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Mohr-Tranebjaerg Syndrome. A case of a new mutation of gene TIMM8A without affection of the front visual system

Introduction
Deafness and dystonia are the key symptoms of Mohr-Tranebjaerg Syndrome (MTS), an X-linked recessive disease and a mutation within the gene TIMM8A. Visual impairment is a common element of this disease and represents symptoms ranging from subclinical visual impairment to complete blindness. The following name is also used for MTS: deafness-dystonia-optic neuronopathy syndrome. The existence of visual symptoms was associated with neurodegenerative changes in the neurones of the retinae and the visual cortex.

Material and Methods:
We introduce the case of a 46-year-old man with MTS, who was diagnosed with bilateral deafness at the age of two years. At the age of 11, a postural and kinetic tremor (trembling) occurred in the right upper extremity. Three years later, neck dystonia was observed. His dystonia spread gradually, including his torso and upper extremities. At the age of 49, cervical dystonia (laterocollis and anterocollis on the left side) and a dystonic position of the torso were predominant (dominated by the bending towards the right side). The irregular tremor in the right upper extremity was provoked when he stretched both hands and tried to carry out precise movements. The patient used his left upper extremity to control involuntary movements of his right hand. He was diagnosed with increased Patellar reflexes, Patellar tremor and bilateral Babinski symptoms. The patient showed a wide-based gait. He could speak single words that were hard to understand. Result of the Burke-Fahn-Marsden Scale: 38/120 points. The patient was able to move around independently but needed help with everyday activities. Injections of Botulinum toxin led to a significant improvement of the cervical dystonia.

Results:
A new nonsense mutation c. 131 G > A (p. W44X) was found within the gene TIMM8A of his patient and his mother. Other relatives did not give their consent to have their genes tested. The patient did not report any visual impairment. His visual acuity was 0.8 for his right eye and 1.0 for his left. The examination of his ocular fundus showed no changes. The spectral optical coherence tomography showed no changes in the retinal ganglion cells. A neuronal degeneration of the front visual system caused by the neurodegenerative process was ruled out. The examination of the visually evoked potentials showed bilateral P100 potentials with significantly impaired morphology and prolonged latency (left eye 185 ms, right eye 173 ms). An MRI of the brain showed a symmetrical cortical atrophy of the temporal and occipital lobe. Conclusion Data analysis suggests that optic neuronopathy is not a permanent characteristic of the MTS phenotype and the use of the term deafness-dystonia-optic neuronopathy syndrome cannot be justified.