Clinical Genetics

AB060. A family with three children of rare intellectual disability syndrome

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Background: Intellectual disability (ID) with dysmorphic features can be caused by many genetic disorders, which commonly overlap with one another, making specific diagnosis difficult.

Methods: Dysmorphology examination of face, limbs, and external genitalia was performed and followed by dysmorphology analysis with POSSUM and Face2Gene software.

Results: A 15-year-old girl with ID, nonspecific muscle weakness, and failure-to-thrive was identified. She sat at the age of 1.5 years and walked at 7 years. She had her menarche at the age of 15 years, and irregular menstrual cycles and oligomenorrhea. Physical examination revealed short stature (117 cm, <3rd percentile), underweight (22 kg, <3rd percentile), and small head circumference (51 cm, <3rd percentile). Dysmorphic features included strabismus, frontal bossing, cupid bow lips, and high arched palate, low posterior hairline, sparse hair, clinodactyly of the 4th finger, disharmony toes and syndactyly of 2nd–3rd toes. Genital examination showed labia minor protrusion and minimal spurt at labia major. Echocardiography and renal ultrasonography are ongoing. Family history revealed parental consanguinity, and that the patient was the 2nd child. Her oldest and youngest sisters were reported to have similar conditions with more severe disability, both died at 7.5 and 9 years, respectively. The youngest sister could not communicate verbally. She cried when urinating. She had keratitis and cataract of both eyes, joint contracture, spasticity with severe muscular atrophy. Detailed clinical data of the oldest daughter was unavailable. All three children were born with low birth weight. The analysis software revealed a possible diagnosis of Verheij syndrome (8q24.3 microdeletion), which is characterized by antenatal and postnatal growth retardation, microcephaly, vertebral anomalies, joint laxity/dislocation, developmental delay (DD), cardiac and renal defects and dysmorphic features.

Conclusions: Dysmorphology analysis software is a very useful tool for clinician to make a specific diagnosis for highly heterogeneous conditions such as ID with dysmorphic features.

Keywords: Intellectual disability (ID); rare disease; Verheij syndrome

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