IMAGES IN MEDICINE.....

Facial and dental appearance of Williams syndrome

Figure 1 Typical facial appearance of a Williams syndrome patient showing wide mouth with long philtrum and thick lips (published with parent's permission).

Figure 2 Orthopantomogram tomographic radiography of a typical Williams syndrome patient showing aplasia and tooth resorption anomaly, with fan shaped positioning of the front teeth.

Williams syndrome is a genetic condition, which predominantly occurs as a sporadic disorder, although some families have been reported showing autosomal dominant inheritance with varying penetrance. The incidence of Williams syndrome is estimated to be one in 20 000–50 000 live births, and it consists of supravalvular aortic stenosis, characteristic dysmorphic facial features named “elfin face” (wide mouth with long philtrum and thick lips; fig 1), mental retardation, and other clinical manifestations including transient infantile idiopathic hypercalcaemia, growth retardation, and frequent dental problems.¹ The syndrome is caused by a submicroscopic deletion in the chromosome 7 implicating the 7q11.23 region (elastic gene).² Early diagnosis of the syndrome is important since many of its features require treatment, and the prognosis can be dramatically improved by timely management. The clinical complexity of this developmental disorder is well known, making it difficult to diagnose on the clinical picture, while genetic testing is expensive and it is not cost effective to screen all patients. A dentist can be of help in diagnosing Williams syndrome early.

We examined 33 children with Williams syndrome, proved by fluorescent in situ hybridisation at the pedodontic and orthodontic department of Semmelweis University in Budapest, Hungary. We found that typical dental findings in Williams syndrome are dental aplasia, which occurred in 90% of our patients, and primary tooth resorption anomaly, which we found in 96% of our patients. Also, fan shaped positioning of the front teeth was seen in the majority of our patients (fig 2). We highlight the fact that early orthodontic consultation is very important to avoid further dental anomalies in Williams syndrome.

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