DISEASE:
CNTNAP2-related developmental and epileptic encephalopathy

NAME:	CNTNAP2-related developmental and epileptic encephalopathy
DESCRIPTION:	A rare, genetic, syndromic neurodevelopmental disorder characterized by moderate to mostly severe intellectual disability, speech impairment with normal or mildly delayed motor development and early-onset seizures often accompanied by developmental regression. Autistic behavior and stereotypic movements are common.
ORPHACODE:	163681
SYNONYMS:	CDFE syndrome CDFES CNTNAP2-related DEE Cortical dysplasia-focal epilepsy syndrome
XREF(S):	Orphanet ICD-10 OMIM
ANALYTE(S):	CNTNAP2
CREATED:	13 May 2019 - 01:02
CHANGED:	22 Jun 2023 - 16:14

Source URL: http://gentest.healthdata.be/disease/143

RELATED CONTENT

Related Genetic Tests

- Epilepsy (gene panel)
- Epilepsy gene panel

Related Laboratories

- Centrum Medische Genetica - UZ Antwerpen
- Centrum Medische Genetica - UZ Brussel VUB

Related Analytes

- contactin associated protein 2

Related Gene Panels

- Epilepsy gene panel - VUB
- Rare epilepsy with developmental delay (> 240 genes) - UZA

Source URL: http://gentest.healthdata.be/disease/143