



Five Novel Variants of *KMT2D*/*KDM6A* Found in Seven Chinese Patients with Kabuki syndrome and a literature review of 39 patients reported in China

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Objective Report 5 new variants of 7 *KMT2D*/*KDM6A* and summarize the clinical manifestations and the mutational spectrum of Kabuki syndrome (KS) by analyzing the reported Chinese cases.

Methods Blood samples were collected for whole-exome sequencing (WES) for 7 patients and their parents if available. Phenotypic and genotypic spectra of 39 previously published unrelated Chinese KS patients were summarized.

Results Genetic sequencing identified six variants (c.3926delC, c.5845delC, c.6595delT, c.12630delG, c.16294C>T, and c.16442delG) in *KMT2D* gene and one variant (c.2668-2671del) in *KDM6A* gene. Of them, 4 variants (c.3926delC, c.5845delC, c.12630delG, and c.16442delG) in *KMT2D* gene and the variant (c.2668-2671del) in *KDM6A* gene were novel. Combining with previously published Chinese KS cases, the patients presented with five cardinal manifestations including facial dysmorphism, intellectual disability, growth retardation, fingertip pads and skeletal abnormalities. In addition, 25% (4/16) patients showed brain abnormalities, such as cerebellar vermis dysplasia, thin pituitary and white matter myelination delay, corpus callosum hypoplasia and Dandy-Walker malformation.

Conclusion We reported five novel variants in *KMT2D*/*KDM6A* genes. A subset of Chinese KS patients presented with brain abnormalities that were not previously reported. Our study expanded the mutational and phenotypic spectra of KS.