GLUCOSE TRANSPORTER DEFICIENCY SYNDROME (GLUT1DS)

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PATIENT

Surname.....

First name.....

Maiden name.....

Sex: I__I

Date of birth: I__I__I / I__I__I / I__I__I__I__I

PRESCRIBER

Obligatory stamp

Signature:

REQUESTED TEST

☐ **FLOW CYTOMETRY ANALYSIS OF ERYTHROCYTE GLUCOSE TRANSPORTER GLUT1 (METAGLUT1 TEST)** (OPL: GLUDS)
- This test cannot be conducted on infants under three months old (contact us)
- Attach blood count results

☐ ***SLC2A1* GENE ANALYSIS** (OPL: SLC2A)
- The medical consultation and consent certificate must be attached to this form to analyse an individual's genetic characteristics

INDICATION

☐ **CLASSIC PHENOTYPE:** epilepsy AND abnormal paroxysmal movements AND development disorder/intellectual deficiency

☐ **ATYPICAL PHENOTYPE** **(INDICATE ONLY THE PRIMARY DISORDER IF IT IS NOT ISOLATED)**

☐ Epilepsy/early childhood convulsion (<4 years)
OR

☐ Ataxia and/or abnormal paroxysmal movements
OR

☐ Paroxysmal exercise-induced dystonia (PED)
OR

☐ Development disorders/intellectual deficiency

☐ **FAMILY TESTING:** testing of family members to find the *SLC2A1* gene variant that causes the pathology (attach copy of results)

CLINICAL AND BIOLOGICAL INFORMATION

Lumbar puncture: ☐ NO ☐ YES CSF glucose = ……..

CSF glucose/blood glucose = ……

Family history: ☐ NO ☐ YES

Complete clinical description:

BLOOD SAMPLING

2 tubes of EDTA whole blood - storage and delivery to Laboratoire CERBA within a maximum of 4 days - temperature: +4°C

Minimum volume required for flow cytometry analysis: 500 µL

We keep the sample for the analysis by flow cytometry for 20 days. Within this period, at your written request and with the relevant documents (informed consent and prescriber's attestation), we are therefore able to perform the secondary analysis of the SCL2A1 gene

Sampling date: I__I__I / I__I__I / I__I__I__I__I Time of sample: I__I__I:I__I__I

Customer no.: C I__I__I__I__I__I__I / I__I

Sampling laboratory stamp: