

# Wadia Journal of Women and Child Health

## Clinical Image

# A rare case of hypertonia in a neonate

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A 24-day-old full-term female baby (birth weight: 3.6 kg), first by birth order and born to a non-consanguineously married couple, presented with the complaints of generalized stiffness of the entire body (increasing with stimulation) and trismus since day 10 of life. The baby was born through cesarean section with no significant antenatal or perinatal course. Mother received two doses of tetanus toxoid antenatally. Neurological examination suggested rigidity in all muscle groups with opisthotonic posturing [Figure 1], exaggerated startle response, and brisk deep tendon reflexes. The hypertonia responded dramatically to Vigevano's maneuver, which is, forced flexion of head and legs toward the trunk [Figure 2].

Preliminary blood investigations, EEG, and MRI brain were all normal.

In view of the suggestive history, diagnosis of hyperekplexia was suspected and a trial of oral Clonazepam at 0.1 mg/kg was started with Baclofen, which decreased the tone and the startle response.<sup>[1]</sup>

Whole-exome sequencing for suspected hyperekplexia was sent which showed heterozygous missense variation in exon 6 of SLC6A5 gene suggestive of hyperekplexia-3 (HKPX3).<sup>[2]</sup> HKPX3 is caused by homozygous or compound heterozygous mutation in the SLC6A5 gene on



**Figure 1:** Opisthotonic posturing at admission.



**Figure 2:** Vigevano maneuver.

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chromosome 11p15 which can be of autosomal dominant or recessive inheritance. Affected individuals present with neonatal hypertonia, exaggerated startle response to tactile or acoustic stimuli and life-threatening apneic episodes. Being a genetic disorder, effects persists thorough out life albeit in less severity. Although the apnoeic episodes reduce over a period of time, the startle episodes persist and older children may still experience exaggerated startle, periods of rigidity, and recurrent seizures. Parents were counseled regarding the illness and the baby was discharged home on oral Clonazepam, Baclofen, and direct breastfeeding.

#### **Declaration of patient consent**

The authors certify that they have obtained all appropriate patient consent.

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Nil.

#### **Conflicts of interest**

There are no conflicts of interest.

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