Indian Journal of Pediatrics (June 2023) 90(6):623
https://doi.org/10.1007/s12098-023-04560-6

SCIENTIFIC LETTER

Ohtahara and West Syndrome due to Pyridox(am)ine-5-Phosphate Oxidase (PNPO) Deficiency with Novel Phenotype and Good Outcome without Pyridoxal-5′-Phosphate

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Received: 19 February 2023 / Accepted: 7 March 2023 / Published online: 16 March 2023

To the Editor: The Pyridox(am)ine-5-Phosphate Oxidase (PNPO) deficiency is characterized by neonatal seizures which show a good response to pyridoxal-5′-phosphate rather than conventional anti-seizure medications (ASMs) [1, 2]. Here we report a case that responded well to pyridoxine and anti-epileptic drugs (AEDs) with normal development.

A 26-mo-old girl with a normal birth history presented with multiple seizure types on day 3 of life and encephalopathy. Seizure control was poor despite starting multiple ASMs. At 3 mo, the child had no neck control and social smile. On examination, the child was lethargic with spasticity. EEG suggested burst suppression. Ohtahara syndrome was diagnosed which responded to pyridoxine. On investigation, hemogram, renal and liver function, serum ammonia and lactate, arterial blood gas, and Brain MRI were normal. Later, the child developed epileptic spasms after discontinuation of pyridoxine at 8 mo which responded to oral prednisolone. At 14 mo, the child again developed one episode of seizures with fever and was started on levetiracetam post which the child was seizure-free. The whole-exome sequencing done at 25 mo of age, showed a homozygous missense variant in exon-4 *PNPO*: NM_018129.4: c.413G>A: p.Arg138His and segregated with the disorder in the family. No recurrence of seizures was noted after starting Pyridoxal phosphate 15 mg/kg/d.

Here we report a child with Ohtahara syndrome with a good response to pyridoxine, who later developed epileptic spasms responding to prednisolone, occasional fever-triggered seizures on levetiracetam alone, and was seizure-free with normal development till the last follow-up at 26 mo. The differentials considered for PNPO deficiency in the current case include hypoxic-ischemic encephalopathy, and etiologies of Ohtahara syndrome like metabolic, structural, and genetic disorders. The Pyridox(am)ine-5-Phosphate Oxidase deficiency should be considered even in seizures that are responsive to pyridoxine and other ASMs.

Declarations

Conflict of Interest None.

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