IMAGES IN MEDICINE

Dental anomalies in Williams syndrome

A 2 year old boy with Williams syndrome was undergoing preoperative evaluation for supra-valvular aortic stenosis. On examination his mouth was wide with full lower lip. He had small, irregular, widely spaced, peg shaped primary teeth with extensive caries (fig 1). These are the typical anomalies of primary teeth in Williams syndrome. In adulthood teeth tend to be crowded. His ionised serum calcium concentration was 1.6 mmol/l (normal 1.1-1.4 mmol/l), and 25-dihydroxycholecalciferol 454 pmol/l (36-144 pmol/l). The serum 25-hydroxycholecalciferol and phosphate levels were normal. He is on calcium, and a vitamin D restricted diet, and is undergoing treatment for dental caries. He is awaiting balloon dilatation for the supra-valvular aortic stenosis.

Williams syndrome usually occurs as a sporadic new dominant condition, although parent-child transmission has been reported.^{1 2} Williams syndrome is caused by a submicroscopic chromosomal deletion on one of the number 7 chromosome pair at 7q11.23. Routine chromosome analysis is usually normal and the deletion is detected by fluorescent in situ hybridisation (FISH) studies using a probe for the elastin gene.^{3 4} Deletion of the elastin gene in Williams syndrome is associated with isolated supra-valvular aortic stenosis and other cardiovascular abnormalities. Williams syndrome, with more protean connective tissue abnormalities and mental retardation, is associated with deletions that include the elastin gene and a gene encoding the protein kinase LIM-kinase-1.

Figure 1 Dental anomalies in patient (photograph reproduced with permission).

A S KASHYAP
Department of Medicine,
Armed Forces Medical College,
Pune 411040, India

H S SHARMA
P KUMAR
Department of Dental Surgery,
Armed Forces Medical College,
Pune 411040, India

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