SEMMELWEIS EGYETEM

*Általános Orvostudományi Kar*

**Genomikai Medicina és Ritka Intézete**
Igazgató: Prof. Dr. Molnár Mária Judit
Cím: 1082 Budapest, Üllői út 78.
Levelezési cím: 1085 Budapest, Üllői út 26. 1428 BP. Pf.2.

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BT
DOB:1983
Admission: 2022.03.22.
Discharge: 2022.03.25.

**Medical history**

Based on his medical history and EESZT (National eHealth Infrastructure) documentation, there is no relevant comorbidity present. He does not take medication regularly.

Around 2020, his coworkers and neighbors noticed his unsteadiness in walking and some involuntary limb movements. He first presented to our outpatient clinic in November 2022 on behalf of the advice of a regional neurologist with a tentative diagnosis of Huntington's disease. He needed further investigation and genetic testing. By this time, his symptoms had worsened, his movement disorders and involuntary movements had intensified, they became disturbing to himself too. He also noticed forgetfulness. Due to the family history and the detected hyperkinetic movement disorder, Huntington's disease was suspected, and genetic testing was initiated. The genetic testing completed in December 2022 confirmed the genetic diagnosis of Huntington's disease, identifying 21/47 +/-1 CAG repeats in the HTT gene.
In January 2022, in the framework of genetic counseling, we handed over his findings, at which time he was scheduled for inpatient admission for the purpose of medication adjustment and physical therapy.

Family history: His father had a similar disease, but no specific diagnosis was made, and there was no genetic test. His symptoms started in his forties and he died at the age of 55. There is no information about paternal grandparents. Father's brother died as a result of alcoholism, nothing more is known about him. He has a half-brother, from his mother's side, he is 53 years old, healthy. He has no children.

**Current complaints**
We are currently taking him for neuropsychiatric tests, medication adjustment and coordination training. He has no new complaints, he notices his excessive movements, which are now bothering him. He describes his mood as indifferent.
Previously the patient was informed about the opportunity to participate in a clinical trial. He agreed to participate in the screening visit of the Novartis CLMI070C12203 clinical trial. He read the consent forms in a separate, quiet place, in the company of his mother. We answered all the raised questions. He signed the patient information leaflet with version number 1. the pharmacogenetic patient information leaflet, and received a copy.

**Physical examination:**
Well developed, hydrated and nourished. Weight: 78 kg, Height: 174 cm. No internal, visceral, cardiac abnormalities. Cranial nerves intact. Sustained tongue protrusion: 4-5 seconds. Muscle trophy corresponds to nutrition. Normotonia throughout the body. Muscle strength was maintained throughout the body. Muscle stretch reflexes throughout the body are 2/4 equal.Accurate cerebellar tests. Stable in Romberg

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position. Mild axial instability. During gait normal contralateral synkinesis. Blind-gait holds direction. Involuntary, non-stereotype dyskinesias in all limbs, sometimes in trunk and facial muscles. Speech assesment: fluency, comprehension, repetition is good. Frontal release signs negative. Mood is characterized by mild anxiety. UHDRS-TMS: 34/124,Functional assessment: 24/25. Independence: 95%.

**Invastigations: Laboratory findings**

| | | | | | | |
|---|---|---|---|---|---|---|
| CRP | | <4.0 | mg/L | | <10.0 | |
| CBC. | | | | | | |
| WBC | | 6.79 | G/L | | 4.00-10.00 | |
| Neutrophil granulocyte % | | 54.3 | % | | 45.0-70.0 | |
| Eosinophil granulocyte % | | 1.6 | % | | <5.0 | |
| Basophil granulocyte % | | 0.6 | % | | <2.0 | |
| Monocyte % | | 8.0 | % | | 2.0-10.0 | |
| Lymphocyte % | | 35.5 | % | | 25.0-45.0 | |
| Immature granulocyte | | 0.1 | % | | <0.8 | |
| Neutrophil granulocyte | | 3.69 | G/L | | 1.80-7.00 | |
| Eosinophil granulocyte | | 0.11 | G/L | | <1.00 | |
| Basophyl granulocyte | | 0.04 | G/L | | <0.20 | |
| Monocyte # | | 0.54 | G/L | | 0.15-0.90 | |
| Lymphocyte # | | 2.41 | G/L | | 1.00-4.00 | |
| RBC | | 4.18 | T/L | | 4.00-5.20 | |
| Hemoglobin | | 125 | g/L | | 120-150 | |
| Hematocrit | | 0.36 | L/L | | 0.34-0.45 | |

| | | | | | | |
|---|---|---|---|---|---|---|
| Iron | | 11.4 | umol/L | | 10.7-32.2 | |
| Transferrin | | 2.49 | g/L | | 2.00-3.60 | |
| Transferrin saturation | | 18 | % | | 16-45 | |
| Ferritin | | 11 | ug/L | | 10-120 | |
| Glucose | | 5.2 | mmol/L | | 4.1-5.9 | |
| Karbamid | | 3.9 | mmol/L | | 2.8-7.2 | |
| Kreatinin | | 62 | umol/L | | 45-84 | |
| eGFR (CKD EPI) | | >90 | ml/min/1.73 m2 | | >90 | |
| Uric acid | | 191 | umol/L | | 154-357 | |
| Bilirubin | | 7.0 | umol/l | | 5.0-21.0 | |
| GOT (ASAT) | | 19 | U/L | | <35 | |
| GPT (ALAT) | | 16 | U/L | | <35 | |
| GGT | | 19 | U/L | | <38 | |
| ALP | | 85 | U/l | | 30-120 | |
| LDH | | 166 | U/L | | <248 | |
| CK | | 99 | U/L | | <145 | |
| Cholesterol | | 4.1 | mmol/L | | 2.0-5.2 | |
| HDL-cholesterol | | 1.77 | mmol/L | H | 1.03-1.5 | |
| LDL-cholesterol | | 2.26 | mmol/L | | <3.30 | |
| Triglycerides | | 0.9 | mmol/L | | <1.7 | |

2023.02.13. Neuropsychological Testing and Assessment: Addenbrooke's cognitive examination: 73/100 point, Orientation 10/10, Attention 3/8 , Memory 23/35, Verbal fluency 4/14, Language 28/28, Visuospatial skills 5/5 , VL/OM 2,13 detects frontotemporal dementia. Apathy evaluation scale not indicating, BDI 2 points=not indicating depression, SHAI 2 points, not indicating, BAI 3 points, not indicating. Opinion: According to his family, Huntington's symptoms started years ago, but he didn't notice it. He says his mood, motivation, and outlook are good, but this contradicts the fact that he does not have a relationship due to his illness and also lost his job due to his illness.

**Diagnosis:** Chorea Huntington

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Epicrisis:

The 40 yo. Huntington's disease patient was admitted for controlling his tetrabenazine treatment due to choreiform hyperkinesis. The gradual increase of Motetis(tetrabenazine) to a dose of 3x1/2 tbl almost completely eliminated the excessive movements. In addition, the patient also benefited from coordination exercises. Psychological tests did not indicate a mood disorder, so we have not started pharmacotherapy for the time being, but we plan to follow the patient closely. There were no pathological abnormalities in laboratories.

Recommendations: Tbl Motetis (tetrabenazine) take half tablet 3 times a day (8 am, 2 pm, 7 pm).

Each day we recommend 30 minutes of moderate intensity physical activity, weekly 2-3 times coordination exercises based on the home exercise plan we gave him.
A regular neurological follow-up is recommended (every 6 months).
If he will be eligible for the clinical trial the patient will be informed of the date of the baseline visit by the study coordinator Noemi Töreki. If he will be not eligible the next appointment as an outpatient in our clinic in 3 months: 05/24/2023 at 9:30 am. Psychological control: 24.05.2023. 10:30 a.m
We draw the attention of the patient to the importance of contraception.

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.

Budapest, 25/03/2022.