SEMMELWEIS EGYETEM
Általános Orvostudományi Kar
Genomikai Medicina és Ritka Intézete
Igazgató: Prof. Dr. Molnár Mária Judit
Cím: 1082 Budapest, Üllői út 78.
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B. A.
Admitted:2022.07.15
Discharged: 2022.07.19.

Medical history:

The 63 yo patient lives in Transylvania, Romania. He noticed in 2017 an unsteady gait, and difficulty when walking down stairs, he also noticed it when walking up stairs, but it bothered him less. He felt more unsteady in the dark. There was no numbness in his limbs. His wife observed twitching in his lower limbs during the night. He complained of intermittent stiffness and a frequent muscular fever in the thighs and calves, but regardless of the load, mainly on the left side. Fine coordination in his hands is maintained, no complaints.

He is known with hypertension, taking Merckformin for insulin resistance since about 2018. Hypercholesterolaemic, started taking rosuvastatin in autumn 2019. In 2009, he had melanoma removed from his thigh and lymph nodes removed from the right inguinal region. No chemotherapy was needed because the sentinel lymph nodes were tumor free.

Previous investigations:

Skull MRI: some small ischaemic lesions in both hemispherea. C spine MRI: mild-grade degenerative lesions, discus protrusions in CV-VI segment without clear radicular compression. Th spine MRI: no abnormalities on the myelon. Polydiscopathy at the level of Th.VII-VII, Th.VIII-IX and Th.IX-X with disc protrusions mainly at the level of Th.VIII-IX with acute bone marrow oedema. Lower limb MEP examination found central conduction times at the upper limit of normal in Oct 2019. Five month later MEP showed elongated CMCT with right-sided dominance. SSEP: slightly elongated latencies right side. EMG in 2019 did not detect ongoing active denervation. Total protein in CSF: 542 mg/l, CSF IgG 40. mg/l. Serum IgG 10 g/l. Oligoclonal gammopathy was found on IEF. IgG detection was present in serum, suggesting systemic inflammation. Abdominal US: Mild diffuse hepatic lesion. Lacerated gallbladder. Right sided renal cysts. Benign prostatic enlargement. Gastroscopy: gastric polyps and GERD, and some signs for erosive gastritis. Colonoscopy revealed polyps and diverticulosis. Echocardiography: normal findings. Carotid Dopler: mild atherosclerosis.
In January 2020, the patient was examined in our department in order to perform lumbar puncture. The reason for the investigation were the previously doubtful paraneoplastic marker test and the OCBs found in CSF and serum. No paraneoplastic antibody was found in the CSF.

Whole Exome Sequencing was performed which revealed mutations in SPG7 gene in compound heterozygous state (p.Gly352ALafsTer87 and p.Ala510Val). The patients was treated symptomatically with baclofen, diazepam and guaifenesin, but did not feel any improvement regarding spasticity. In September 2020 the baclofen and merckformin were discontinued. He found it harder to stand up and walk after a short sitting.

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He first presented in our outpatient clinic in Jan. 2022. He complained about mild progressive lower limb stiffness, which was treated ineffectively with tizanidine. HbA1C levels were at the upper limit of normal. DEXA examination found no osteoporosis. No pain. No complaints of vision, but when driving he has to turn his head to look sideways, looking sideways with his eyes is not quick. His balance has also deteriorated.

Family history: his father had Alzheimer's disease, he died at the age of 75. His mother is healthy. His sister is also healthy. He has a son 24 yo, healthy, but in infancy he needed surgical treatment of patent ductus arteriosus Botalli.

Current complaints: worsened lower limb stiffness and gradual deterioration of the gait. Because of the infectivity it is admitted for muscle relaxant infusions. He also complained of significant lumbar spine pain, and stiffness in the lower back. No complaints for fever, cough, dyspnoea, chest pain, loss of consciousness, nausea,vomiting. Stools,urine normal.

Physical examination:
Well developed, hydrated and nourished, mild central overweight. Weight: 88 kg, Height: 181 cm. Skin in warm, dry and intact without rashes or lesions. No masses, hepatomegaly, or splenomegaly are noted. The feet are in a left supination in varus position. The peripheral veins are well palpable, varicose veins on the left lower limb. The head is normocephalic and atraumatic without tenderness, visible or palpable masses, depressions, or scarring. Neck and back are without deformity. Curvature of the cervical, thoracic, and lumbar spine are within normal limits. Signs of lumbar paraspinal muscle spasm. Lasegue's sign is negative. The patient states good olfactory sensation bilaterally. Symmetric visual fields. Extraocular muscle movements intact, pupils equally reactive to light. Their direct and consensual light response is retained. Accommodation and convergence normal. Mild conjugated horizontal gaze palsy (not following finger all the way to end position), vertical gaze palsy, unable to converge. No nystagmus or diplopia. In all three branches of the trigeminal nerve symmetric facial sensation. Symmetric facial expression, normal motor function. No nasolabial fold flattening. Normogeusia. Bilateral normacusis. The gag reflex is triggered by touching both sides of the pharynx. The soft palate arches are at equal height, the uvula is centrally located. Normophonia, no articulation disorder. The soft palate reflexes are equal. No dysphagia. Tip of tongue is in midline when extended, tongue moves well in all directions, no atrophy, no fibrillation. Normotrophic. Spasticity in both lower limbs with left predominance. Examined in lying:, MAS 2 in knee and in left ankle. MAS 3+ right ankle. In upper limbs normotonia. In lying muscle strength is maintained throughout the body. Negative Barré's and Mingazzini paresis tests. In squats the patient is slow and needs little support. Normal plantar flexion, normal dorsiflexion but not holding. Brisk deep tendon reflexes, except of bilateral poor Achilles reflex. No Babinski sign, but dorsiflexion tendency. At the present examination, protopathetic sensitivity intact, mild epicritic sensory disturbance noted: no vibrationsense at the basal ankle of halluxes, but at ankles, joint position the sense is adequate, graphaesthesia good. No dysdiadochokinesis.

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Accurate cerebellar tests. Stable in Romberg position. Narrow based gait, slower than normal, with left lower limb paraspasticity (knees slightly rocking, circumducts with left lower limb). During gait normal contralateral synkinesis (normal arm swing during gait). Blind-gait holds direction, but complains instability. Bárány's test no lateralisation. Speech assesment: fluency, comprehension, repetition is good. Frontal release signs are no longer elicited. Patient has no urinary or bowel complaints. Appropriate mood and affect. Awake, alert and oriented. No visual or auditory hallucinations.

2022.07.14 - 10:00 VAS: 7/10 16:00 7/10 20:00 6/10
2022.07.15 - 10:00 VAS: 5/10 16:00 6/10 20:00 5/10
2022.07.16 - 10:00 VAS: 5/10 16:00 4/10 20:00 4/10
2022.07.17 - 10:00 VAS: 4/10 16:00 2/10 20:00 3/10
2022.07.18 - 10:00 VAS: 2/10

Investigations:
2022.07.15 - Complete blood count (CBC): showed normochromic normocytic anaemia, normal iron level.

Diagnosis: Hereditary Spastic Paraplegia, Paraplegin Type (SPG7) - Lumbal back pain - Prediabetes mellitus - Hypertension - Hypercholesterolemia - Polydiscopathia thoracalis et lumbalis. – St. post osteosynth colli fem. l.d.

Epikrízis:
The 62 yo male patient was admitted because of hereditary spastic paraplegia, paraplegin type, linked to the SPG7 gene mutation confirmed by WES (whole exome sequencing). Relevant medical history includes hypertension, insulin resistance, right thigh melanoma excision and right inguinal lymphadenectomy, thoracolumbar polydsicopathy, right femoral neck fracture. He is currently admitted to our inpatient care unit for muscle relaxant infusion treatment due to increasing lower limb spasticity, specifically ankle joint stiffness, consistently worsening in gait disturbance and because of the acute lower back pain.
IV infusions of Relaxil-G (guaifenesin) and Seduxen (diazepam) were started for muscle relaxation, but the patient showed gradual decrease in his lower back pain, but the lower limb stiffness, his gait was still very spasticoparetic with left predominance. As he had previously tried oral muscle relaxants, but only at low doses, it is recommended to try again, gradually increasing the dose.
He has been receiving i.v. administrated muscle relaxants, physiotherapy, coordination exercises.

He was discharged to his home in good general condition with pain relief. He is expected to be followed up as an outpatient in his country. The patient was reffered to Dr. Szabolcs Szatmári at 2nd Clinic of Neurology, Târgu Mureș County Emergency Clinical Hospital (Address: Mun. Târgu Mureș, Târgu Mureș, Gheorghe Marinescu str. 50. Romania. Phone: 0265212111). In our department, a follow-up examination is recommended every 6 months within the framework of a telemedicine consultation.

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Recommendations: Lioresal 10mg 3x1, Myoqinon 100mg M:1 N:1, C-vitamin 1000mg M:1, Folsav 3ug M:1, Milgamma M:1, Thiogamma 600mg M:1, B-vitamin komplex M:1 + Cardura XL 4mg M:1, Egiramlon10/5mg M:1, Rawel SR 1,5mg M:1, Rosuvastatin 20mg M:1, Talliton 12,5mg 2x1

Each day we recommend 10-15 minutes of modified McKenzie exercises beside regular physical therapy and massage therapy, which the patient receives regularly at home. For symptomatic pain relief transcutaneus electrical nerve stimulation (TENS) device is recommended to use at home. . The patient was educated by a physical therapist on how to use the TENS equipment.

After your hospitalization, please see your GP as soon as possible to present your final report. In one week You will be contacted for an outpatient appointment by the medical team from Târgu Mureş.

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, 19/07/2022