Orofacial findings and dental treatment in 18r- syndrome: case report

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Introduction

Ring formation of chromosome 18 (18r- syndrome) results from the "welding" of the two ends of the chromosome after deletion of both terminal segments. In 1969, de Grouchy collated the findings in 53 reported cases of autosomal aberrations of chromosome 18. These comprised 23 cases of the 18p- syndrome in which there is a deletion of the short arm of the chromosome, 20 cases of the 18q- syndrome in which there is a partial deletion of the long arm of the chromosome, and ten cases of 18r- syndrome. The 18q- syndrome is a clinically well-defined syndrome. The 18p- syndrome has many malformations in common with 18q- syndrome although a well-defined syndrome does not emerge from the reported cases. However, the diagnosis of 18r- is almost impossible a priori, but may be suspected when a combination of symptoms belonging to the 18p- and 18q- syndromes occurs.

Intellectual deficit is constant in 18r- syndrome. Its intensity varies from borderline to severe mental retardation. Newborns are hypotrophic and hypotonic, seizures have been reported, and microcephaly is nearly constant. Features of the 18p- syndrome reported in 18r- patients include hypertelorism, bilateral epicanthic folds, and — less frequently — abnormal slanting of the palpebral fissures and strabismus. Both high-arched palate and cleft palate have been reported frequently. The most commonly observed feature of the 18q- syndrome in 18r- patients is the carp-shaped mouth. The ears are often low set, but not malformed.

Other malformations reported in 18r- patients common to both 18p- and 18q- syndromes include: congenital heart disease, renal malformations, club feet, and hip dislocations. de Grouchy reported dental caries and micrognathia in the 18p- syndrome. In 1982 Taylor and Peterson reported a case of 18p- syndrome and its craniofacial and dental implications. They described a child with a retrognathic mandible, a tendency to an anterior open bite, a carp-shaped mouth, and a high palatal vault. The child had chronic marginal gingivitis, but few carious lesions.

Case report

A female (OM) aged three years four months with nursing bottle caries was brought for dental treatment to the pediatric dentistry department of the Hadassah Faculty of Dental Medicine, Jerusalem, Israel. She had been referred previously to the oral surgery department because of a large and asymmetric tongue. The child had been born without problems in the 38th week of an uneventful pregnancy and weighed 2.76 kg with an Apgar score of 9. A systolic heart murmur detected at birth subsequently disappeared. Also noted immediately was an asymmetrically enlarged tongue, which was part of a generalized hypertrophy of the right side of the body. Owing to the association of congenital hypertrophy with Wilms' tumor, the child received biannual abdominal ultrasound examinations.

Neurological review showed hypotonia and delayed psychomotor development, so at age 19 months the child was hospitalized for evaluation. At this hospitalization she was an alert little girl with head circumference of 44 cm, anterior fontanelle open, and measuring 3x2 cm. She had remnants of a hemangioma on the forehead. The eyes had pronounced epicanthal folds and the pupils were equal and responded well to light. The nose was small and slightly bossed. The palate was high and the lower lip protuberant. The tongue was large, protruded from the mouth, and the right side was significantly larger than the left, which was part of the diffuse unilateral muscular hypertrophy. Measurements were made of the lower limbs, and the

Fig 1. Karyotype illustrates ring formation of chromosome 18 (arrow).
muscles of the right thigh and calf were found to have a greater circumference than those of the left. The right leg was also longer. OM was not yet sitting unsupported and was only verbalizing monosyllabically.

At this time genetic examination revealed ring formation of chromosome 18 (Fig 1). The family history revealed that the parents were healthy and neither was of advanced age. An older daughter also suffers from mild hypotonia. A son born since, after amniocentesis, is healthy and of above-average intelligence.

Dental findings and treatment

OM presented with nursing bottle caries. All the maxillary teeth were affected by caries. The mandibular teeth were unaffected. Unusually large spacing was detected between the mandibular right primary canine and first primary molar. The presence of all the developing tooth germs was confirmed except for that of the mandibular right second premolar. Radiographs (Fig 2) confirmed the clinical findings. Asymmetric macroglossia was noted with the tongue always protruding from the mouth (Fig 3).

Treatment was performed with conscious sedation using hydroxyzine and nitrous oxide/oxygen and a Papoose Board® (Olympic Medical Group, Seattle, WA). Fissure sealants were placed on all mandibular and maxillary second primary molars, stainless steel crowns on the maxillary first primary molars and polycarbonate crowns on the maxillary primary canines and central incisors. The maxillary primary lateral incisors were extracted. At 6-1/2 years old, OM underwent tongue reduction. At that time, her physical development was appropriate for her age; she had marked mandibular prognathism, which was later discovered to be postural owing to the protrusive tongue. There was a concomitant anterior open bite. Other dental findings were normal. The partial glossectomy was performed under general anesthesia. The child was treated with intravenous antibiotics and discharged in good condition after a week. Six months postglossectomy the tongue no longer protruded from the mouth (Fig 4).

At age 7 years 7 months, the child was seen for routine dental treatment, which was performed as be-
Fig 5 and 6. Extraoral and intraoral views one year postglossectomy show the persistence of the anterior open bite although the tongue is well inside the mouth.

Fig 7. Intraoral lateral view one year postglossectomy showing the persistent spacing between the mandibular right first primary molar and canine.

Table. The frequencies of various features in reported cases of 18p- and 18q- syndromes compared with those found in OM

<table>
<thead>
<tr>
<th>Feature</th>
<th>OM</th>
<th>18p (%)</th>
<th>18q (%)</th>
<th>18r (%)</th>
</tr>
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<tbody>
<tr>
<td>Pronounced epicanthal folds</td>
<td>√</td>
<td>26</td>
<td></td>
<td>50</td>
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<tr>
<td>Bossed nose</td>
<td>√</td>
<td>34.8</td>
<td></td>
<td></td>
</tr>
<tr>
<td>High-arched palate</td>
<td>√</td>
<td></td>
<td>20</td>
<td></td>
</tr>
<tr>
<td>Macroglossia</td>
<td>√</td>
<td>34.8</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Dental decay</td>
<td>√</td>
<td></td>
<td>50</td>
<td>40</td>
</tr>
<tr>
<td>Carp-shaped mouth</td>
<td>√</td>
<td>52.2</td>
<td>20.8</td>
<td>50</td>
</tr>
<tr>
<td>Micrognathia</td>
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<td></td>
<td></td>
<td>40</td>
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<tr>
<td>Macroglossia</td>
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<tr>
<td>Midface dysplasia</td>
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<td>58.3</td>
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</tr>
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<td>4.3</td>
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<td>100</td>
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<td>Mental deficit</td>
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Discourse

Among the findings cited by de Grouchy in the 18p- syndrome are dental caries and micrognathia. It would seem unlikely that dental caries per se is an integral part of any of these syndromes although the phenomenon described may have been dental hypoplasia. In the case presented, the caries was due to nursing bottle use. Strikingly, all of the mandibular teeth were protected by the inordinately large tongue, and thus were unaffected. The asymmetric, large tongue was probably the cause of the unilateral large space between the mandibular right canine and primary first molar. Micrognathia was not a feature here — after the glossectomy the teeth occluded. Macroglossia does not seem to have been a feature in previous reports of the 18p, 18q-, or 18r-syndromes. In the present case, macroglossia was the most prominent feature, and it was the first symptom noticed at birth, that led to assessing the child and subsequently diagnosing the disorder.

The primary idiopathic hypertrophy of the tongue diagnosed here was part of a unilateral hypertrophy of the right side as classified by Shafer. The decision as to whether or not to perform a partial glossectomy should be based on assessing both the level of functional disturbance as well as psychologic considerations. Severe macroglossia can interfere with normal speech, can result in episodic respiratory difficulties and dysphagia, and can cause drooling. Also, the tongue and oral mucosa may dry out causing ulceration. Anterior and lateral open-bite malocclusions are frequent, while mandibular prognathism and Class III malocclusions also occur. If the child can approximate his teeth with the tongue inside the mouth and not bite it, there is no clear indication for surgical correction.

Psychologic considerations have two aspects. First is the trauma caused to a patient who is sufficiently aware of his appearance to be sensitive to a protruding tongue. Second, several investigations have shown that a person's appearance greatly affects his acceptance by society. Teachers tended
to have higher expectations of the more attractive students than of their less attractive peers. It has been found that infants spent a great deal more time gazing at pictures of women who had previously been graded as attractive than at pictures of women who had been classified unattractive. In one investigation, a group of Down syndrome patients underwent plastic surgery in order to correct the characteristic stigmata of the syndrome (including macroglossia). Parents and teachers were then asked to rate the improvement both in esthetics and function, and the partial glossectomy was considered the most successful in improving appearance while function (eating, drinking, and speech) was also considered vastly improved.

In this child, esthetics and speech difficulties particularly disturbed the parents. The child was unable to close her mouth without biting down on the tongue. After the partial glossectomy, there was a great improvement in the child’s speech and the family was very pleased with the esthetic result.

FUTURE ANNUAL SESSIONS

1994 May 26–June 31
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