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LETTER TO THE EDITOR

Eye movement abnormalities in a patient with Zellweger spectrum disorder

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Dear Sir,

Zellweger spectrum disorders (ZSDs) are included in peroxisomal biogenesis disorders, a wide spectrum of diseases due to mutations in genes (PEX), leading to loss of peroxisomal metabolic functions. ZSDs have an autosomal recessive transmission and encompass a continuum of three different phenotypes, i.e. Zellweger syndrome, neonatal adrenoleukodystrophy and infantile Refsum disease. The onset is in the newborn period or later in childhood. Though the prognosis is usually poor, milder phenotypes with clinical heterogeneity have been reported [1]. Visual system abnormalities, including retinal degeneration, optic nerve atrophy, cataracts, corneal changes and glaucoma are commonly described in ZSDs [2]. Moreover, pendular nystagmus (PN) is almost always present [3]. However, given the precocious appearance of hypovision and the usually young age of the patients, little is known about quantitative eye movements features in the various phenotypes of ZSDs.

We examined the eye movements features of a 56-year-old man, already diagnosed as having a peculiar mild phenotype of ZSD caused by two heterozigous mutations of the PXMP3 (PEX2) gene (c.355 C>T (p.Arg119X) and c.865_866insA (p.Ser289-LysfsX36), with onset at age 3 years and slow progression. Neurological examination showed predominant cerebellar signs as gait ataxia, dysartria, dysmetria, and gaze-evoked nystagmus, mild retinopathy and bilateral neurorim pallor with preserved good visual acuity, hypoacusia, generalized areflexia, and bilateral pes cavus; brain MRI revealed marked atrophy of cerebellum, cerebellar peduncles and brainstem, particularly of pons, and moderate atrophy of the supratentorial regions, with no signal abnormalities in the white matter. Patient's detailed clinical and biochemical aspects have already been described [1].

Eye movements were recorded by means of an eye tracker device (ASL 504, Applied Science Laboratories, Bedford, MA, USA). An interactive procedure based on nine static points of calibration was performed to ensure a minimization of spatial error. The subject's head movements were minimized by a chinrest.

We recorded horizontal (target of 10° and 18° of amplitude) and vertical (8°) visually guided saccades, elicited, after the disappearance of the central fixation target (1500 ms) and a period of Gap (200 ms), by a peripheral target appearing randomly at left–right or up–down position for 1500 ms. The antisaccade task was analogous to the horizontal visually-guided task, but the subject had to make a saccade opposite to the target. In the fixation task, the patient was required to hold his eye still in response to target in central (90 s) and eccentric (10°–18°, 15 s each) position. Standard saccadic parameters of horizontal and

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