Williams Syndrome: A Case Series

SUBAPRIYA KANDASAMY, DEEPTI SAXENA, YOUGAL KISHORE AND SHUBHA R PHADKE

From Department of Medical Genetics, Sanjay Gandhi Postgraduate Institute of Medical Sciences, Lucknow, UP, India.

Correspondence to: Dr Shubha R Phadke, Professor and Head, Department of Medical Genetics, Sanjay Gandhi Postgraduate Institute of Medical Sciences, Lucknow, Uttar Pradesh, India. shubharaophadke@gmail.com

Pediatricians' awareness about malformation syndromes can help in their timely diagnosis. Williams syndrome is a microdeletion syndrome associated with characteristic facial features and behavioral phenotype. Diagnosis can be confirmed by fluorescence-in-situ hybridization or multiplex ligation probe amplification. Correct diagnosis can help in diagnosing hypercalcemia and cardiac defects, and providing genetic counseling to the family.

Keywords: *FISH, Microdeletion, Williams syndrome.*

Williams syndrome (OMIM 194050) is caused by hemizygous microdeletion on chromosome 7; the estimated prevalence varies from 1: 20000 to 1:50000 live births. It is characterized by cardiovascular disease, distinctive facies and personality, mild intellectual disability, unique personality, and connective tissue growth and endocrine abnormalities. The aim of this communication is to describe the typical facial and other diagnostic features of Williams syndrome.

The data about the facial and other phenotypic features of 11 patients (8 boys) of Williams syndrome diagnosed at our center from 2007 to 2012 were collected. The diagnosis was confirmed by Fluorescence-in-situ- hybridization (FISH) or Multiple Ligation Probe Amplification (MLPA).

The mean age was 8 years. The presenting complaints were developmental delay and/or intellectual disability (in 6 children) or hyperactive behaviour (in 2 children). ***Fig.* 1** shows facial features of 9 children with Williams syndrome. Malar hypoplasia, thick lower lip and bulbous nasal tip were present in 10 cases; periorbital fullness and long philtrum were seen in 8 cases. Seven patients had bi-temporal narrowing and full cheeks, while 6 had short nose and wide mouth. Prominent ear lobes were seen in 5 patients. Epicanthal folds, small jaw, malocclusion of teeth and widely spaced teeth were seen in 3 children. The cardiac anomalies (aortic stenosis, pulmonary stenosis, ventricular septal defect and mitral valve prolapse) were present in 5 out of 9 children in whom echocardiography was done. Four patients had short stature and six had microcephaly.

Jurado, *et al*. [1] suggested that postnatal growth deficiency may be associated with deletion of maternal copy of chromosome 7. One of our patients presented at 3.5 months of age with cholestatic jaundice, paucity of bile ducts and facial dysmorphism. The diagnosis of Williams syndrome was made at 1 year when developmental delay and characteristic facial features became obvious.

The facial phenotype of Williams syndrome is characteristic and is seen in all children. The fullness of cheeks is lost with age and face becomes long and coarser. Cardiac malformations are commonly seen and are a

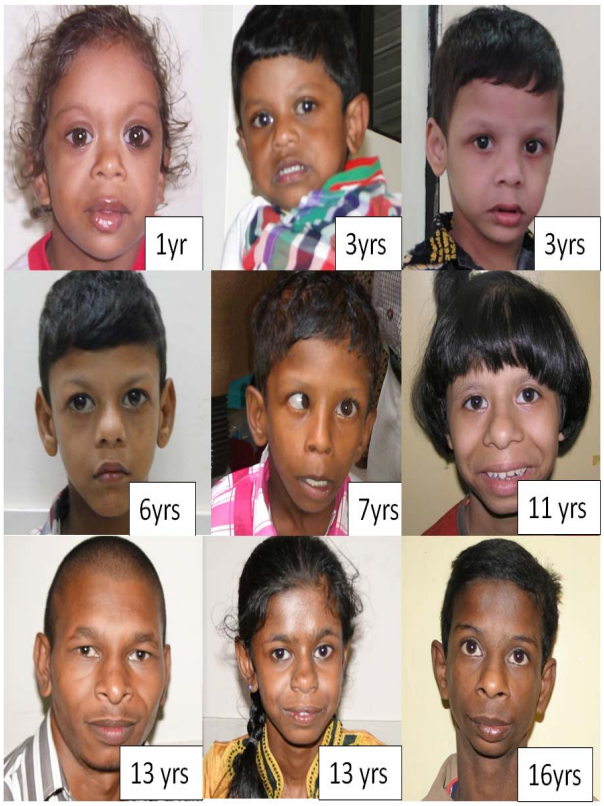

FIG 1: *Facial features of Williams syndrome.*

common reason for referral [2,3]. In a study from Brazil [4], out of 23 clinically suspected cases, 15 were confirmed to have Williams syndrome. In addition to characteristic facial phenotype, joint laxity, cardiac malformation and overfriendly personality help in clinical diagnosis. Rauch, *et al*. [5] reported the prevalence of William syndrome to be 1.3% in cases with intellectual disability.

There was no infant that had hypercalcemia in our case series. The photographs in this series are expected to give visual impression of the facial features that can help in diagnosis by gestalt.

Acknowledgement: Indian Council for Medical Research.

REFERENCES

1. Pérez Jurado LA, Peoples R, Kaplan P, Hamel BC, Francke U. Molecular definition of the chromosome 7 deletion in Williams syndrome and parent-of-origin effects on growth. Am J Hum Genet. 1996;59:781-92.
2. Patil SJ, Madhusudhan BG, Shah S, Suresh PV. Facial phenotype at different ages and cardiovascular malformations in children with Williams-Beuren syndrome: a study from India. Am J Med Genet A. 2012;158A:1729-34.
3. Ferrero GB, Biamino E, Sorasio L, Banaudi E, Peruzzi L, Forzano S, *et al*. Presenting phenotype and clinical evaluation in a cohort of 22 Williams-Beuren syndrome patients. Eur J Med Genet. 2007;50:327-37.
4. Viana MM, Stofanko M, Gonçalves-Dornelas H, da Silva Cunha P, de Aguiar MJ. Phenotype of Williams-Beuren syndrome in Brazilian patients: comments on the article by Patil, et al. [2012] and discussion of variable phenotypes in distinct populations. Am J Med Genet A. 2013;161A:637-8.
5. Rauch A, Hoyer J, Guth S, Zweier C, Kraus C, Becker C, *et al*. Diagnostic yield of various genetic approaches in patients with unexplained developmental delay or mental retardation. Am J Med Genet A. 2006;140:2063-74.

Accuracy of Mothers' Perceptions of Their Child's Weight Status

R JANI, *S MIHRSHAHI, †S MANDALIKA AND ‡KM MALLAN

*From the Institute of Health and Biomedical Innovation, *School of Population Health, and ‡Institute of Health and Biomedical Innovation and School of Exercise and Nutrition Sciences, QUT, Brisbane, Australia and †College of Home Science, Nirmala Niketan, (affiliated to University of Mumbai), India.*

Correspondence to: Dr KM Mallan, Institute of Health & Biomedical Innovation, Queensland University of Technology, 60 Musk Ave, Kelvin Grove, Brisbane QLD 4059, Australia. kimberley.mallan@qut.edu.au

The study examined the accuracy of maternal–perceived child weight. Urban affluent mothers of 111 children aged 2-5 years were recruited. Nearly a quarter of mothers overestimated their underweight child as 'normal weight' and all overweight/obese children were perceived as 'normal weight'. Mothers, therefore, were unable to recognize their child's true weight status.

Keywords: *Obesity, Preschool children, Underweight.*

For a comparable Body mass index (BMI), Indian children have higher body fat (2-8%) and greater risk of disease (e.g. higher insulin resistance) than Caucasian children [1]. Mothers play a role in shaping the early (0-5 years) eating and activity behaviors related to childhood obesity [2]. Poor maternal recognition of their child's true weight status could therefore be a potential risk factor for obesity. In a systematic review, 6-73% of predominantly Caucasian parents under-estimated the weight status of children aged 2-12 years [3]. The accuracy of maternal perceived child weight has been examined in populations (e.g. Caucasians and Hispanics in US) where obesity is a public health concern [3], but not in populations of urban affluent Indian children, for whom obesity is an increasing concern [4]. The present study examined the accuracy of mothers' perception of their preschool (2-5 years) children's weight, in Mumbai.

In total, 111 mothers with children aged 2-5 years (mean±SD, 44.1±9.7 mo) attending private medical clinics (n=5) in the affluent areas of Mumbai were recruited after receiving approval from the QUT Human Research Ethics Committee, Australia. Child's height and weight were measured by the researcher using standard equipments/protocols. Maternal perception of the child's weight status was measured using a single item from the NOURISH questionnaire [5]: Do you think your child is...(*i*) underweight, (*ii*) healthyweight, (*iii*) somewhat overweight, (iv) very overweight, or (v) don't know.

According to the BMI z-score categories [6], 51.4% of children were healthyweight. The prevalence of