X-linked neurodegenerative syndrome, Bertini type

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. X-linked neurodegenerative syndrome, Bertini type. ORPHA:85334

An X-linked syndromic intellectual disability characterized by congenital ataxia and generalized hypotonia, global developmental delay with intellectual disability, myoclonic encephalopathy, progressive neurological deterioration, macular degeneration, and recurrent bronchopulmonary infections.