

MUSCLE DISEASES

UPDATE AND REVIEW OF CONGENITAL MYOPATHIES

Congenital myopathies are reviewed by neuropathology researchers in New Delhi, India, and Mainz, Germany. The term 'congenital myopathy' (CM) was introduced with the discovery of 'central core disease,' a non-progressive myopathy described by Shy and Magee (1956). Molecular genetics, enzyme and immunohistochemical tests and electron microscopy have led to a better understanding of CM and their classification. CM is either structured or unstructured, with or without structural changes. Structured CM include, *central core disease* (autosomal dominant, mildly progressive or static; or autosomal recessive, more severe with onset in the first decade); *multi-minicore disease* (proximal muscle weakness, spinal rigidity, scoliosis, respiratory impairment, and external ophthalmoplegia); *myotubular myopathies* (type 1 fiber atrophy and central nuclei); *X-linked myotubular myopathy* (rapidly fatal in newborn boys, presenting with hypotonia and respiratory insufficiency, and arthrogryposis multiplex); *centronuclear myopathy* (autosomal dominant or recessive, or sporadic, neonatal and childhood forms, mildly progressive, central nuclei, type 1 predominance); *nemaline myopathy* (thread like rod inclusions on Gomori trichrome stains, 6 different forms, congenital to adult); *actin aggregate myopathy* (similar to nemaline myopathy, early onset, rapid course, rarely benign); *desminopathy* (slowly progressive, second to fourth decade distal weakness onset, cardiomyopathy, autosomal dominant or recessive); *a-B-crystallinopathy* (similar to desminopathies, a myofibrillary myopathy); *hyaline body myopathy* (subsarcolemmal hyalinized bodies, rich in myofibrillary ATPase and myosin). Unstructured CM: *congenital fiber type disproportion* (non-progressive childhood CM with relatively good prognosis, type 1 fiber predominance).

No clinical symptomatology is specific for an individual CM, but some clinical findings are more frequent in certain CMs; eg. ptosis or extraocular muscle weakness in multicore disease, nemaline myopathy, centronuclear myopathy, and congenital fiber type disproportion; scoliosis in desminopathies, and central core disease. (Sharma MC, Jain D, Sarkar C, Goebel HH. Congenital myopathies – a comprehensive update of recent advancements. **Acta Neurol Scand** May 009;119:281-292). (Respond: Dr MC Sharma, Department of Pathology, All India Institute of Medical Sciences, New Delhi-110029, India. E-mail: sharmamehar@yahoo.co.in).

COMMENT. An expertise in enzyme and immunohistochemical methods, electron microscopy, and molecular genetics is required to distinguish the rapidly expanding differential diagnosis of congenital myopathies. Clinical symptomatology is largely nonspecific.

BENIGN RECURRENT SIXTH CRANIAL NERVE PALSIES

A retrospective chart review of a cohort of 253 pediatric patients with sixth nerve palsies uncovered 30 cases of benign sixth nerve palsy, of which 9 were recurrent, in a study at University of Pennsylvania School of Medicine, Philadelphia. Sixth nerve palsy occurred alone in 225 patients, and the etiologies were as follows: 90 (40%) had neoplasms, 25