DISEASE:
Early-onset progressive diffuse brain atrophy-microcephaly-muscle weakness-optic atrophy syndrome

NAME:	Early-onset progressive diffuse brain atrophy-microcephaly-muscle weakness-optic atrophy syndrome
DESCRIPTION:	A rare, severe early-onset neurodegenerative encephalopathy characterized mainly by developmental delay (DD) / developmental regression (DR), epilepsy, cortical atrophy, secondary hypomyelination and thin corpus callosum. Additional features include secondary microcephaly, hypotonia, spasticity, optic atrophy and skeletal anomalies.
ORPHACODE:	496641
XREF(S):	Orphanet ICD-10 OMIM
ANALYTE(S):	TBCD
CREATED:	13 May 2019 - 01:02
CHANGED:	22 Jun 2023 - 16:14

RELATED CONTENT

Related Genetic Tests

- Epilepsy (gene panel)

Related Laboratories

- Centrum Medische Genetica - UZ Antwerpen

Related Analytes

- tubulin folding cofactor D

Related Gene Panels

- Rare epilepsy with developmental delay (> 240 genes) - UZA