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4.02 Visual electrophysiology testing in infants with congenital vertical nystagmus

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Purpose: In the present study, we describe the visual electrophysiology findings and underlying aetiologies of congenital vertical nystagmus (CVN) with special attention to inherited retinal diseases. The main goal was to establish a strategy of exams in the setting of CVN.
Methods: Our retrospective cohort included all paediatric patients with diagnosis of CVN. All patients underwent complete ophthalmic examination with nystagmus video, visual electrophysiology testing (VEP + ffERG), neuropediatric examination and brain MRI.
Results: We recruited 45 patients with CVN from 2006 to 2017. On the basis of nystagmus video analysis, 35% of infants had an up-beat nystagmus, 30% had a down-beat nystagmus, 25% had a pendular vertical nystagmus, and 10% had a vertico-torsional nystagmus. Overall, 37% of patients had abnormal visual electrophysiology findings (14/45 had abnormal VEP and ERG; 3/45 had abnormal VEP only) and have been diagnosed with anterior visual pathway disease. The main ophthalmic aetiologies of CVN were: stationary photoreceptor dysfunction syndromes (achromatopsia, blue cone monochromacy, congenital stationary night blindness), and early retinal degenerations (non-syndromic Leber amaurosis, Joubert syndrome, neuronal ceroid lipofuscinosis). Two cases of optic nerve gliomas were

discovered. 38% of CVN were "neurological" in origin (inborn encephalopathies). 17% were diagnosed as idiopathic. Interestingly, there were no abnormalities of VEP/ERG in the group of down-beat congenital nystagmus.

Conclusions: Visual electrophysiology testing is highly productive in the setting of CVN. Anterior visual pathway diseases were as frequent as neurological conditions in patients with CVN. In our cohort, stationary retinal dysfunctions and early retinal degenerations were prominent ophthalmic aetiologies of CVN. We suggest that a thorough ophthalmic examination, including visual electrophysiology testing (VEP + ffERG), combined with neuropediatric examination and a brain MRI should be obtained in all infants with CVN.