Universitair Ziekenhuis Brussel

Centrum voor
Medische Genetica

CRITERIA FOR MUTATION ANALYSIS OF L1CAM GENE

Dear Colleague,

Hereby you find a checklist that should be completed for each L1CAM gene mutation analysis you request.

Notify that this analysis will only be performed when the following criteria are fulfilled:

- male gender
- in case of a prenatal diagnosis it should concern a case of isolated hydrocephaly (no associated malformations) **with** a positive familial history.
- in case of postnatal diagnosis:
  - When there is at least one other affected family member besides the index patient, the index patient should present at least one typical feature.
  - When there is no positive familial history, at least two typical features should be present in the index patient.

Typical features:

- hydrocephaly
- spasticity
- mental retardation
- agenesis / hypoplasia of the corpus callosum
- adducted thumbs

If your index case does not fulfill the above-mentioned criteria, please contact the lab **before** sending your sample to us.

Please return the enclosed checklist to:

W. Lissens, PhD.
Centre for Medical Genetics
University Hospital, VUB
Laarbeeklaan 101
1090 Brussels
Belgium.

CHECKLIST L1CAM mutation search

- NAME OF THE PATIENT: _____
  (or MOTHER in case of PRENATAL DIAGNOSIS)
- DATE OF BIRTH: _____
- KARYOTYPE: _____
- PEDIGREE: _____

PRENATAL:

- gestational age at detection: _____
- microbiological screening:
  - ☐ Toxoplasmosis: _____
  - ☐ CMV: _____
  - ☐ Rubella: _____
  - ☐ Herpes: _____
- anomalies on ultrasound
  - ☐ cerebral: _____
    - ventricular measurements: _____
    - other: _____
  - ☐ extra cerebral: _____
- prenatal cerebral MRI: _____

POSTNATAL:

birth weight: _____
birth length: _____
OFC: _____

clinical manifestations typical for L1 disease:

- ☐ hydrocephaly
- ☐ mental retardation / developmental delay
- ☐ spasticity
- ☐ adducted thumbs

other clinical manifestations

- ☐ brain imaging
- ☐ US
- ☐ CT scan
- ☐ MRI