Kabuki syndrome (KS) is a rare malformation syndrome caused by mutations in KMT2D and KDM6A genes. Besides the cardinal manifestations, including distinctive facial appearance, mild-to-moderate intellectual disability, dermatoglyphic abnormalities such as persistence of fetal fingertip pads, skeletal anomalies and postnatal growth retardation, a wide spectrum of other anomalies is documented so far. We present clinical features and diagnostic findings in our four patients.

Methods During the 2016-2019 period, four of our patients were diagnosed with KS based on clinical observation and gene-targeted testing or comprehensive genomic testing (exome sequencing).

Results Three of our patients were males, one aged 17 and two aged 3 years, and one female aged 16 years at the time of diagnosis. Mutation of the KMT2D gene was identified in two patients, mutation KDM6A in one patient, while molecular analysis of one male patient is still in progress. In addition to the five cardinal findings, one male patient had cleft palate, one had aortal coarctation with bicuspid aortic valve and one had multiple severe respiratory infections. Although susceptibility to autoimmune disorders is a known feature in these patients, our female patient with KMT2D gene mutation was recognized as the first KS patient with systemic lupus erythematosus.

Conclusion KS is a rare disorder with highly variable clinical features making the diagnosis difficult. Consequently, a close collaboration between pediatric subspecialists, clinical geneticists and molecular biologists is essential in the terms of clinical recognition and genetic confirmation of this syndrome.