

DISEASE:
Myoclonic-astatic epilepsy

NAME:	Myoclonic-astatic epilepsy
DESCRIPTION:	A rare, childhood onset epilepsy syndrome characterized by multiple seizure types including myoclonic-tonic (MA) seizures that occur usually in previously healthy children.
ORPHACODE:	1942
SYNOMYS:	Doose syndrome EMAS Epilepsy with myoclonic-astatic seizures Epilepsy with myoclonic-tonic seizures MAE Myoclonic atonic epilepsy Myoclonic-astatic epilepsy in early childhood
XREF(S):	Orphanet ICD-10 OMIM OMIM OMIM

ANALYTE(S):	<u>SLC2A1</u> <u>SYNGAP1</u> <u>NEXMIF</u> <u>SCN1A</u> <u>CHD2</u> <u>SLC6A1</u> <u>AP2M1</u>
CREATED:	13 May 2019 - 01:02
CHANGED:	22 Jun 2023 - 16:14

Source URL: <http://gentest.healthdata.be/disease/477>

RELATED CONTENT

Related Genetic Tests

- [Epilepsy \(gene panel\)](#)

Related Laboratories

- [Centrum Medische Genetica - UZ Antwerpen](#)

Related Analytes

- [adaptor related protein complex 2 subunit mu 1](#)
- [chromodomain helicase DNA binding protein 2](#)
- [neurite extension and migration factor](#)
- [sodium voltage-gated channel alpha subunit 1](#)
- [solute carrier family 2 member 1](#)
- [solute carrier family 6 member 1](#)
- [synaptic Ras GTPase activating protein 1](#)

Related Gene Panels

- [Rare epilepsy with developmental delay \(> 240 genes\) - UZA](#)