Gaze Palsy, Organomegaly, and Inspiratory Stridor: A Diagnostic Triad

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An 11-mo-old girl presented with developmental delay and squint (Supplementary file: Video 1). She was first-born to non-consanguineous healthy parents with a smooth perinatal transition. Examination revealed normal head size, central hypotonia, stridor, massive hepatosplenomegaly, and non-concomitant squint (Supplementary file: Video 1). Investigations revealed a normal brain MRI, pathologically increased chitotriosidase activity ($>1500 \mu\text{mol/L/h}$; reference: $\leq 351.9 \mu\text{mol/L/h}$) and reduced beta-glucocerebrosidase activity ($1.2 \mu\text{mol/L/h}$; reference: $\geq 4.1 \mu\text{mol/L/h}$). The child succumbed to aspiration pneumonia at 13 mo of age. Clinical exome sequencing confirmed the diagnosis [homozygous pathogenic variant-c.1448T > C (p.Leu483Pro) in *GBA* gene]. Both parents were carriers and were advised antenatal counseling.

Acute neuronopathic Gaucher disease is an extremely rare storage disorder with an abysmal prognosis [1–3]. Its incidence accounts for nearly 5% of all patients with Gaucher disease [3]. The deficiency of glucocerebrosidase leads to the accumulation of glucosylceramides in the reticuloendothelial system and brainstem degeneration. The clinical suspicion is primarily based on pattern recognition. Organomegaly, gaze restriction, stridor, and recurrent apneas with relatively normal neuroimaging are sine qua non of acute neuronopathic Gaucher disease. Refractory seizures may occur later in the course of the disease [3]. Although squint is a common association of developmental delay, the importance of careful

ophthalmological examination for gaze restriction in children with developmental delays cannot be understated [4].

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Compliance with Ethical Standards

Conflict of Interest None.

Consent Written informed consent obtained from parents.

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