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

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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.