ALDH18A1-related De Barsy syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. ALDH18A1-related De Barsy syndrome. ORPHA:35664

A rare, genetic, neurometabolic disease characterized by prenatal and postnatal growth retardation, hypotonia, failure to thrive, large and late-closing fontanel, development delay, cutis laxa, joint laxity, progeroid appearance, and dysmorphic facial features. In addition, corneal opacities, cataracts, myopia, seizures, hyperreflexia and athetoid movements have also been associated.