# Autism spectrum disorder-epilepsy-arthrogryposis syndrome

INSERM

## Source

*INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Autism spectrum disorder-epilepsy-arthrogryposis syndrome. ORPHA:370943*

SLC35A3-CDG is a form of congenital disorders of N-linked glycosylation characterized by distal arthrogryposis (mild flexion contractures of the fingers, deviation of the distal phalanges, swan-neck deformity), retromicrognathia, general muscle hypotonia, delayed psychomotor development, autism spectrum disorder (speech delay, abnormal use of speech, difficulties in initiating, understanding and maintaining social interaction, limited non-verbal communication and repetitive behavior), seizures, microcephaly and mild to moderate intellectual disability that becomes apparent with age. The disease is caused by mutations in the gene SLC35A3 (1p21).