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A case of the Roussy-Levy syndrome family

The Roussy-Levy Syndrome (RLS), also known as hereditary areflexic dystasia, is a rare genetic neuromuscular disorder that typically becomes apparent during early childhood. It was described in 1926. In individuals with RLS, there is a failed communication of certain nerve signals to muscles of the lower legs. Active demyelination is restricted to childhood. It leads to hypertrophic "onion bulb" changes and is accompanied and followed by progressive axonal loss. The disorder is characterised by inherited gait ataxia, absence of tendon reflexes (areflexia) of the lower legs and, eventually, the hands, weakness and degeneration (atrophy) of muscles of the lower legs, abnormally high arches of the feet (pes cavus) with increased extension of the toes and tremors of the hands. Many affected individuals also have an abnormal front-to-back and sideways curvature of the spine (kyphoscoliosis) (2, 5). We report a family, whose members in three generations (grandmother, mother, daughter) were showing these clinical signs of Roussy-Levy Syndrome.

Generation I – grandmother

A 69-year-old woman. It is difficult to gather an accurate history. The patient with the signs of dementia. It has been found that at the age of 35-40 she complained of lumbosacral pains and then difficulties in walking. At the moment, the paint is diagnosed with right-side scoliosis of the thoracic-lumbar spine, considerable degenerative-productive changes in the spine, equinovarus talipes with a contracture in the crural articulations, arch-like feet with a bayonet-like position of the big toe. On neurological examination no signs of paresis have been found. There is, however, an apparent ataxia and tremor within the upper extremities. The tendon reflexes in the extremities are absent, hypoesthesia of the radical type L4-L5-S1. She does not move on her own due to pains in the spine and inability to stand upright.

Generation II – mother

A 42-year-old woman. The history: in childhood her locomotor and intellectual functions developed normally, she reported neither pains nor difficulties in walking. She was always shorter than her peers, and as an adult was 146 cm tall. At the age of about 12 she was diagnosed with talipes equinovarus, and orthopedic shoes were recommended. The patient did not comply with the above recommendations, but would regularly wear high-heeled shoes. At the age of 18 she started to complain of pains in the lumbosacral area, occurring episodically while walking. Beginning from the age of 21 onwards the patient reported "uncertainty" on walking. These problems increased gradually, but did not make her everyday activities impossible. At 35 she had pains and numbness within the hypogastric area and

both thighs. They were associated with some episodic irregularities in urination such as urine retention. The problems were of remitting character. Her thoracic and lumbar spine was X-rayed. On examination right-sided scoliosis of the thoracic part, small degenerative changes and lateral position of the lumbar bone were diagnosed. Consequently, she was treated in an outpatient department. She received non-steroid non-inflammatory drugs, which reduced the problems. At 41 there was a sudden onset of pains in the abdomen and numbness within both thighs, considerable fatigability and uncertainty on walking. The patient could not stand on the whole foot without an orthopedic shoe. She would involuntarily stand on her toes instead. On clinical examination she was diagnosed with talipes equinovarus, a contracture in the crural articulation, an arch-like foot with the bayonet-like position of the big toe, small bilateral ataxia of both lower extremities, lack of tendon reflexes, hypoesthesia from the level TH 11 down to the knees. The patient complained of difficulties in urination. On biochemical examinations she was diagnosed with GGTP 29.3j. levels, whereas other parameters (blood morphology, transaminase, liver tests, urea, creatinine, alkaline phosphatase, protein electrophoresis, coagulator system, electrolytes, urine test) were normal. MRI of the lower half of the thoracic and lumbosacral spine was performed. She was diagnosed with thoracic scoliosis, weak degenerative-productive changes, including the posterior surface of the corpus of the lumbar vertebra, which could cause a small pressure on the meningeal sack and irritate the roots of the tail of epididymis. The intervertebral discs L3-L4, L4-L5 and L5-S1 showed an insignificant reduction in the intensiveness of the signal with small protrusion. EMG examination proved disturbances in conduction which testify to a neurogenic process at the level of the trunks of the peripheral nerves. A urodynamic examination was negative. Neuropsychological examination showed diminution of the pace of psychical processes with considerable neglect in education, an insignificant reduction of intelligence below the average. Subjectively, her mood was improved and numbness reduced. Objectively, however, the neurological status has not changed.

Generation III – daughters

A 25-year-old woman. Was born as a premature baby in 7th month of pregnancy and weighed 1,250g at birth. Then she developed normally. At the ages 4 and 5 there were single feats of epilepsy. She graduated from a secondary school and a college. She complains of pains in the thoracic and lumbosacral spine on longer standing. On clinical examination: the patient is fairly short (152 cm), left-sided scoliosis of the thoracic spine. Talipes equinovarus, arch-like feet with a bayonet-like big toe, without contracture in the crural articulation, no signs of paresis in the extremities. The signs of discreet ataxia in the lower extremities. The knee reflexes present, yet very weak without crural reflexes.

A 23-year-old woman. Was born as a premature baby in the 8th month of pregnancy. She weighed 2,450g at birth. She developed normally. On clinical examination no changes in the costal-articulatory system have been diagnosed. Neurological examination showed no aberrations from the norm. The tendon reflexes are vivid and symmetric.

Roussy-Levy Syndrome is inherited as an autosomal dominant genetic trait and is connected with segmental duplication at chromososme 17p11.2 (1, 3). This genetic defect is commonly found in patients with the hypertrophic form of the Charcot-Marie-Tooth syndrome. Also pathologic and neurophysiologic study show that RLS is a variant of demyelinating Charcot-Marie-Tooth Syndrome but has original morphological and symptomatological features (1, 4). Based on clinical signs the women could be classified as having the RLS. All have displayed gait ataxia, areflexia, pes cavus and sideways curvature of the spine (kyphoscoliosis). Usually in RLS, the onset of symptoms is in the first decade of life, but in our patients was later. During the first decade of life, the women had the changes in the costal-articulatory system (pes cavus, kyfoscoliosis) but no or minimally gait ataxia. It

is interesting, that in this family were suffered only women suffered. The members of this family require a genetic study.

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SUMMARY

Roussy-Levy syndrome, also known as hereditary areflexic dystasia, is a very rare genetic neuromuscular disorder that typically becomes apparent during early childhood. The disorder is characterised by inherited gait ataxia, pes cavus and areflexia which are eventually associated with distal muscle atrophy, postural tremor and minor sensory loss. We report a family whose members in three generations (grandmother, mother, daughters) were showing these clinical signs of Roussy-Levy syndrome. All of these women have displayed gait ataxia, areflexia, pes cavus and sideways curvature of the spine (kyphoscoliosis).

Przypadek rodziny z zespołem Roussy-Levy

Zespół Roussy-Levy, znany również jako dziedziczna dystazja, jest bardzo rzadkim, genetycznie uwarunkowanym zespołem nerwowo-mięśniowym, którego pierwsze objawy najczęściej pojawiają się we wczesnym dzieciństwie. Są to zazwyczaj niepewność chodu, wydrążona, łukowata stopa i brak odruchów ścięgnistych, połączone z występowaniem drżenia spoczynkowego rąk, zaników mięśni dłoni oraz niewielkich zaburzeń czucia powierzchniowego. Przedstawiamy rodzinę, w której w trzech kolejnych pokoleniach stwierdzono kliniczne objawy zespołu Roussy-Levy.