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Intellectual disability-alacrima-achalasia syndrome

INSERM

Source

INSERM. (1999). *Orphanet: an online rare disease and orphan drug data base. Intellectual disability-alacrima-achalasia syndrome. ORPHA:289483*

Intellectual disability-alacrima-achalasia syndrome is a rare, genetic intellectual disability syndrome characterized by delayed motor and cognitive development, absence or severe delay in speech development, intellectual disability, and alacrima.

Achalasia/dysphagia and mild autonomic dysfunction (i.e. anisocoria) have also been reported in some patients. The phenotype is similar to the one observed in autosomal recessive Triple A syndrome, but differs by the presence of intellectual disability in all affected individuals.