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.

(J Med Genet 1994;31:758-760)

Rapp-Hodgkin syndrome was first described by Rapp and Hodgkin in 1968.¹ They reported a family (mother, son, and daughter) affected by a pleiotropic form of hypohidrotic ectodermal dysplasia with variable involvement of teeth, hair, nails, oral clefting, heat intolerance, and a characteristic facies that included pronounced midfacial hypoplasia. This report was the first to describe an autosomal dominant form of hypohidrotic ectodermal dysplasia associated with cleft lip and palate. This condition is generally distinguished from other forms of ectodermal dysplasia by its mode of transmission and the presence of clefts of the lip and palate, narrow dystrophic nails, hypospadias in males, and variable short stature. Since this initial report a number of additional clinical findings have been reported by several investigators and the condition has come to be known as Rapp-Hodgkin syndrome (RHS).²⁻⁶ A summary of these clinical findings are listed in table 1. It has been proposed that the pro-

nounced midfacial hypoplasia observed in RHS may result from a hypoplastic maxilla,⁷ or be the result of maxillary retrusion.⁶ However, it is not known if this is because of a true maxillary tissue hypoplasia or displacement of a normal maxilla. Cephalometrics is the scientific measurement of the bones of the cranium and face, using a fixed reproducible position for lateral radiographic exposure of the skull and facial bones. This method permits study of the measurements of the head with relation to specific reference points, and may be used to evaluate facial growth and development. We have identified a family with three persons who displayed the characteristics associated with RHS. The purpose of our study was to use skeletal cephalometric analysis to characterise the nature of the midfacial hypoplasia in Rapp-Hodgkin syndrome.

Table 1 Clinical features associated with Rapp-Hodgkin syndrome

Growth failure
Hypohidrosis or normohidrosis
Sparse, thin, and dry hair, pili torti ("kinky hair")
Sharp nose, small nasal bridge
Small mouth
Cleft lip and palate
Hypodontia with small and conical teeth
Nail dysplasia
Hypospadias
Absent lacrimal punctae
Photophobia
Hypoplastic dermatoglyphics
Midfacial hypoplasia

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 = sphenoethmoidal 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.

Department of
Dentistry,
Bowman-Gray School
of Medicine,
Medical Center Blvd,
Winston-Salem,
NC 27157, USA
T C Hart

Department of
Orthodontics,
Eastman Dental
Center, Rochester,
NY, USA
S Kyrkanides

Correspondence to Dr Hart.
Received 25 October 1993
Revised version accepted for
publication 26 April 1994

Figure 1 Pedigree of affected kindred.

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.

Table 2 Clinical findings of affected subjects

Clinical findings	II·2	III·1	III·2
Age	38	13	10
Weight (kg) (centile)*	63 (77)	63 (93)	36 (75)
Height (cm) (centile)*	157 (25)	159 (70)	140 (60)
Submucous cleft palate	—	—	+
Heat intolerance	+	+	+
Nasolacrimal duct atresia	+	+	+
Hearing loss	+	+	+
Midfacial hypoplasia	+	+	+
Nail dysplasia	+	+	+
Mild keratoconus	+	+	+
Subdermal abscess (arm/genital)	+	—	—
Keratotic lesions (chest/trunk)	+	+	—
Hypodontia	+†	+	+

* Body weight and height centile according to Hall.¹¹

† 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.¹⁰ 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 intraoral photograph 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 3 Cephalometric measurements

	II·2	III·2	III·1
Linear measurements (mm) (Z score)			
A-PNS	43·5 (−1·38)	42 (−2·51)	47 (−1·24)
Ba-A	97 (0·76)	91 (−1·38)	101 (0·43)
S-Ba	44 (−0·68)	43 (−0·74)	48·5 (0·68)
S-PNS	43 (−1·01)	42 (−3·0)	44 (−1·9)
S-A	82 (−0·21)	78·5 (−2·34)	80 (−2·07)
N-S	76·5 (1·45)	71·5 (1·38)	69·5 (−1·93)
ANS-PNS	49 (−1·8)	45 (−3·4)	47 (−3·3)
SE-PNS	38 (−2·29)	41 (−2·6)	38·5 (−4·0)
UFH	49 (1·0)	49 (2·11)	40·5 (0·85)
Angular measurements (degrees) (Z score)			
Angle of convexity	−5 (−5·0)	−13 (−9·6)	−23 (−18·3)
Lande's angle	85 (−0·97)	88·5 (0·16)	93 (2·0)
Facial angle	86·5 (0·96)	95 (8·3)	95 (4·6)
ANB angle	−2·0 (−2·21)	−3·0 (−2·77)	−1·0 (−1·67)
Saddle angle (N-S-Ba)	137 (1·45)	134·5 (0·82)	132 (0·42)

Figure 2 Lateral cephalometric headplate of III·1.

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

Figure 3 Cephalometric tracing of lateral headplate seen in fig 2.

Figure 4 Intraoral photograph of III-1 showing dental anomalies.

this deficiency ranged from mild (II-2) to severe (III-1) (table 3). We found that not only was there a deficiency in the length of the palatal plane, but additionally the maxilla itself was displaced dorsally closer to the posterior cranial base and cephalad, closer to the anterior cranial base. Although the maxilla was shorter and vertically displaced, it was positioned within normal limits anterior-posteriorly relative to the forehead (N). These findings indicate an underdeveloped nasomaxillary complex and suggest that the midfacial hypoplasia is the result of both deficiency and displacement of maxillary tissue. Posterior-anterior radiographs

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.

We would like to thank Dr J D Subtelny for his expert advice in the cephalometric analysis, Dr M Aslani for help in patient management, Pat Ragusa for photographic illustrations, and Kimberly Ladley for manuscript preparation.

- 1 Rapp RS, Hodgkin WE. Anhidrotic ectodermal dysplasia: autosomal dominant inheritance with cleft palate and lip anomalies. *J Med Genet* 1968;5:269-72.
- 2 Watson RM, Harwick CE. Hypodontia associated with cleft palate. *Br Dent J* 1971;130:77-80.
- 3 Witkop CJ Jr, Brearley LJ, Centry WC Jr. Hypoplastic enamel, onycholysis and hypohidrosis inherited as an autosomal dominant trait. *Oral Surg* 1975;39:71-86.
- 4 Silengo MN, Davi GF, Bianco R, Demarco A, Vernoa R, Franceschini P. Distinctive hair changes (pili torti) in Rapp-Hodgkin ectodermal dysplasia syndrome. *Clin Genet* 1982;21:297-300.
- 5 Salinas CF, Marcos-Montes GGM. Rapp-Hodgkin syndrome. Observations on ten cases and characteristic hair changes (pili canaliculi). *Birth Defects* 1988;24:149-68.
- 6 Crawford PJJ, Aldred MJ, Clarke A, Tso MSY. Rapp-Hodgkin syndrome: an ectodermal dysplasia involving the teeth, hair, nails, and palate. *Oral Surg* 1989;67:50-62.
- 7 Broadbent BH. A new X-ray technique and its application to orthodontia. *Angle Orthodont* 1931;1:45-66.
- 8 Breslau-Siderius EJ, Lavrijns APM, Otten FWA, van der Schroeff JG, Swart JGN. The Rapp-Hodgkin syndrome. *Am J Med Genet* 1991;38:107-10.
- 9 Downs WB. Analysis of the dentofacial profile. *Angle Orthodont* 1956;26:191-219.
- 10 Riolo MC, Moyers RE, McNamara JA Jr, Hunter WS. *An atlas of craniofacial growth*. Monograph 2. Craniofacial Growth Series. Ann Arbor: University of Michigan, Center for Human Growth and Development, 1974.
- 11 Hall JH. *Handbook of normal physical measurements*. Oxford: Oxford University Press, 1989.