

Newborn With Abnormal Eye Movements

Teaching NeuroImages

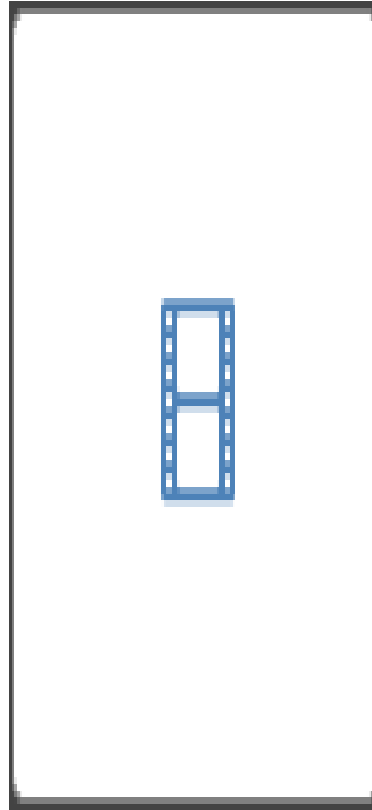
Neurology[®]

Resident & Fellow Section

Vignette

- Full-term female
- Developed focal motor seizures at 1 hour of life
- Day 5 developed paroxysmal non-epileptic abnormal eye-head movements developed (Video)
- Cerebrospinal fluid (CSF) revealed mild hypoglycorrachia, borderline low CSF:serum glucose, and low pyridoxal-5'-phosphate (P5P)

Video



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- Genetic testing showed compound heterozygous mutations in the pyridoxamine 5'-phosphate oxidase (PNPO) gene consistent with P5P-dependent epilepsy
- Aberrant gaze saccades with head jerks are classically described in GLUT-1 deficiency¹
- These eye movements are not described in other conditions with associated hypoglycorrhachia
- PNPO-associated epilepsy has been associated with abnormal eye movements² and should be considered when neonates present with seizures and aberrant saccades with coordinated head movements