Pediatric Neurology Part III: Chapter 140. Myofibrillar myopathies (Handbook of Clinical Neurology)

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Myofibrillar myopathies (MFMs) are rare, inherited or sporadic, progressive neuromuscular disorders with considerable clinical and genetic heterogeneity. MFMs are defined morphologically by foci of myofibril dissolution that begins at the Z-disk, accumulation of myofibrillar degradation products, and ectopic expression of a large number of proteins including desmin. To date, mutations in six genes are known to cause MFMs, accounting for approximately half of the MFM patients identified. The causative genes encode mainly sarcomeric Z-disk(-related) proteins: desmin, αB-crystallin, myotilin, Z-band alternatively spliced PDZ motif containing protein (ZASP), filamin C and the antiapoptotic BCL2-associated athanogene 3 (Bag3). Although in most MFM patients the disease presents in adulthood and evolves slowly, some patients with desminopathy, αB-crystallinopathy or Bag3opathies have an infantile or juvenile disease onset. Cardiac involvement is very common in desminopathies and can sometimes be the initial or only symptom of the disease. Respiratory symptoms are noted during childhood in αB-crystallinopathies. Early severe cardiac and respiratory involvement is seen in Bag3opathies. Optical microscopic and immunohistochemical features are similar in MFMs; however, ultrastructural findings can be useful to differentiate between the distinct MFM subtypes. No curative treatment for MFMs is currently available. Careful follow-up, especially of cardiac and respiratory function, is important.

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