Open Peer Review on Qeios

Congenital cataract-hearing loss-severe developmental delay syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Congenital cataract-hearing loss-severe developmental delay syndrome. ORPHA:300313

Congenital cataract-hearing loss-severe developmental delay syndrome is a rare, genetic, lethal, neurometabolic disease characterized by congenital cataracts, sensorineural hearing loss, severe psychomotor developmental delay, severe, generalized muscular hypotonia, and central nervous system abnormalities (incl. cerebellar and cerebral hypoplasia, hypomyelination, wide subarachnoid spaces), in the presence of low serum copper and ceruloplasmin. Nystagmus and seizures have also been reported.