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Whole Exome Sequencing unraveled the mystery of neurodevelopmental disorders in three Iranian families.

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Abstract

Neurodevelopmental disorders are genetically and phenotypically heterogeneous disorders encompassing wide classification of intellectual disabilities (ID) and autism spectrum disorders (ASD). In the present study, we performed whole exome sequencing (WES) in three unrelated patients with developmental delay, hypotonia and ID. We found novel heterozygous mutations in two ID and ASD associated genes namely *POGZ* and *KMT2D* (c.1918C>CT (p.R640RC) in the *POGZ,* c.14531 delG (p.G4844Vfs*13) and c.13818C>CG (p.Y4606YX) in the *KMT2D* genes). Segregation analysis showed all three mutations are *de novo* mutations. Our data add information to the available literature and enlightens a new era of genotype-phenotype correlation. Molecular genetic testing will accelerate and simplify genetic counseling and promote accurate and immediate prognostic information allowing patients to benefit from precise diagnosis and risk assessment schemes for further conception.