DISEASE:
Cobblestone lissencephaly without muscular or ocular involvement

NAME:	Cobblestone lissencephaly without muscular or ocular involvement
DESCRIPTION:	A rare, genetic, cobblestone lissencephaly disease characterized by the presence of a constellation of brain malformations, including cortical gyral and sulcus anomalies, white matter signal abnormalities, cerebellar dysplasia and brainstem hypoplasia, existing alone or in conjunction with minimal muscular and ocular abnormalities, typically manifesting with severe developmental delay, increased head circumference, hydrocephalus and seizures.
ORPHACODE:	352682
SYNONYMS:	Cobblestone lissencephaly without muscular or eye involvement Lissencephaly type 2 without muscular or eye involvement Lissencephaly type 2 without muscular or ocular involvement
XREF(S):	Orphanet ICD-10 OMIM
ANALYTE(S):	LAMB1
CREATED:	13 May 2019 - 01:02
CHANGED:	22 Jun 2023 - 16:14

Source URL: http://gentest.healthdata.be/disease/1903

RELATED CONTENT

Related Genetic Tests

- Malformations of cortical development (235 genes)

Related Laboratories

- Centrum Medische Genetica - UZ Brussel VUB

Related Analytes

- laminin subunit beta 1