DISEASE:
Autism spectrum disorder-epilepsy-arthrogryposis syndrome

NAME:	Autism spectrum disorder-epilepsy-arthrogryposis syndrome
DESCRIPTION:	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.
ORPHACODE:	370943
SYNONYMS:	SLC35A3-CDG
XREF(S):	Orphanet OMIM ICD-10
ANALYTE(S):	SLC35A3
CREATED:	13 May 2019 - 01:02
CHANGED:	22 Jun 2023 - 16:14

RELATED CONTENT

Related Genetic Tests

- Congenital disorders of glycosylation (79 genes)

Related Laboratories

- Centrum Menselijke Erfelijkheid - KUL

Related Analytes

- solute carrier family 35 member A3

Related Gene Panels

- Congenital disorders of glycosylation (79 genes) - KUL