DISEASE:
Lissencephaly due to LIS1 mutation

NAME:	Lissencephaly due to LIS1 mutation
DESCRIPTION:	Lissencephaly due to LIS1 mutation is a cerebral malformation with epilepsy characterized predominantly by posterior isolated lissencephaly with developmental delay, intellectual disability and epilepsy that usually evolves from West syndrome to Lennox-Gastaut syndrome. Additional features include muscular hypotonia, acquired microcephaly, failure to thrive and poor control of airways leading to aspiration pneumonia.
ORPHACODE:	95232
SYNONYMS:	PAFAH1B1-related lissencephaly
XREF(S):	Orphanet ICD-10 OMIM
ANALYTE(S):	PAFAH1B1
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RELATED CONTENT

Related Genetic Tests

- Epilepsy (gene panel)
- Lissencephaly (LIS1 gene)
- Lissencephaly / subcortical band heterotopia
- Malformations of cortical development (235 genes)

Related Laboratories

- Centre de Génétique-Institut de Pathologie et de Génétique (IPG)
- Centrum Medische Genetica - UZ Antwerpen
- Centrum Medische Genetica - UZ Brussel VUB

Related Analytes

- platelet activating factor acetylhydrolase 1b regulatory subunit 1

Related Gene Panels

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