



Oral manifestations seen in association with a case of trisomy for the short arm of chromosome 9

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Abstract

A case of trisomy for the short arm of chromosome 9 in a 13-year-old boy is described. Particular emphasis is placed upon a number of abnormal dental findings, which include enamel hypoplasia, hypodontia, and severe dental crowding. The difficulties of providing comprehensive dental treatment in cases such as these is discussed. (Pediatr Dent 18:465-68, 1996)

Trisomy for the short arm of chromosome 9 (9p+) was first described in 1970 by Rethore et al.¹ Five children were reported who had extra chromosome substance identified as the short (p) arm of chromosome 9. Although this was prior to the widespread availability of banding techniques, the children exhibited sufficient common clinical features to suggest a new syndrome. In 1973 Rethore et al.² summarized the nine cases reported up to that time and added one of their own. Since then, more than 125 cases have been reported³ and 9p trisomy is one of the best known chromosomal disorders.

The most important clinical features of trisomy 9p are summarized in Table 1. There is a 2:1 female predilection with classic features being the characteristic facies, variable mental retardation, and hypoplasia/dysplasia of the terminal phalanges.⁴

Patients who are trisomic for the entire short arm of chromosome 9 have a well-defined phenotype, however there have been several other reports of complete and partial trisomy for segments of 9p. Lewandowski⁵ described three patients trisomic for only the distal half of the short arm of chromosome 9 (p21—pter), which represents a milder form of the trisomy 9p syndrome. Alfi et al.⁶ described three patients with deletion of the distal third of the short arm (p22—pter) with partial antithetical features of the trisomy 9p syndrome. It would appear that the distal one-third to one-half of the short arm of chromosome 9 is important in the partial expression of trisomy 9p syndrome. Thus, the clinical features in general vary with the amount of excess chromosomal material present, as seen in the mild neuro-

logical retardation of partial trisomy 9p, moderate retardation of trisomy 9p, and neonatal death in trisomy 9.⁷

The purpose of this article is to describe another case of partial trisomy of chromosome 9p, with particular emphasis on a number of unusual oro-dental features that were seen in association with this case.

TABLE 1. PRINCIPAL CLINICAL FEATURES OF TRISOMY 9P

<i>Facies</i>
<i>Head</i>
High, broad forehead
Mild microbrachycephaly
Flat occiput
Large fontanelle and open metopic suture in childhood
<i>Eyes</i>
Small eyes, deep set in their sockets
Horizontal or down-slanted palpebral fissures
Mild hypertelorism
<i>Nose</i>
Large, full nose with globular tip
Downturned nares
<i>Mouth</i>
Large mouth with downturned angles
Everted lower lip
Micrognathia
High arched palate
<i>Hands</i>
Hypoplasia ± dysplasia of terminal phalanges (particularly digits 2 and 5)
Single palmar crease
Cyanosed hands and feet
Dysplastic, claw-like nails
Disproportionately long palms for fingers
<i>Variable Mental Retardation</i>
IQ from 30 to 65
Speech impairment common

Case report

This 13-year-old boy was referred to the Department of Children's Dentistry of the Eastman Dental Hospital by a community dental officer for his dental management because of poor cooperation. He had also been previously seen by a consultant orthodontist for an opinion. Trisomy of the short arm of chromosome 9 had been diagnosed, and the child was under the care of a consultant pediatrician.

Medical history

This boy was born of a pregnancy complicated by oligohydramnios at 37 weeks of gestation via spontaneous vaginal delivery. He was the first baby of a 32-year-old mother. Birth weight was 1.76 kg, well below the 10th centile. The child remained in the Special Care Baby Unit until he weighed 2 kg, when he was discharged.

The child had a number of dysmorphic features: a wide carp-shaped mouth, micrognathic lower jaw, thick lips, drooling, long beaky nose with narrow nasal passages, hypertelorism, low set ears with bilateral pre-auricular pits, and narrow auditory canals. Bilateral blocked tear ducts also were present. He had proximally inserted thumbs with bilateral clinodactyly and left transverse and partial right transverse palmar creases. The hands were large, erythematous, swollen, and often cold. Large big toes were also present, the third toe underlying the second toe with marked valgus hindfoot. The testes were initially undescended, became palpable in the upper part of the scrotum at age 2 or 3 years, but then retracted and became undescended. He also had bilateral inguinal herniae and bilateral orchidopexy and herniorrhaphy. Computerized tomography revealed a left ventricle a little larger than the right. There was no cerebellar vermis and the cisterna magna was in continuity with the region of the fourth ventricle. There was no abnormality in cerebral substance.

His growth was well below and diverging from the third centile. At age 2 years and 4 months his length (77 cm) was 3.5 SD below the mean, and by age 6 years and 4 months his height (90 cm) was 5.4 SD below the mean. The bone age was always retarded, and at a chronological age of 2 years, the ossification centers of the capitate and hamate had not appeared (both are usually seen at 3 months). At a chronological age of 6 years the bone ages of the phalanges and the carpal bones corresponded to those of the 5-year and 2-year standards, respectively. At age 7 years and 8 months, with a height of 98 cm he was started on growth hormone that was still being given at 14 years. At this age he was still 4 SD below the mean. Pubic hair developed at age 13 years.

The child was severely mentally retarded with little or no speech. He did not walk until age 5 and showed inversion of both feet. At 5 years he developed epilepsy. Thyroid function tests were normal, but an insulin

stress test gave a blood glucose level of 2.1 mmols with a maximum growth hormone level of 7.7×10^{-6} L. Thyroid-stimulating hormone and cortisol level were normal during the insulin stress test. The rise in testosterone following HGC was normal.

The child was hospitalized on several occasions because of seizures and upper respiratory infections. At age 12 years he underwent operations to correct his bilateral undescended testicles and a blocked tear duct.

Chromosome studies

The chromosomes were first assessed shortly after birth in 1980 and were found to be apparently normal on low-resolution GTG-Banding. A provisional diagnosis of Coffin-Siris syndrome was made in 1982. The chromosome examination was repeated in 1985 and on higher-resolution GTG-Banded chromosomes it was found that there was an unbalanced translocation, with an unidentified segment attached to the short arms of chromosome 10. Blood from both parents was karyotyped and the mother was found to be a carrier of a balanced reciprocal translocation, 46, XX, t(9,10)(p13;p15).

The child was therefore trisomic for the region 9p13—pter and monosomic for the telomere region of 10pter, due to inheritance of the derived chromosome 10, 46, XY, -10, +der (10)t(9;10)(p13;p15) mat. Following this, the diagnosis was changed to trisomy for the short arm of chromosome 9.

TABLE 2. SUMMARY OF THE CLINICAL AND RADIOGRAPHIC DENTAL FINDINGS

Teeth Present	$\frac{764321}{6E321} / \frac{12456}{1234E6}$
Teeth Unerupted	$\frac{85}{75} / \frac{7}{578}$
Incisors	Uncomplicated crown fractures $\frac{1}{2}$ Proclined upper incisors Severe crowding (lower central incisors labially placed) Marked attrition of lower incisors Pitted enamel hypoplasia
First Permanent Molars	All had occlusal carious lesions Taurodont
Hypodontia	$\frac{\quad}{38}$ 84/
Periodontal	Poor oral hygiene Generalized marginal gingivitis throughout

Dental history

A complete dental examination at initial presentation was not possible because the patient couldn't cooperate. We decided to arrange for a general anaesthetic to allow a thorough clinical and radiographic examination and to perform any necessary dental treatment. The findings are summarized in Table 2.

The first permanent molars were restored occlusally with amalgam and the maxillary incisors with etched composite resin. It was decided that the labially placed mandibular central incisors should be extracted to relieve anterior crowding. These were sent for histological analysis.

Discussion

The development of differential banding techniques for human chromosomes has resulted in the reporting of large numbers of partial duplication syndromes in the literature. It has been shown that chromosome number 9 is particularly susceptible to breakage and rearrangement.⁸ The syndrome of trisomy 9p has been described in more than 120 cases and was reviewed extensively by Centrewall et al.⁹ The child we describe showed a number of oral abnormalities, some of which have not been described previously.

The case we discuss showed hypodontia of the upper left canine, lower right first premolar, upper left third molar, and lower right third molar (Figs 1 and 2). The absence of a lower first premolar and upper canine is relatively unusual.¹⁰ Although hypodontia is not well described in association with trisomy 9p, the presence of microdontia (which is often associated with hypodontia) has been described.¹¹ Taurodontism of molars, seen in our case, is also associated with hypodontia.¹² and can present possible difficulties with endodontic treatment or extraction.¹³ The failure of the lower second premolars and second molars to erupt may be related to retarded somatic growth. Delayed eruption has been described in association with trisomy 9p¹¹ and is also a feature of hypodontia.¹⁴

The boy had enamel pitting of all the teeth; the incisors were particularly severely affected, showing severe pitting of a random nature on the labial faces of all the crowns (Fig 3). Such enamel pitting has not been described specifically in association with trisomy 9p, although the presence of dystrophic teeth has been recorded.¹⁵ Pitted enamel has been described in association with other conditions exhibiting severe seizure activity such as tuberous sclerosis,¹⁶ pit-shaped enamel defects being most evident on examination of the labial faces of the premolar surfaces.¹⁷

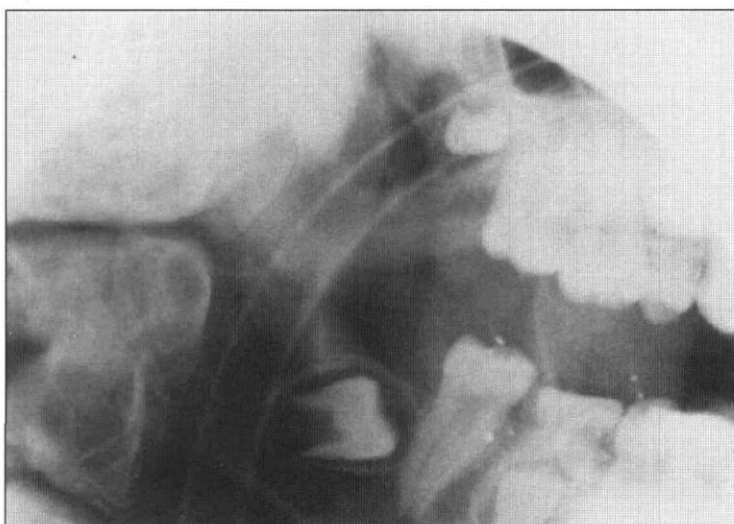


Fig 1. Right bimolar radiograph at age 13 years.



Fig 2. Left bimolar radiograph at age 13 years. These radiographs confirm the failure of the maxillary left permanent canine, mandibular right premolar, maxillary left and mandibular right third molars to develop.

The shape of the jaws has been well described in trisomy 9p—a narrow, high-arched palate being seen in 60% of patients in one review.³ The presence of mandibular retrognathia also has been described.^{18,19} Such jaw anomalies would certainly contribute to dental crowding, and the severe lower incisor crowding seen in our case (Fig 4) almost certainly was influenced by a degree of retrognathia. It is interesting to note that the presence of exposed upper teeth also has been described.²⁰ This was indeed seen in our case, and a contributory factor would certainly be the retruded lower jaw and resulting lower lip trap proclining the upper incisors. Cleft lip with or without cleft palate has been described in association with trisomy 9p as an occasional feature,¹⁷ having a frequency of between 16%³ and 33%.⁴ There is certainly an association between cleft lip and palate and dental arch crowding, but it was not possible to obtain any correlation between the two from

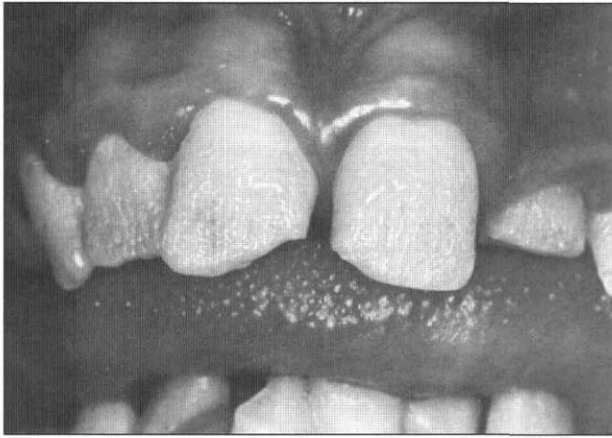


Fig 3. Marked random pitted enamel hypoplasia of the upper and lower permanent incisors.

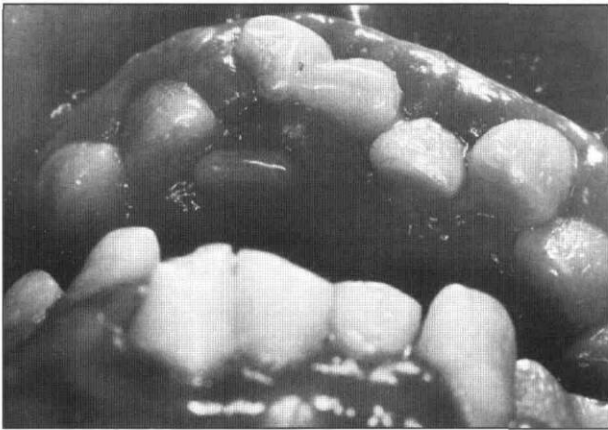


Fig 4. Mirror view showing severe lower labial segment crowding.

the literature because there are few records of the respective children's malocclusions. There was, however, no evidence of clefting in the case we describe.

It is evident from the literature that children with trisomy 9p syndrome frequently have a number of dental abnormalities. The degree of mental retardation seen in these patients causes problems in oral hygiene maintenance and dental treatment. Our case exhibited carious lesions that required intervention, and it is not surprising, in view of frequent crowding and poor oral hygiene, that caries has been reported before.^{5, 11} It is unfortunate, but inevitable, in these cases where the degree of mental retardation results in such poor cooperation that general anaesthesia is usually required to facilitate dental treatment. The dental treatment for this case was relatively straightforward, with the restorations required being fairly minimal. Difficulties arise when the treatment required is more extensive; decisions then have to be made regarding extractions versus complex restorations to save teeth. In this case, the decision was made to extract the lower central incisors to help alleviate crowding and so improve oral hygiene, thereby minimizing the risk of periodontal disease. It was felt that any form of orthodontic appliance therapy

to align the lower incisors was impossible. As with all such difficult management cases, a degree of compromise will always be required.

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