Oculodigital Sign: A Clinical Clue for Diagnosis

Sir,
A one-and-half-year-old boy, second born to a third-degree consanguineous couple, was brought for the evaluation of poor tracking of objects noticed since early infancy. Mother’s antenatal period and neonatal transition were uneventful. There was history of mild delay in attaining milestones since infancy. Child had one episode of left focal motor seizures in the second week of life and was treated elsewhere with phenobarbitone. Deep-set eyes, vertical nystagmus, and bilateral alternate divergent squint were observed. Prolonged pressing of eyes with thumb [Figure 1] and repetitive eye-poking were observed. The child had poor eye contact and did not fix and follow the light. Menace reflex was absent. Pupils were dilated and sluggishly reacting to light. Fundus examination was normal. Motor system examination was also normal. Hand and leg flapping stereotypies were observed. Differentials of visual impairment due to hypoglycemic brain injury, intrauterine infection, and inherited retinal diseases were considered. Brain MRI did not reveal any structural abnormality. Visual evoked potentials (VEPs) were absent. Clinical exome revealed a homozygous nonsense variation, c.2952C>A in the exon 16 of the GUCY2D gene [ENST00000254854.5; genome assembly: GRCh37.p13; rs1395017892] that results in a stop codon and premature truncation of the protein at codon 984 (p.Cys984Ter). The p.Cys984Ter variant has not been reported in the 1000 genomes, and ExAC databases. The in silico prediction
The prevalence of LCA has been estimated as 1:33,000–1:81,000.[1] Our patient with GUCY2D-associated LCA had visual impairment, nystagmus, and relatively normal fundus in contrast to retinal disease caused by mutation in other genes.[1] Fundus examination findings described in patients with LCA are disc pallor, peripheral pigmentary retinopathy, optic drusen, nummular pigmentation, and falcated retina.[1]

Ocular auto-stimulation may be difficult to control with physical restraints or negative reinforcement. It often tends to decrease with advancing age and may be controlled by engaging in play activities that keep the hands occupied.[3] Oculodigital sign in children must alert the clinicians to search for vision impairment of varied etiology.

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Conflicts of interest
There are no conflicts of interest.

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