Rothmund–Thomson Syndrome with partial anodontia: A rare case report

INTRODUCTION

Rothmund–Thomson syndrome (RTS), was first illustrated by the German ophthalmologist Auguste Rothmund in 1868. He observed poikiloderma, growth retardation developing at a young age with rapidly progressive bilateral juvenile cataracts in Bavarian children [2]. Later in 1936, Thomson, a British dermatologist described patients with “Poikiloderma congenitale” and growth retardation who exhibited skeletal defects, but no cataracts. [2, 3] The two groups represents the same syndrome was supported by Carlton in 1943 and by Sexton in 1954. [2] The two syndromes were known as Rothmund Thomson Syndrome (RTS) and the eponym was coined by Taylor in 1957 [3].

Clinically it is an uncommon autosomal recessive dermatosis presenting in infancy [5]. It is characterized by broad clinical expressivity. Till now, approximately 400 RTS patients are reported[6] and generally poikiloderma, the cutaneous erythematous rash is the hallmark sign. Within the initial two years of life, Erythema, blisters and swelling are visible at the sun-exposed areas, mainly on the face, dorsal aspect of hands and extensor aspects of arms and forearms. It then evolves into a permanent rash characterized by skin atrophy with hyper- and hypo-pigmented areas and telangiectasia (collectively known as poikiloderma). Other common features manifested in early childhood are, stunted growth, juvenile cataracts and skeletal abnormalities seen in around 75% patients. [5, 6, 7, 8] The skeletal abnormalities include frontal bossing, saddle nose, small hands and feet, and long-bone abnormalities, including radial ray defects. Hyperkeratosis, sparse and thin hair, eyelashes and eyebrows, dystrophic nails, hypogonadism, premature aging may be observed in adult age. Patients with RTS are also characterised by an immunological impairment and have an augmented possibility of malignancies such as osteosarcoma and nonmelanotic skin cancer [5, 7]. However, life expectancy may not be affected [12].

Mutation of the RECQL4 gene was found as the root cause of RTS in 1998. The RECQL4 protein is believed to be important in keeping up genomic stability and may assume a part in commencement of replication. Till date, roughly two thirds of RTS patients have mutations in RECQL4. [5] Depending on clinical and molecular investigations; two major forms of this condition are described: type I RTS is portrayed by poikiloderma, juvenile cataracts and ectodermal dysplasia, and are negative for the RECQL4 gene mutation scan and type II RTS is characterized by poikiloderma, inherited bone defects and an increased risk of osteosarcoma and is related to pathogenic mutation of RECQL4 gene. [8]. However, the role of RECQL4 in bone development and homeostasis, remains to be assessed. [8]

Dental abnormalities, first observed by Rothmund are associated with a wide range of malformations. These include microdontia, rudimentary or hypoplastic teeth, numerous unusual crown formations and short roots, predominantly in maxillary dentition. Structural defects of connective tissue on a gingival biopsy are also seen in certain patients. An increased caries incidence has also been observed. In some patients craniofacial abnormalities including frontal bossing and saddle nose may also be present. [2, 5, 9]

The goal of Prosthetic rehabilitation in young RTS patient with complete or partial anodontia is: maintaining alveolar bone integrity, ensuring correct patterns of chewing, swallowing, articulation and restoration of normal facial characteristics.

Transitional dentures prove to be a good treatment option for growing patients in which extraction of teeth may lead to psychological trauma, compromised mastication and speech. [13] Cu sile denture, a newer type of transitional denture, can be one of the treatment option for prosthetic rehabilitation in such patients. As stated by De Van “the perpetual preservation of that which remains and not the meticulous replacement of that which has been lost.” [14, 17]
A Cu-sil denture is basically a denture with holes allowing the remaining natural teeth to emerge into the oral cavity. It also includes a soft elastomeric seal which clasps the neck of natural teeth, cushioning them from hard acrylic denture and reducing the masticatory forces transmitted by the prosthesis to the underlying tissues.\[15, 16\]

**CASE PRESENTATION**

A 10 year-old male patient was referred to a Postgraduate Dental College, Department of Pedodontics and Preventive Dentistry with regard to problem with mastication due to absence of teeth. Patient history revealed that he was diagnosed with Rothmund Thomson syndrome in the Department of dermatology of the same hospital. He was the first child of consanguineous parents. There was no family history associated with his condition. He was born after 40 weeks of gestation with a birth weight of 3,000 g. Talangiectasic lesions with hypo and hyperpigmentation started at the age of 3 months on the face of child and progressed gradually to the extremities.

At present, the patient weighed 30 kg, measured cm in height. The patient had no family history of similar clinical features. His hair was fine, sparse, lustreless, and fair and he had normal nails. He had partial anodontia denoted by the developmental absence of teeth in both primary and secondary dentitions. The mental age of the patient appeared normal.

The clinical body examination revealed, the child exhibiting short stature, hypopigmented and hyperpigmented skin lesions over face, lips, trunk and extremities, sparse eye lashes and eyebrows, dystrophic nails which can be seen in Figure 1. The skeletal deformity with difficulty in walking was also observed.

![Figure 1: Clinical body examination of the patient](image)

The important craniofacial features included flat nasal bridge, straight profile and increased nasiolabial angle as described in Figure 2.

![Figure 2: Extraoral features](image)

Intraorally, on soft tissue examination, the gingiva appeared fragile and bright red in colour with absence of stippling and rounded margins. Gingival bleeding occurred spontaneously in many sites. There was marked gingival recession with respect to all teeth. Ankyloglossia with thick, short frenulum, restricted tongue protrusion, and lifting of the tip of the tongue was seen.
On hard tissue examination, a partial anodontia was seen with presence of only thirteen teeth.

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A deep carious lesion was found with respect to #36 which was tender on percussion. A slight mobility was seen #31, #41 with other teeth showing no increased mobility. An edge to edge relationship of the jaws was seen.

The Radiographic investigations were done and orthopantomogram (OPG) revealed multiple missing teeth with absence of impacted teeth, confirming partial anodontia, thin alveolar ridges, reduced vertical bone height and short roots. An intraoral periapical radiograph #36 revealed irreversible pulpitis with widening of periodontal space.

The treatment plan was formulated keeping in mind the patient's complaints and the underlying systemic condition. Oral prophylaxis was done on the first visit. On subsequent visits, root canal treatment of 36 was done. Conservative prosthetic rehabilitation was planned until the growth is ceased, so a Cu sile denture was fabricated in subsequent visits.

**TECHNIQUE FOR FABRICATION OF CU SILE DENTURE**

In order to improve appearance, mastication, and speech, Cu-sile denture was considered to be the best treatment choice.

The technical stages for the fabrication included conventional denture processing techniques. Preliminary impressions of maxillary and mandibular arch were made with irreversible hydrocolloid material (Alginate-Dentsply) in a metal stock tray. This step was followed by pouring with dental stone and obtaining primary
casts. For the maxillary cast, custom tray with double spacer was fabricated followed by border moulding with green stick impression compound and secondary impression with addition silicone light body impression material. For the mandibular arch, conventional steps were performed. The secondary impressions were then poured with dental stone. Over this, acrylic denture bases were made for the master casts and occlusal rims were fabricated.

After the recording of maxillo-mandibular relationship, both casts were mounted on articulator and artificial teeth arrangement was done which was followed by try in procedure. The patient’s phonetics and esthetics were evaluated. Acrylization of the final maxillary and mandibular denture was done.

After the final insertion, routine hygiene instructions for the dentures were given to both the child and his parents. Further adjustments were made to eliminate interferences at recall appointments. After a few recall appointments, he was fully adapted to use the dentures, and his parents reported that he was able to eat well and in addition, his speech and phonetics were improved and the patient was happy with the dentures.

DISCUSSION

The overall incidence of dental involvement in RTS has been estimated at 27% to 59% of the cases. The diagnosis of RTS is made mainly by characteristic clinical features at presentation, as observed in our patient. Differentiation from other causes of childhood poikiloderma such as Kindler’s syndrome, Cockayne’s syndrome, acrokeratotic poikiloderma, sclerosing poikiloderma, dyskeratosis congenita, acrogeria, and xeroderma pigmentosum is to be done. Rare disorders with prominent telangiectasias, such as Bloom’s Syndrome, Fanconi’s anemia, and ataxia telangiectasia are also to be differentiated from this disorder.

Dental abnormalities associated with RTS were first noticed by Rothmund in his original article. From the dental perspective, till date, a wide variety of malformations of the erupted teeth, ectopic eruption, and failure of 1 or more teeth to erupt have been described in most detailed studies. In the current medical literature, however, aberrations in tooth development of RTS patients are usually mentioned using less definite terms, such as microdontia, hypodontia, or dystrophic teeth, adding little to our understanding of the precise nature of tooth involvement in this disorder. In the following case, generalized short root anomaly with normal crowns of the affected teeth is reported in an adult male patient with RTS.
The prosthodontics rehabilitation of children should ensure restoration of normal function of the developing masticatory system, improving aesthetics, preventing speech defects and thus allowing normal functioning of the child. It helps to improve both the sagittal and vertical skeletal relationship during craniofacial growth and development.

A Complete denture generally leads to alveolar bone loss and loss of proprioception and a conventional RPD often have deleterious effects on the periodontally compromised natural teeth. So, a Cu-sil denture, which involves making holes in a complete denture through which the natural teeth emerge, has been considered to be the best treatment option. It offers the patient, a psychological satisfaction of retaining the natural teeth in their mouth.

Dental implants combined with implant-supported dentures are often recommended as a treatment choice for patients with hypodontia. But early implant placement in a growing child may result in infraocclusion since the implant act as ankylosed teeth, and do not follow the movement of surrounding structures,

CONCLUSION

The dental manifestations of RTS can cause considerable social problems in the affected individuals. In this case report, the prosthetic rehabilitation of a 10 year old boy with Rothmund Thomson syndrome associated with partial anodontia was described. Since oligodontia or complete anodontia leads to atrophy of the alveolar bone, prosthetic treatment is of great value to these patients from functional, psychological, and psychosocial standpoints.