Autosomal dominant limb-girdle muscular dystrophy type 1E

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Autosomal dominant limb-girdle muscular dystrophy type 1E. ORPHA:34517

A subtype of autosomal dominant limb-girdle muscular dystrophy characterized by an adult onset of progressive cardiac conduction defects that begin with cardiac dysrhythmia. Congestive heart failure and symptoms of progressive muscle weakness (present in a proximal distribution) tend to occur later. Affected patients may present only the cardiac features of the disease. Additional features include exertional dyspnea, calf hypertrophy, elevated creatine kinase serum levels and muscle cytoplasmic inclusions.