

Coexistence of Kabuki Syndrome and Autoimmune Thyroiditis

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Kabuki syndrome (KS) is a multiple congenital anomalies/intellectual disability syndrome characterized by developmental delay, specific facial features, skeletal and visceral abnormalities. This syndrome is caused by mutations in the *MLL2* and *KDM6A* genes. The autoimmune abnormalities have been described in very rare patients with KS. Herein, we present a very rare condition, KS coexisting with autoimmune thyroiditis and vitiligo. A seven-year-seven-month-old girl presented with short stature. There was no consanguinity between her parents. She was born as a term neonate weighing 2100 gram as a twin baby with no perinatal complications. On physical examination, her typical facial features were large and low-set ears, broad and arched eyebrows, elongated palpebral fissures with eversion of the lateral third of the lower eyelid, high and narrow palate. Other phenotypic malformations were numerous vitiligo lesions of different size in the neck, brachydactyly, prominent fetal finger pads, and joint hyperlaxity. Her laboratory findings revealed autoimmune thyroiditis. Thyroid-stimulating hormone (TSH) was 242 mIU/L (reference range: 0.55-6.7) and free thyroxine (fT4) was 0.42 ng/dL (reference range: 0.91-1.92). Anti-microsomal antibody was 450.6 U/mL (reference range: 0-9) and anti-thyroglobulin antibody was 2766 U/mL (reference range: 0-4). Patient's thyroid ultrasonography was consistent with thyroiditis with reduced parenchyma and rough pattern. Levothyroxine-replacement therapy (50 µg/day) induced euthyroid state (TSH: 3.65 mIU/L and fT4: 1.15 ng/dL). We detected a *de novo* heterozygous p.R2471* (c.7411C >T) mutation in the patient. No mutation was detected in the *MLL2* gene in her parents and brother. To our knowledge, this mutation was not reported in KS patients to date.

Key words: Kabuki syndrome, autoimmune thyroiditis, vitiligo, *MLL2* gene, mutation

Crouzon Syndrome with Hypoplasia of Corpus Callosum and Inferior Vermis

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Crouzon syndrome with acanthosis nigricans (CSAN) is a clinically and genetically distinct entity caused by a mutation in the *FGFR3* gene, featuring craniosynostosis, characteristic facial features, and atypical and extensive acanthosis nigricans. CSAN is genetically and clinically different from classic CS. We report a CSAN case with neurological findings as hypoplasia of corpus callosum and inferior vermis. A ten-month-old girl presented with a facial dysmorphism at birth. There was no consanguinity between her parents. She was born as a term neonate weighing 3380 g with no perinatal complications. She has undergone surgery because of choanal atresia at the ninth day of life. Additionally, she has undergone coronal craniotomy surgery because of craniosynostosis due to bilateral coronal stenosis at the age of nine months. On physical examination, her atypical facial features were midface hypoplasia, hypertelorism, craniosynostosis, brachycephaly, maxillary hypoplasia, exophthalmos, bilateral distinctive and low-set ears. Other phenotypic findings were lateral nistagmus bilaterally and widespread acanthosis nigricans on all of curve regions as neck, bilateral axillae. Cranial magnetic resonance imaging revealed hydrocephaly and hypoplasia of corpus callosum and inferior vermis. There was no pathology in abdominal ultrasonography and echocardiography. We detected a *de novo* heterozygous *A391E* (c.1172C>A) mutation in our patient. No mutation was detected in the *FGFR3* gene in her parents and sisters.

Key words: Crouzon syndrome, hypoplasia, corpus callosum, inferior vermis, *FGFR3*