Cephalometric analysis of Rapp-Hodgkin syndrome

Thomas C Hart, Stephanos Kyrkanides

Abstract
Rapp-Hodgkin syndrome (RHS) is a rare form of ectodermal dysplasia with variable involvement of the hair, eyes, sweat glands, nails, and teeth. Oral findings may include hypodontia, hypoplastic enamel, cleft lip/palate, and a pronounced midfacial hypoplasia. The objective of this study was to determine if the pronounced midfacial hypoplasia is the result of a true tissue hypoplasia or displacement of midfacial tissues. We have identified a family in which three members, a mother and two daughters, display clinical features characteristic of RHS. Clinical and intraoral examination of these affected persons showed manifestations characteristic of RHS, and several new findings were observed, including subcutaneous abscesses, palmoplantar keratosis, and keratotic lesions located on the chest and trunk. To assess the midfacial hypoplasia, lateral cephalometric analysis was performed on lateral headplates. Results of the cephalometric analysis suggest that the midfacial hypoplasia results from both a deficiency and a displacement of the maxilla. These findings are significant because successful dental treatment of RHS relies upon accurate assessment of current and projected orofacial development, particularly for the skeletal relations of the maxilla and mandible.

Table 1 Clinical features associated with Rapp-Hodgkin syndrome

<table>
<thead>
<tr>
<th>Feature</th>
<th>Description</th>
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<tbody>
<tr>
<td>Growth failure</td>
<td>Hypohidrosis or normohidrosis</td>
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<tr>
<td>Hypodontia</td>
<td>Sparse, thin, and dry hair, pili torti (&quot;kinky hair&quot;)</td>
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<tr>
<td>Sharp nose</td>
<td>Small nasal bridge</td>
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<tr>
<td>Cleft lip and palate</td>
<td>Hypodontia with small and conical teeth</td>
</tr>
<tr>
<td>Nail dysplasia</td>
<td>Hypoplasia</td>
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<tr>
<td>Absent lacrimal punctae</td>
<td>Photophobia</td>
</tr>
<tr>
<td>Hypospadias</td>
<td>Hypoplastic dermatoglyphics</td>
</tr>
<tr>
<td>Midfacial hypoplasia</td>
<td></td>
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Material and methods
A family with three persons affected by RHS was studied (fig 1). Each person was examined for general medical and dental findings. Lateral cephalometric x rays were taken on the three affected subjects using a GE XR machine (16 mA, 90 Kv, 21/48 seconds) with patients positioned in a Welmer cephalostat (distance characteristics: a = 5", b = adjustable). Radiographs were traced and cephalometric tracings assessed as previously described.8-10 The following linear and angular measurements were determined. Linear measurements (in mm): A-PNS, ANS-PNS, Ba-PNS, Ba-A, N-S, S-A, S-PNS, S-Ba, SE-PNS, UFH. Angular measurements (in degrees): angle of convexity, Lande's angle, facial angle, ANB angle, saddle angle. Where A = A point, N = nasion, S = sella, Pr = porion, Pg = pogonion, O = orbitale, Ba = basion, SE = sphenoidai point, ANS = anterior nasal spine, PNS = posterior nasal spine, UFH = upper facial height, angle of convexity = N-A-Pg, Lande's angle = (O-Pr)-(N-A), facial angle = (O-Pr)-(N-Pg), saddle angle = N-S-Ba.
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II-2 | III-1 | III-2
---|---|---
Age | 38 | 13 | 10
Weight (kg) (centile)* | 63 (77) | 63 (93) | 36 (75)
Height (cm) (centile)* | 157 (24) | 159 (70) | 140 (60)
Submucous cleft palate | + | - | +
Heat intolerance | + | + | +
Nasolacrimal duct atresia | + | + | +
Hearing loss | + | + | +
Midfacial hypoplasia | + | + | +
Nail dysplasia | + | + | +
Mild keratocoucus | + | + | +
Subdermal abscess (arm/ | - | - | -
genital)
Keratic lesions (chest/trunk) | + | + | +
Hypodontia | + | + | +

* Body weight and height centile according to Hall.15
† Teeth subsequently extracted for full dentures.

Linear and angular cephalometric measurements are shown in table 3. These measurements are compared to sex, race, and age matched controls.16 To illustrate the cephalometric analysis, the following data for III-1 are presented: lateral headplate for III-1 (fig 2) and the resulting cephalometric tracing (fig 3). Fig 4 is an introra oralograph of III-1 showing dental anomalies of tooth morphology and number, as well as an anterior cross bite, typical of a class III angle relationship, where the lower teeth occlude in front of the upper teeth.

Cephalometric analysis showed a mild to moderate maxillary tissue hypoplasia in all affected members of the family (ANS-PNS, A-PNS). The maxilla was also displaced closer to the cranial base (S-PNS, SE-PNS, and S-A) in all affected persons; however, it was positioned within normal limits anterior-posteriorly relative to the forehead (Lande's angle). The presence of a prognathic mandible also contributed to a concave skeletal profile (angle of convexity).

**Table 2 Clinical findings of affected subjects**

**Table 3 Cephalometric measurements**

**Figure 2 Lateral cephalometric headplate of III-1.**

**Results**

The clinical findings of the three affected subjects are presented in table 2. Two new clinical findings were observed. II-2 had a history of subcutaneous abscesses in the armpit and genital areas. II-2 and III-1 had keratotic lesions on their chest and trunk. The overall clinical findings of these persons support the variable clinical expression of the syndrome previously reported.

**Discussion**

Pronounced midfacial hypoplasia is a noted characteristic of Rapp-Hodgkin syndrome. To date, no studies have determined whether this hypoplasia is the result of a lack of maxillary bony tissue (tissue deficiency) or a displacement of the bony tissue present. Maxillary tissue deficiency may be determined from cephalometric analysis by measuring the length of the palatal plane (ANS-PNS, and A-PNS) (table 3). Displacement of the bony maxilla may be assessed by its position relative to (1) the cranial base (S-A, S-PNS) and (2) the forehead (point N, Lande's angle) (table 3).

Results of this study suggest that both tissue hypoplasia and tissue displacement contribute to the midfacial hypoplasia observed in RHS. Cephalometric analysis showed a mild to moderate maxillary tissue hypoplasia in all three affected members of this family. The degree of
could provide important confirmation of this observation if the maxillary breadth is also significantly reduced. An additional finding contributing to the concave profile of these patients was the presence of mandibular prognathism. Measurement of the facial angle showed the mandible to be prognathic relative to the forehead (N) in all three affected persons. Mandibular prognathism would tend to exaggerate the concave nature of the skeletal profile.

These findings have important treatment implications. Persons with Rapp-Hodgkin syndrome are frequently missing teeth. This may affect appearance, speech, and mastication. This is particularly important to children and adolescents, who are often concerned with the appearance of missing teeth. Dental implants can successfully replace missing teeth, permitting restoration of an aesthetic appearance and helping with speech and chewing ability. Dental implants offer several advantages over traditional dentures. Implants may be strategically placed to conform to the individual needs of the patient and they remain in place. Dental implants are likely to be an increasingly significant part of dental rehabilitation of persons with Rapp-Hodgkin syndrome. Understanding the relationship between the maxilla and mandible is crucial to successful planning and implementation of dental implant therapy. Therefore, the effects of midfacial hypoplasia and mandibular prognathism on dental occlusion must be considered in dental treatment. This class III angle relationship will significantly affect occlusal relationships in these patients. Such a relationship complicates dental implant therapy. If other studies confirm the generality of these findings, and determine that midface hypoplasia common to RHS patients results from the maxillary deficiency and displacement, other treatment strategies such as maxillary expansion therapy may be desired before dental implant therapy.

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