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SCIENTIFIC LETTER

Genetically Confirmed Case of Aspartylglycosaminuria (AGU)

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To the Editor: Aspartylglycosaminuria (AGU) is a lysosomal storage disorder caused due to mutations in the *AGA* gene [1]. Clinically, the disorder is characterized by mucopolysaccharidosis (MPS) phenotype and radiologically by dysostosis multiplex [2]. A 9-y-old girl born of consanguinity, with uneventful birth history presented with developmental delay, behavioral abnormalities, and progressive abdominal distension from infancy. Child had hyperactivity, did not mingle with others, inappropriate laughing, open mouth, exaggerated startle, and needed assistance in her daily activities like dressing and bathing. On examination, weight: 29 kg (50th to 75th percentile), height: 129 cm (25th to 50th percentile), head circumference: 59 cm. Further, coarse facies, wide nasal bridge, muddy conjuctiva, short neck, open mouth, drooling of saliva, short stubby fingers, protruded abdomen, dry scaly skin and right knee contracture were noted. Systemic examination revealed inattention, dysarthria, spasticity, power of 4/5, exaggerated deep tendon reflexes in all limbs, and hepatosplenomegaly. On investigation, complete blood count, liver and kidney function tests, hearing assessment, skeletal survey and echocardiography, were normal. MRI brain showed corpus callosal thinning, hyperintensities in the bilateral periventricular region with cerebellar atrophy. Exome sequencing showed a novel likely pathogenic homozygous frameshift deletion c.831_835del, p.(Gly279fs*39) in exon-8 of *AGA* gene. Glycosylasparaginase activity from leukocytes showed deficiency, 0.9 nmol/24 h/mg [Normal: 53–122 (89 ± 20)].

Here we report an Indian child who presented with global developmental delay with MPS phenotype. We considered the following differentials: MPS-3, mucolipidosis type-3, alpha mannosidosis type-2 and fucosidosis. Milder forms of MPS type-3 and alpha mannosidosis type-2 also have coarse facial features, developmental delay and behavioral abnormality and is one of the major differentials. Mucolipidosis-3 has similar presentation but has joint involvement. Absence of dysostosis multiplex, hypomyelination and basal ganglia changes ruled out fucosidosis [3, 4]. To conclude, aspartylglycosaminuria should be considered in children who present with phenotype of storage disorder more so mucopolysaccharidosis phenotype.

Declarations

Conflict of Interest None.

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