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Williams-Beuren syndrome

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Williams-Beuren syndrome, Mendelian inheritance in Man number 194050, is a genetic disorder caused by microdeletion of the Williams-Beuren syndrome chromosomal region on chromosome 7, spanning 1.5 million to 1.8 million base pairs and containing around 26–28 genes. The genotypic and phenotypic correlation is still not fully understood. Clinically, patients present with multiple systemic disorders include distinctive facial features (Fig. 1). Infant hyperglycemia, feeding difficulties, colic, and growing problems may be encountered. The adult height is usually below the third percentile. Gross developmental delays, poor physical co-ordination, and connective tissue abnormalities such as overly loose joints are obvious. Moderate intellectual disability but with specific strengths in verbal short-term memory and language, overfriendliness, and an empathetic personality are commonly observed. Elastin arteriopathy occurs in 75% of patients with this syndrome. Any artery may be narrowed, and peripheral pulmonic stenosis is common in early infancy. Supravalvular aortic stenosis is the most common arteriopathy requiring surgical correction. Genetic diagnosis using fluorescence *in situ* hybridization to demonstrate deletion of the Williams-Beuren syndrome chromosomal region, which encompasses the elastin gene, is the mainstay of laboratory diagnosis (Fig. 2).

Our patient was a girl aged 4 years and 8 months. The diagnosis of Williams syndrome was made soon after birth by dysmorphic

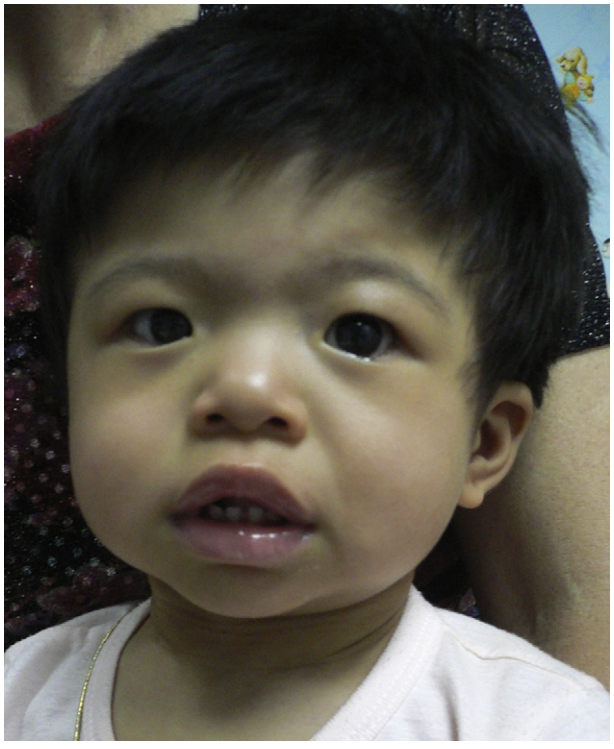

Fig. 1. Typical facial features of Williams syndrome: broad forehead, periorbital puffiness, flat nasal bridge, upturned nose tip, full cheeks, full prominent lips, wide mouth, increased interdental spaces, and a small chin. (Published with permission from the patient's parents and grandparents.)

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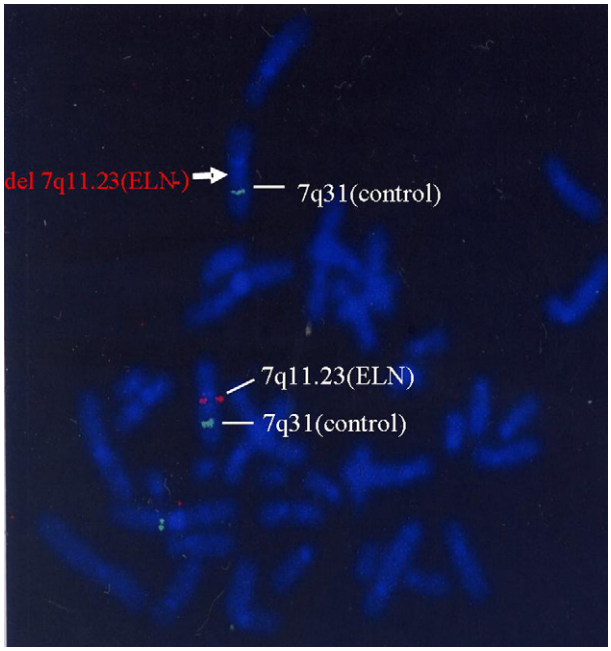

Fig. 2. Fluorescence *in situ* hybridization results. Green signal: control (7q31); red signal: Williams-Beuren syndrome chromosomal region (*ELN*) probe. A red signal is missing. White arrow demonstrates the deleted allele of *ELN*.

facial features and fluorescence *in situ* hybridization study. No hyperglycemic episodes, hypercalcemia, feeding difficulties, or cardiovascular disease was found. Follow-up of her growth showed that her body weight and height were all within the 10th to 25th percentiles. She was very friendly, although Bayley Scales of Infant Developmental assessment revealed significant delay in the mental development index (<50, <0.1 percentile). Comprehensive biopsychosocial and behavioral health care supervision is being carried out through an interprofessional medical team (Fig. 3).

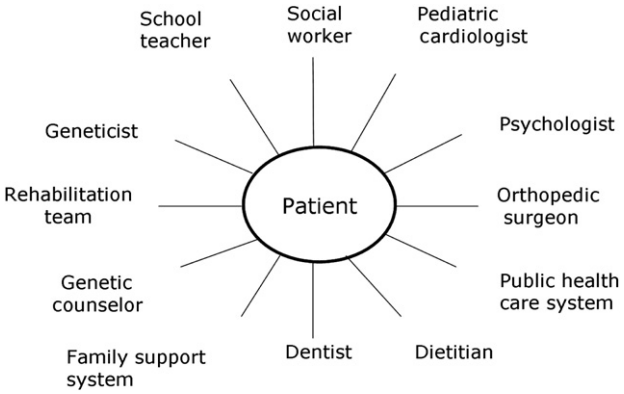

Fig. 3. Interprofessional patient care model.

Further reading

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