**Research paper**

**Targeted next generation sequencing of a panel of autism-related genes identifies an *EHMT1* mutation in a Kleefstra syndrome patient with autism and normal intellectual performance**

István Bock\(^a\), Krisztina Németh\(^b\), Klára Pentelényi\(^c\), Péter Balicza\(^c\), Anna Balázs\(^b\), Mária Judit Molnár\(^c\), Viktor Román\(^d\), József Nagy\(^e\), György Lévay\(^f\), Julianna Kobolák\(^a\), András Dinnyés\(^a\).

\(^a\) BioTalentum Ltd., Gödöllő, Hungary  
\(^b\) Autism Foundation, Budapest, Hungary  
\(^c\) Institute of Genomic Medicine and Rare Disorders, Semmelweis University, Budapest, Hungary  
\(^d\) Laboratory of Neurodevelopmental Biology, Gedeon Richter Plc., Budapest, Hungary  
\(^e\) Laboratory of Molecular Cell Biology, Gedeon Richter Plc., Budapest, Hungary  
\(^f\) Laboratory of Cognitive Pharmacology, Gedeon Richter Plc., Budapest, Hungary

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**Highlights**

- We developed a NGS-based workflow to screen for syndromic causes of autism.
- A pathogenic, de novo mutation was found in an autistic patient with dysmorphisms.
- The mutation leads haploinsufficiency of *EHMT1* mRNA and causes Kleefstra syndrome.
- A *DPP6* missense variant segregated with the autism within the patient's family.
- Multiple genetic factors might contribute to autistic syndromes of KS patients.