Semmelweis Egyetem
Genomikai Medicina és Ritka Betegségek Intézete

Intézetigazgató: Prof. Dr. Molnár Mária Judit
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Clinical Trial – Medical Record

Name: L.K.
D-O-B: 07.10.1999.
Date: 2022. 06. 15.

Medical History

She had delayed movement and speech development.
Her epilepsy (GM seizures) started at the age of 6 (2005), she gets antiepileptic treatment as Levil and Etopro.
Gait disorder is known since her youth, which began to worsen around the age of 20 and is associated with dizziness.
Genetic testing (sequence analysis) determined pathogenic variants /c.1880G>A (p.Arg627Gln) és c.2542G>A (p.Gly848Ser)/ in compound heterozygous form in POLG1 gene.

She applied for the clinical trial, protocol number: SPIMD-301 (EudraCT number: 2021-003907-16). Today is a screening visit. She is not planning to become pregnant and has no such relationship. There was no hospitalization in the last 30 days. He does not have and has not had any clinically significant respiratory or cardiovascular disease, neuropathy, malignancy, or organ transplantation procedure. There has been no GM episode for more than 70 days. There were no previously diagnosed HIV, HBV, HCV or autoimmune diseases. In the last 30 days, you has not participated in either interventional or non-interventional clinical trials. There was no abuse. So far, no side effects, drug allergies, or anaphylactic reactions have occurred with regard to the medications taken.

Family history: Her older brother (László Róbert) deceased in the age of 24 cause of serious epilepsy and ataxia, Depakine caused serious hepatic failure.

Status

Weight: 94,8 kg. Height: 165 cm. RR:20/min. BP: 118/92 Hgmm. Temp.: 36,7 C. Bilateral horizontal nystagm when viewing direction, hypermetric saccades. Mild paresis in proximal muscles. Mild deep reflexes. Dysdiadochokinesis. She can walk with rollator max. distance: 15 m. Euthym. EQ-5D-5L: 50. 6MWT with rollator: 207,10 cm. 3TUG: 1. 75,2sec – loss her balance, not able to complete. 5XSST: 82 sec.
Demography: Caucasian

Epicrisis

Diagnoses: Epilepsy- Depression - Mitochondrial recessive ataxia syndrome (POLG1 gene associated).- Obesity.- Hypercholesterinaemia. Axonal senso-motor polyneuropathy.

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Screening visit of clinical trial /protocol number: SPIMD-301 (EudraCT number: 2021-003907-16)/ was performed.

Today, we provided enough time and a calm environment to read the documents (the Patient information and declaration of consent Hungarian version SPIMD-301_Hungary_Main ICF_v3.1.0_30Mar2022_HUN Identification number: 301-004) and answered her questions to give all the essential information. After the patient signed the "Patient Information" and "Declaration of Consent", she was recruited in the clinical trial protocol number: SPIMD-301 (EudraCT number: 2021-003907-16).
Vital parameters were measured, EQ-5D-5L questionnaire was filled. ECG was performed after 5 minutes of rest. Blood and urine samples were taken. Functional tests have been assessed. The screening visit was performed. Regarding enrolment in the clinical trial, the study coordinator of the Institute (Ms Noémi Töreki) will notify the patient within 2 weeks. The 1st visit will take place on 14 July 2022.

It is recommended to continue the current medications:
Tbl. Levil 1000mg 2x1,5
Tbl. Etopro 50mg 2x2
Tbl. Sertalin 50mg 1x1
Tbl. Frontin 0,25mg R:1, E: 2.

The patient has been informed orally about his medical condition, the proposed tests and interventions, the possible benefits, and risks of having or not having them, the planned dates of the tests and interventions, his right to decide about the proposed tests and interventions, the possible alternative procedures and methods, the course of treatment and the expected outcome, further care, the proposed lifestyle.