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9.01 Abnormal flash ERGs in patients suffering from Pelizaeus-Merzbacher disease

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Purpose: Our goal was to describe abnormal full-field ERGs found in patients (N = 10) suffering from Pelizaeus-Merzbacher disease (PMD, OMIM 312080). PMD is a rare recessive X-linked leukodystrophy leading to hypomyelination of the central nervous system. It is caused by abnormal expression of the proteolipid protein PLP due to mutations in PLP1 gene. Although numerous studies described abnormal VEPs with increased latency or absence of response related to central nervous system hypomyelination, ERGs have always been reported normal, most probably due to the fact that retinal axons are not myelinated. To our knowledge, no study combining ERG and VEP recordings in PMD patients has been conducted.

Methods: Full-field ERGs and VEPs were recorded on alert patients with natural pupils after verifying that pupil diameter was at least 3 mm. Flash VEPs were first recorded according to ISCEV protocol with active electrodes at O1 and O2 to evaluate the severity of white matter involvement. Three flash ERGs were subsequently recorded with skin electrodes: dark-adapted combined rod-cone standard flash ERG, light-adapted standard flash cone ERG, and light-adapted 30 Hz flicker ERG to verify the normality of retina functioning. All examinations were performed with Metrovision MonPackOne Monitor and portable stimulator at Lariboisière Hospital, Paris (France).
Results: A total of 10 PMD patients (all male) were included. All had well documented PLP1 mutations. Mean age (range) at VEP/ERG recordings was 10.5 (3–25) years. All 10 patients had abnormal VEPs with abnormal waveforms (n = 2), or decreased amplitude and delayed peak time (n = 5), or non-detectable VEPs (n = 3). Seven of the 10 patients had abnormal full-field ERGs. Amplitude of the flicker ERG was decreased (n = 1, n° 6); amplitude of the b-wave combined rod-cone ERG was decreased or electronegative with normal cone and flicker ERG (n = 3, n° 1, 5, 2), electronegative b-wave of the combined rod-cone ERG with electronegative b-wave of the cone ERG and normal flicker ERG (n = 2, n° 8, 9); amplitude of the b-wave combined rod-cone ERG was decreased with non-detectable cone and flicker ERGs (n = 1, n° 7).
Conclusions: No study combining ERG and VEP recordings had previously been reported in a series of PMD patients. In addition to abnormal VEPs already described in the literature, we found abnormal ERGs in seven of the 10 subjects studied. These abnormalities mostly involved the inner layers of the

neuroretina and cannot be explained only by the primitive hypomyelination described in PMD. We hypothesize a possible retrograde retinal involvement secondary to axon loss in the optic nerve, itself a consequence of the hypomyelination as described in animal models.