Rapp-Hodgkin Syndrome: Clinical and Dental Findings

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Rapp-Hodgkin syndrome is a rare form of ectodermal dysplasia involving the hair, eyes, sweat glands, nails, teeth and palate. This syndrome is characterized by stiff, sparse hair with the appearance of steel wool, sparse eyebrows and lashes, cleft palate, absence of lacrimal punctae, epiphora, a decreased number of sweat glands, and dystrophic nails. Cleft palate, hypodontia, abnormal tooth shape, multiple caries, delayed eruption of teeth are the main oral manifestations. In this paper we describe the clinical and dental findings of this syndrome in a 7 year old girl referred to the dental clinic because of severe tooth ache. The dental treatment included root canal treatment, composite restorations, extractions and removable prostheses. This paper highlights features of Rapp-Hodgkin Syndrome and its dental rehabilitation.

Keywords: *Rapp-Hodgkin syndrome, hypodontia, cleft palate*
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INTRODUCTION

Ectodermal dysplasias (EDs) represent a large and complex group of diseases comprising more than 170 different clinical conditions.[1-3] They are caused by impaired development of epidermal appendages and are characterized by a primary defect in at least two of the following tissues: nails, hair, teeth and sweat glands.[3,4]

Rapp-Hodgkin ectodermal dysplasia or Rapp-Hodgkin Syndrome (RHS) is a rare autosomal dominant disorder [5] but sporadic mutations do occur.[6] This syndrome was first described over 30 years ago in an affected mother, son and daughter with a combination of anhidrotic ectodermal dysplasia, cleft lip and cleft palate.[5] RHS does display some clinical overlap with other ectodermal dysplasias, notably ectrodactyly-ectodermal displasia-cleft lip-palate syndrome (EEC), and ankyloblepharon-ectodermal dysplasia-clefting(AEC).[7] Recent genetic molecular studies have shown that AEC, EEC and RHS represent a wide phenotypic spectrum of a single genetic disorder associated with mutations in the *TP63* gene.[7,8,9] The characteristic features of this syndrome, summarized by Witkop et al. in 1975[10] are stiff, sparse hair with the appearance of steel wool, sparse eyebrows and lashes, cleft palate, the absence of uvula and lacrimal punctae, hypodontia, hypoplastic enamel, epiphora, ectropion, photophobia, hypoplastic dermatoglyphics and a decreased number of sweat glands.[11–13] The oral manifestations of RHS include small teeth with enamel defects, missing teeth, multiple caries, delayed eruption of teeth, tooth shape anomaly, and reduced production of saliva.[4, 6, 9, 12]

The aim of this case report is to present the general features of RHS and the dental rehabilitation of its symptoms.

CASE REPORT

A 7-year-old female patient was referred to our clinic with toothache. After extraoral and intraoral examinations, the patient was referred to the Pediatric Endocrinology Department with a prediagnosis of ectodermal dysplasia. After consultation Rapp Hodgkin Syndrome was diagnosed. Born full-term after an uneventful pregnancy, weighing 3000g and measuring 50cm, she was the oldest of three children and the only affected sibling. Her consanguineous parents were systemically healthy and there was no family history of genetic disease. The pedigree of the patient is shown in Figure 1. At birth she had clefting of the soft palate for

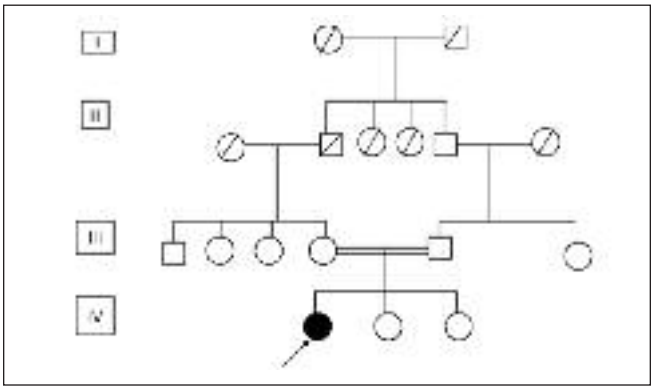

Figure 1: Pedigree of patient's family. Symbols marked by a slash indicate that the subject is deceased. Double line is representative of consanguineous union. Squares indicate males, and circles females. Proband is indicated by arrow.

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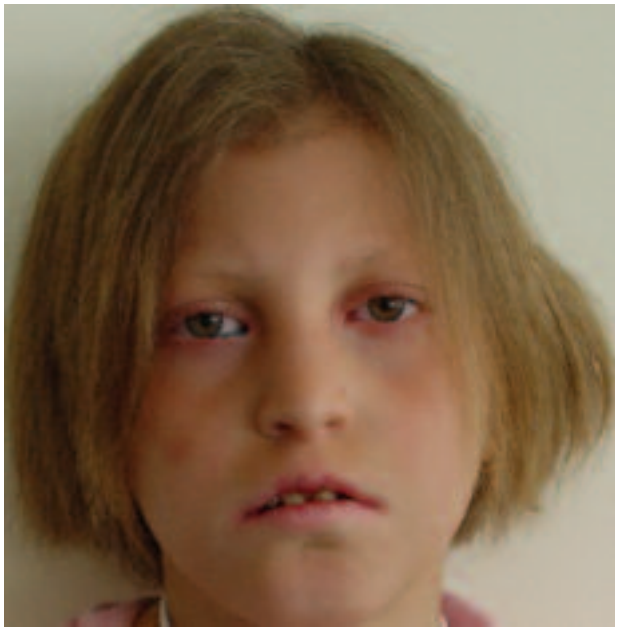

Figure 2: The patient's facial appearance; light-colored hair, sparse eyelashes and eyebrows, thin lip and flat nasal bridge with hypoplastic alae nasi,

which she underwent surgery when she was 17 months old.

On physical examination, she presented as a verbal and active young girl, 120 cm tall, weighing 24 kg. Her hair was

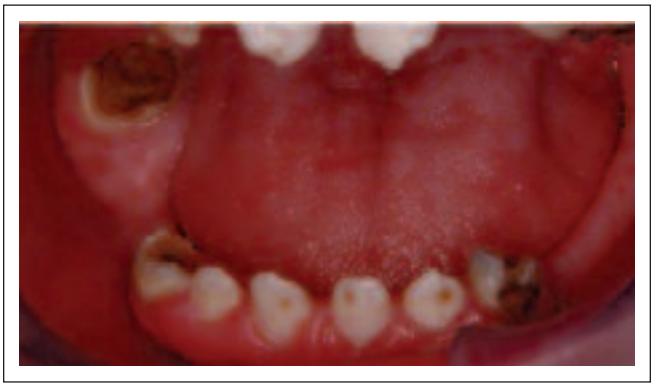

Figure 4: Intraoral view- mandible with small and conical incisors.

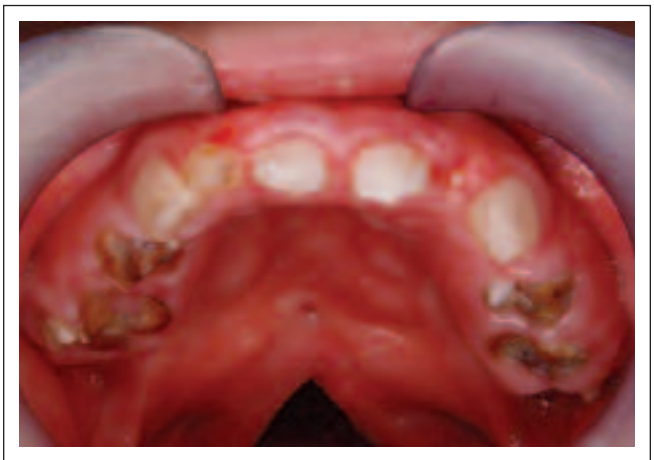

Figure 5: Intraoral view- maxilla;operated cleft soft palate and carious posterior teeth.

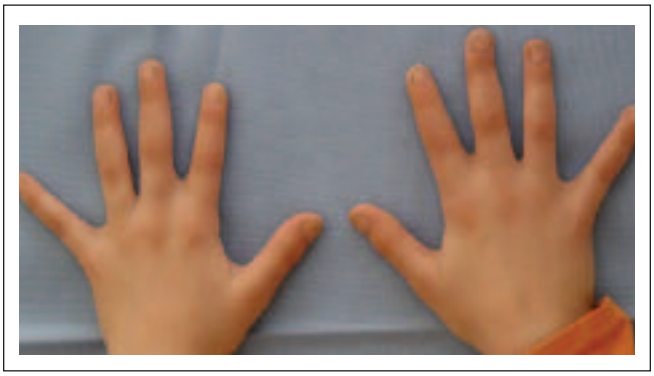

Figure 3: Incomplete syndactyly between second and third fingers and dystrophic nails.

light colored, slow growing, very fragile and broke easily when combed. The eyelashes and eyebrows were sparse. She had thin lips and a narrow nose with hypoplastic nasi alae (Figure 2). Her fingernails and toenails were dysplastic, small and displayed soft tissue tufting (Figure 3). There was incomplete bilateral syndactyly between the second and third fingers (Figure 3). She had obstructed lacrimal ducts leading to recurrent conjunctivitis. She underwent surgery for this problem. Before she came to the pediatric dentistry clinic, her parents were not aware that she had vision problems and after an examination in the ophthalmology department, she started to wear eye glasses.

Renal ultrasound and a cardiac echocardiogram showed no abnormalities. Her blood count (including calcium, phosphorus, magnesium, zinc levels), TSH, FSH, LH and immunoglobulin measurements were normal. Chromosomal analysis revealed a normal female karyotype 46, XX.

An intraoral examination revealed mixed dentition with right and left maxillary deciduous first molars, right maxillary second incisors, right and left maxillary–mandibular canines. Extensive caries in the permanent first molars was noted. The crowns of the mandibular and maxillary incisors were small, rather conical and dysmorphic (Figures 4-6). The clefting of the soft palate, which had been operated on, was observed during the intraoral examination. Agenesis of

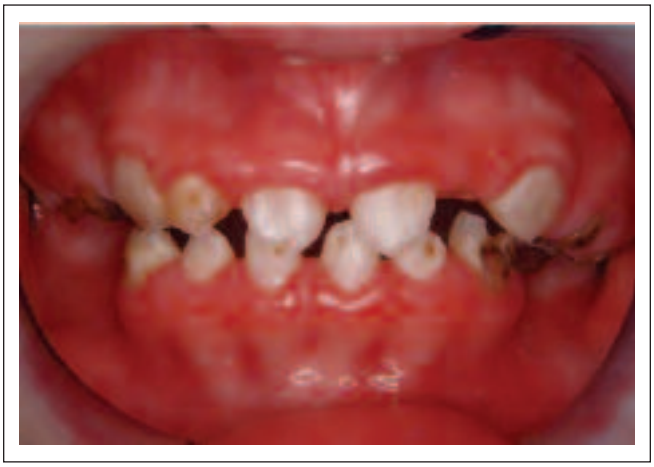

Figure 6: Occlusion.

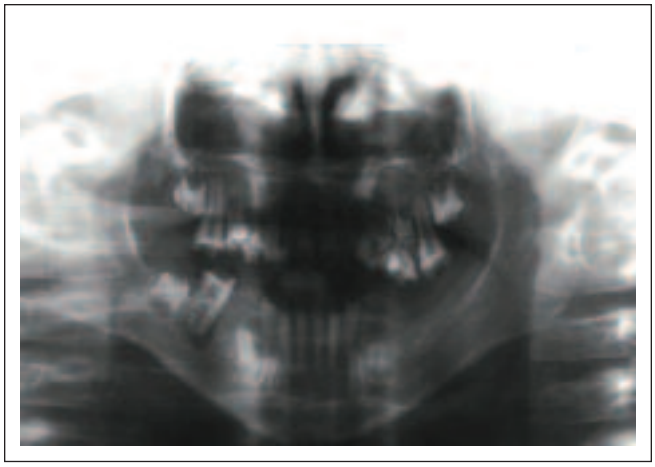

Figure 7. Panoramic radiograph revealed hypodontia, taurodontism in permanent right mandibular first molar and reduced enamel layer on the crown of the all teeth.

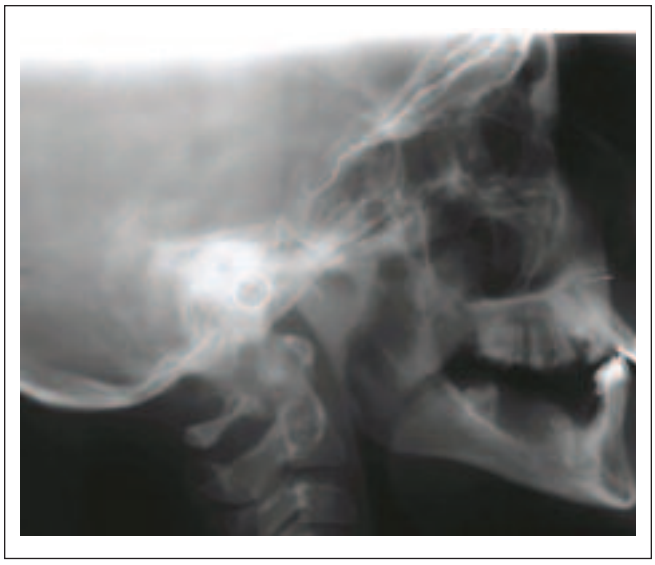

Figure 8: Cephalometric radiograph revealed Class III tendency due to mandibular prognathism.

the permanent left and right mandibular second premolars as well as taurodontism in the permanent right mandibular first molar were observed in the panoramic radiograph which also showed a reduced enamel layer on the crowns of all the teeth (Figure 7). The mandibular left first molar had been extracted the previsions year due to severe caries.

Lateral cephalometric analysis showed the effective mid-facial length was lower than normal values according to McNamara analysis. There was a tendency to Class III angle relationship due to mandibular prognathism (Figure 8). Treatment in the present case involved the extraction of the mandibular and maxillary posterior teeth because of gross caries. The maxillary central right and left incisors and mandibular central and lateral incisors were restored using composite celluloid strip crowns. The left and right mandibular canines needed root canal treatment after which a root canal sealer was applied and the teeth restored with a composite restorative material. On completion of restorative

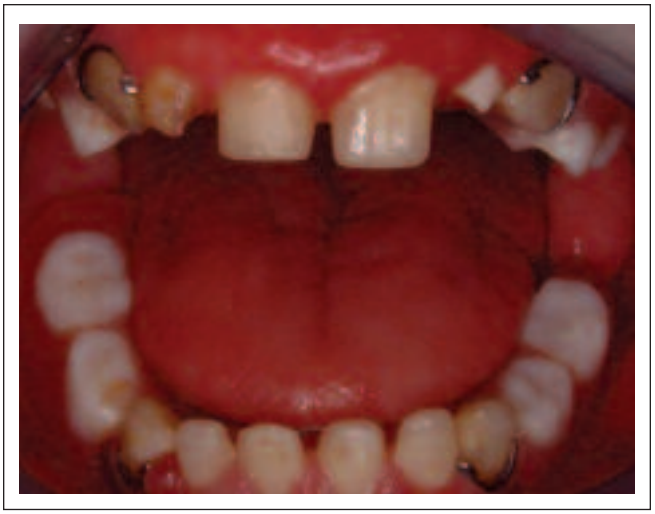

Figure 9: Final restorative results with removable prosthesis

treatment, impressions of the maxilla and mandible were taken for a diagnostic model. Custom trays were fabricated and a final impression made. When the removable prosthetic denture was completed, it was delivered with wear and care instructions for the patient and her parents (Figure 9). The follow up time is 1 year.

DISCUSSION

Our patient was compared with previous cases (Table 1). The overall clinical findings for this patient support the variable clinical expression of the syndrome previously reported.

RHS is considered one of the groups of disorders categorized by findings of ectodermal dysplasia and clefting. Of the four areas particularly affected- the hair, nails, eyes and palate-the two most distinctive features are pili torti and cleft palate.[1,10,14] Pili torti is a rare hair abnormality that is associated with many syndromes. It is caused by irregular thickening of the outer root sheath of the hair, with the flattening and rotation of hair shafts resulting in fragile corkscrew hairs.[15,16] There are seven forms of ectodermal dysplasia associated with pili torti; of these only RHS syndrome also shows cleft palate.[1] Therefore, cleft palate is the cardinal feature of this syndrome.[1, 6, 9, 11]

Almost all reported cases of RHS describe the hair as sparse, coarse, dry, strawlike, wiry, thin, short and hypopigmented. These features are present at birth or may appear during infancy or throughout childhood. Hair loss at puberty is also a reported characteristic.[17] In our patient this was not observed because she was seven years old but her hair was dry, fragile, sparse and blondish white.

Individuals with Rapp-Hodgkin Syndrome generally have striking faces that include a high forehead, hypoplasia maxilla, a low nasal bridge, syndactyly and a thin upper lip[18] We observed similar findings to those in the literature with respect to the high forehead, thin lips and low nasal bridge, dry skin, and dystrophy of the nails. Incomplete bilateral syndactyly between the second and third fingers was also

Table 1 : Comparison of present case with previous cases

	Our patient	Kim and Shin[5]	Plotkova-puech *et al*[14]	Neilson *et al* [17]: case 1	Neilson *et al* [17]: case 2	Atasu *et al* [20]: case 1	Atasu *et al*[19]: case 2	Atasu *et al* [19]: case
Cleft palate/ Cleft lip	+ / -	+	Ogival palate	+ / -	+ / -	+/ +	-/ -	- / -
Short nose/ broad root	+ / +	/+	+/ -	Thin and hypoplastic nasal alae	Hypoplastic nasal alae	+ /+	+ / +	+ / +
Maxillary hypoplasia/ small mouth	-/ +	+	+	+	- / -	+ / -	+ / +	- / -
Hypoplastic enamel/ severe caries/ hypodontia	+ / + / +	?/ ? / +	+ / + /+	+ /+ / -	+ /+ / -	anodontia	+ / + / +	Anodontia (two im teeth within both a
Nail dysplasia	+	+	+	+	+	+	-	+
Nasolacrimal duct stenosis /absent lacrimal puncta	+ / -	- / -	- / -	-/+	-/ +		- / -	- / -
Sparse, thin / dry hair	+ / +	- / +	- / +	- / +	-/ +	- /+	- / +	- / +
Facial dysmorphism	-	+	+	-	+	+	+	+
Diffuse erythroderma	-	-	+	-	-	-	-	-
Sweat problem	-	+	+	+	+	-	-	-

observed in our patient.[18]

Several oral and dental manifestations of RHS such as cleft palate, small teeth with enamel defects and missing teeth, severe caries, late eruption of teeth and reduced production of saliva[4,10,11,12] have been reported. Kantaputra *et al*[18] observed multiple tooth caries, glossy tongue and large dental pulp space. Atasu *et al*[19] described three siblings with RHS, all with, cleft lip and palate and anadontia. Similar findings to those in the literature with respect to tooth shape anomalies and multiple tooth caries were seen in our patient in addition to hypodontia and taurodontism.

Lateral cephalometric analysis performed on RHS patients by Hart and Kyrkanides,[20] showed midfacial hypoplasia to be a feature of RHS.[20] Our patient showed mild midfacial and mandibular deficiencies consistent with Hart and Kyrkanides' findings.[20] These findings are significant because successful dental treatment of RHS relies upon accurate assessment of current and projected orafacial development, particularly for the skeletal relationship between the maxilla and mandible.

Hypodontia and malformation are the most frequently observed characteristics of RHS. Children suffering from hypodontia and multiple caries may have severe functional and esthetic problems often leading, to psychological problems. The adverse aesthetic and functional effects of hypodontia in RHS children can be minimized by early diagnosis and treatment planning. The pediatric dentist should keep in mind that there are good possibilities using removable prostheses to help these patients. However, fixed prosthodontic approaches are inadvisable because of the dynamics of developing dentition and continuing growth of the jaws in children.[21] When growth has stabilized, implants or fixed prosthetic rehabilitation may be an optional aid to support, stabilize and retain the prostheses.[21, 22] Prosthetic rehabilitation not only improves speech and masticatory function, but also has positive psychological implications that may help greatly in regaining self confidence.

Because severe caries is features of RHS oral hygiene instruction is a significant part of dental management and professional care, including frequent dental examinations, oral hygiene prophylaxis and preventive dental applications are very important for these patients.

CONCLUSION

Patients with Rapp Hodgkin Syndrome often have difficulty eating because of missing teeth, abnormal dentition and multiple caries and the physical appearance of these, in addition to cleft palate, may also result in psychological problems.

These problems can be solved with dental treatment, professional care and a multidisciplinary approach. Because the pediatric dentist is the most important member of the multi-

disciplinary team, he/she should be aware of the symptoms of RHS so corrective measures can be taken to solve existing problems.

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