Case Description

We report an 8-year-old boy with bilateral optic nerve hypoplasia and cortical visual impairment with an ASTN1 (OMIM#600904) variant. EEG was ordered for screening purposes because there was no history of seizures/epilepsy. EEG showed absence of posterior dominant rhythm and focal needle-like spike-and-wave discharges in the left occipital region (Figure). It is unclear whether the ASTN1 mutation contributed to this patient’s phenotype. EEGs of children with visual dysfunction commonly show absence of posterior dominant rhythm and may show occipital needle-like spikes, which are considered innocuous and unrelated to epilepsy thus a normal EEG variant, and may be due to functional deafferentation.1,2

Figure

Routine EEG

Sensitivity 15 μV/mm, low frequency 1 Hz, high frequency 70 Hz, notch on 60 Hz. Bipolar (A) and average reference (B) showing a run of 100–150 μV focal spike-and-wave discharges with a needle-like morphology in the left occipital region (red arrows)—maximal negativity at O1.
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References

Appendix
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