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Clinical Observations

Biotin and Acetazolamide for Treatment of an Unusual Case of Autism with Lack of Nail and Hair Growth

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ABSTRACT:

BACKGROUND: Patients with autism spectrum disorder (ASD) and developmental delay/encephalopathy rarely demonstrate no or negligible hair and nail growth suggesting a biotin responsive clinical disorder.

PATIENT: A 10-year-old girl presented to Genetic Clinic with features of ASD, isolated headaches and episodes of headaches and limb shaking. The medical history revealed that her hair and nails did not grow. Administration of biotin restored her nail and hair growth and improved intellectual ability and school performance. Her episodes of headaches, single limb shaking and loss of consciousness responded to administration of acetazolamide, and her school performance and social skills further improved.

RESULTS: A de novo c.1091 C>T, p.T364M pathogenic variant was found in the *ATP1A2* gene by whole exome sequencing, but a genetic etiology in the biotin responsive metabolic pathways was not identified. **CONCLUSION:** The combination of biotin and acetazolamide treatment were successful in restoring normal mental function and school performance. Poor or no clinical nail and hair growth in any child with a developmental delay-ASD presentation should be considered as evidence for a biotin-responsive genetic disorder even when Exome testing is negative.

Key Words: Autism Spectrum Disorder, biotin, hemiplegic migraine, *ATP1A2* gene
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