and Magee [28] in 1956 several cases have been reported and variously referred to as the same type of affection. Nevertheless, many points are still far from clear, so that new cases deserve some interest. This report concerns two members of a family where the disease had an autosomal dominant pattern of inheritance with different penetrance.

Case Report

The two patients, father and daughter, belong to a large family (Table 1). The father, aged 45 (III8), recalled difficulty in running as a child. According to him the trouble had completely regressed with advancing age, although he still complained of not being able to climb stairs while carrying a weight. On physical examination there was a mild proximal weakness, mainly of the pelvic girdle. The daughter (IV1), aged 11 years, suffered since birth from weakness of the shoulder and pelvic girdles which had been diagnosed as progressive muscular dystrophy although it was nonprogressive. She had been treated with casts for one year because of bilateral congenital dislocation of the hip. A careful family history revealed that the paternal grandmother of the girl complained of similar muscle troubles. Hip dislocation was referred in two family members who were not examined. In both father and daughter serum CPK was normal, while electromyography exhibited a myopathic pattern. Muscle biopsy was performed. Analogous thorough investigation of healthy members of the family was planned but they did not cooperate.

Muscle biopsy was performed under local anesthesia on the right quadriceps muscle in both cases. The specimen was rapidly frozen in isopentane cooled in −160°C liquid nitrogen and cut in the cryostat. Various histological and histochemical methods were carried out: haematoxylin-eosin, Gomori’s trichrome, oxidative enzymes (NADH diaphorase, LDH, SDH), ATPase at pH 9.4 and after incubation at pH 4.3 and pH 4.6, PAS, phosphorilase, Gomori for reticulin.

Table 1. Family tree of the cases