A novel variant of KAT6B caused Say-Barber-Biesecker-Young-Simpson syndrome

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Introduction

Say-Barber-Biesecker-Young-Simpson syndrome (SBBYSS, OMIM 603736) is a rare syndrome with multiple congenital anomalies. The phenotype is characterized by distinctive facial appearance with severe blepharophimosis, an immobile mask-like face, a bulbous nasal tip, and a small mouth with a thin upper lip. Herein, we report one de novo heterozygous KAT6B truncaing variants c.5124delC (p.L1709Sfs*5) in a 5-year-old girl presenting with some typical SBBYSS traits.

Method

This study was approved by ethics committees of our hospital and consent was obtained from the patient's parents. Genomic DNA was extracted from peripheral blood and isolated via standard procedures using a QIAamp DNA Blood Mini Kit (Qiagen, Hilden). Sequence analysis was done on a HiSeq 2000 sequencing machine (Illumina, San Diego, CA, USA). Data were filtered by the genetalk software.

Results and discussions

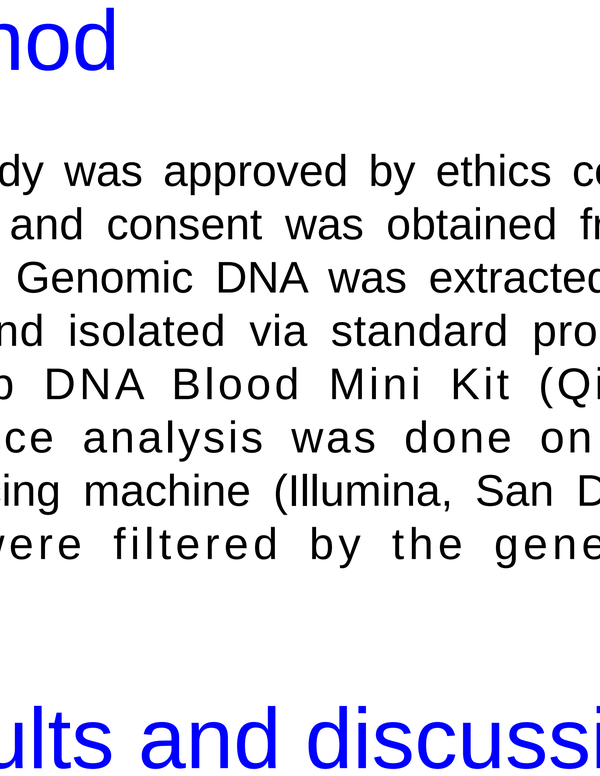

Figure 1. Appearance of the patient at the age of 5 years. A: Facial features including severe blepharophimosis, a bulbous nasal tip; B: Great toes; C: Left hand: abnormally long thumbs; D: Right hand.

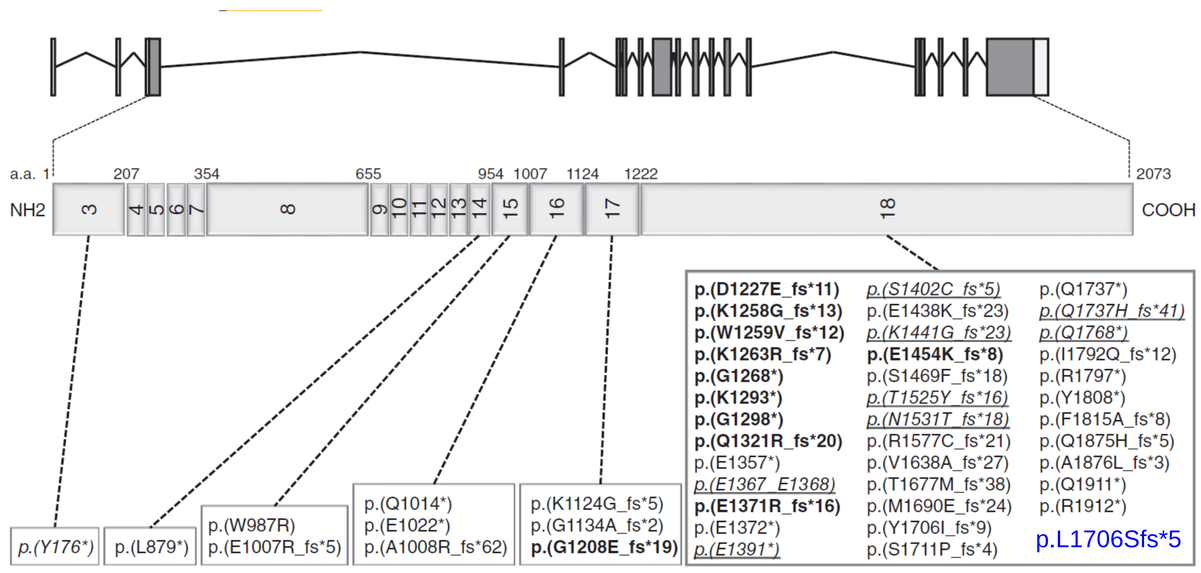

Figure 2 Schematic representation of the KAT6B transcript, and published variants. The sequence variant in exon 3 leading to a not-typical SBBYSS case is shown in italics. (From Clinical genetics. 2019; 95: 253-261)

Table 1 Clinical manifestations of Say-Barber-Biesecker-Young-Simpson syndrome

Category	Features
Major features	Long thumbs/great toes
	Immobile mask-like face
	Blepharophimosis/ptosis
	Lacrimal duct anomalies
Minor features	Patellar hypoplasia/agenesis
	Congenital heart defect
	Dental anomalies (hypoplastic teeth and/or delayed eruption of teeth)
	Hearing loss
	Thyroid anomalies
	Cleft palate
	Genital anomalies (males: cryptorchidism)
	Hypotonia
	Global developmental delay/intellectual disability

(From Clinical genetics. 2019; 95: 253-261)

Conclusion

In the present study, a novel de novo mutaton was identified in the KAT6B gene in a Chinese pediatric patient with ASD symptoms. The truncating variant c.5124delC led to a short protein. The correlation analysis of genotype-phenotype indicated distincetive clinical features comparing with the classical mutations of the KAT6B gene. This work ecriches the mutant spectrum of the KAT6B gene and adds our understanding of the phenotype.

Acknowledgement

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