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Letter to the Editor

Sialuria due to GNE pathogenic variant masquerading as cerebral palsy

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Dear Editor,

Sialuria (OMIM#269921) is an inborn error of metabolism that leads to abnormally high free sialic acid and occurs due to dominant mutations in the *GNE* gene.[1] A total of ten case studies have been published to date. Here, we report the 11th case from the world and the first Indian case of Sialuria, which masqueraded as cerebral palsy.

A 7-year-old male child, without any significant family and birth history presented with developmental delay and seizures from 6 months of age. The child had a global developmental delay with a predominant delay in the motor domain. He currently walks with difficulty, needs help for activities of daily living, and speaks in phrases with dysarthria. Seizures were noted at 6 months of age, with frequency of 1–2 episodes per year, requiring 2 antiseizure medications. The child also had stiffness of limbs starting from 2 years of age. On examination, weight -15.5 kg, height -101 cm, and head circumference -48 cm all are <3. Standard deviation, spasticity in all four limbs, exaggerated deep tendon reflexes, and extensor planters. Hepatosplenomegaly and coarse facies were absent. On investigations, complete hemograms, liver and kidney function tests, ammonia, lactate, blood gases, and tandem mass spectrometry were normal. Magnetic resonance imaging (MRI) brain showed mild cerebral atrophy with dilated ventricles. Exome sequencing identified a likely pathogenic missense variant c.2086G>A, p.(Val696Met) in the *GNE* gene. Parents did not consent to testing. Urinary total N-Acetylneuraminic acid: 1.4 (0.41 ± 0.23 mmol/g creatinine) and Free N-Actylneuraminic acid: 1.0 (0.24 + 0.14 mmol/g creatinine) was elevated.

In summary, the child has a global developmental delay, seizures, and spastic quadriparesis with no organomegaly, which prompted us to think of cerebral palsy, hereditary spastic paraparesis, and arginase deficiency. Exome sequencing gave us a clue like the case identified by Champaigne *et al.*[2] On reverse phenotyping and biochemical investigation, the diagnosis of sialuria was confirmed. Mutations affecting the allosteric site Arg 263 and Arg 266 and other variants like p.Asp84 His have been described.[3] The mutation in the current case does not lie in the allosteric domain and awaits explanation by further functional studies. In conclusion, any child with cerebral palsy and no significant birth history or changes on an MRI brain scan should be investigated for genetic aetiology.

Ethical approval

The Institutional Review Board has waived the ethical approval for this study.

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.

Use of artificial intelligence (AI)-assisted technology for manuscript preparation

The authors confirm that there was no use of artificial intelligence (AI)-assisted technology for assisting in the writing or editing of the manuscript and no images were manipulated using AI.

REFERENCES

1. Enns GM, Seppala R, Musci TJ, Weisiger K, Ferrell LD, Wenger DA, *et al*. Clinical course and biochemistry of sialuria. J Inherit Metab Dis 2001;24:328-36.
2. Champaigne NL, Leroy JG, Kishnani PS, Decaestecker J, Steenkiste E, Chaubey A, *et al*. New observation of sialuria prompts detection of liver tumor in previously reported patient. Mol Genet Metab 2016;118:92-9.
3. Martinez NN, Lipke M, Robinson J, Wilcken B. Sialuria: Ninth patient described has a novel mutation in GNE. JIMD Rep 2019;44:17-21.

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