Alpha-B crystallin-related late-onset myopathy

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Alpha-B crystallin-related late-onset myopathy. ORPHA:399058

A rare, genetic, alpha-crystallinopathy disease characterized by adult-onset myofibrillar myopathy, variably associated with cardiomyopathy and/or posterior pole cataracts. Patients typically present progressive proximal and distal muscle weakness and wasting of lower and upper limbs, often with velopharyngeal involvement including dysphagia, dysphonia and ventilatory insufficiency. Electromyography shows myopathic features and muscle biopsy reveals myofibrillar myopthay changes.