Belgian Genetic Tests database

DISEASE:
SYNGAP1-related developmental and epileptic encephalopathy

NAME:	SYNGAP1-related developmental and epileptic encephalopathy
DESCRIPTION:	A rare genetic developmental and epileptic encephalopathy (DEE) characterized by developmental delay, generalized epilepsy consisting of eyelid myoclonia with absences and myoclonic-atonic seizures, intellectual disability and autism spectrum disorder (ASD).
ORPHACODE:	544254
SYNONYMS:	SYNGAP1-related DEE
XREF(S):	Orphanet
ANALYTE(S):	SYNGAP1
CREATED:	04 Feb 2020 - 15:13
CHANGED:	22 Jun 2023 - 16:14

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