Mevalonic aciduria

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Mevalonic aciduria. ORPHA:29

Mevalonic aciduria (MVA) is a rare, very severe form of mevalonate kinase deficiency (MKD; see this term) characterized by dysmorphic features, failure to thrive, psychomotor delay, ocular involvement, hypotonia, progressive ataxia, myopathy, and recurrent inflammatory episodes.