CASE REPORT

Dental management of Rothmund-Thomson syndrome with partial anodontia

Nilesh Vithaldas Rath, ¹ Mayur Shrigopal Bhattad, ² Nilima Thosar, ¹ Sudhindra Baliga ¹¹Department of Pedodontics and Preventive Dentistry, Sharad Pawar Dental College, Wardha, Maharashtra, India²Department of Pedodontics & Preventive Dentistry, HSRSM Dental College and Hospital, Hingoli, Nanded, Maharashtra, India**Correspondence to**
Dr Nilesh Vithaldas Rath,
drnileshrathi@gmail.com

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SUMMARY

Rothmund-Thomson syndrome (RTS) is a rare autosomal recessive trait disease. It is characterised by skin, eye and skeletal abnormalities. Abnormalities associated with teeth include abnormal crown and root formations, rudimentary or hypoplastic teeth, microdontia and multiple missing teeth. In the present case, there were multiple decayed primary teeth and multiple congenitally missing permanent teeth. Mandibular left primary first molar (tooth 74) was pulpally involved and obturated with mineral trioxide aggregate. Follow-up after 2 years revealed successful obturation.

BACKGROUND

Rothmund-Thomson syndrome (RTS) is a rare autosomal recessive trait disease. The disease was named after a German ophthalmologist, Rothmund,¹ who observed poikiloderma, growth retardation and rapidly progressive bilateral juvenile cataracts. Poikiloderma, the diagnostic hallmark of the syndrome, is a chronic 'non-progressive' lesion consisting of telangiectatic lesions, reticulated areas of hypo and hyperpigmentation, and punctate dermal atrophy, evolving from previous acute erythematous rashes.² Thomson,³ a British dermatologist, reported similar cases but without cataracts, and labelled them poikiloderma congenitale. Reliable prevalence and incidence data of RTS are not available and, to date, about 300 patients' records have been found in the medical literature.⁴ RTS is basically characterised by skin, eye and skeletal abnormalities that include progressive poikiloderma of early onset, photosensitivity, alopecia and hyperkeratosis. Other abnormalities encompass juvenile cataract, short stature, abnormal teeth, congenital bone defects, soft tissue contractures, osteogenesis imperfecta, sparse, brittle, thin or absent scalp hair eyelashes and/or eyebrows.⁵ Patients with RTS are also characterised by an immunological impairment and have an augmented possibility of malignancies such as osteosarcoma and non-melanotic skin cancer.^{4, 6} The eponym RTS was coined by Taylor,⁷ in 1957, to describe a group of patients with the aforementioned disorders.

Mineral trioxide aggregate (MTA), an endodontic cement, is composed mainly of tricalcic silicate, tricalcic alluminate and bismuth oxide. It is a radio-opaque, biocompatible, dentinogenic and antibacterial material. It is calcium releasing and forms a strong barrier of calcium salts when in contact with tissue fluids.⁸

In the present case of RTS (OMIM: 268400), MTA was used as an obturating material in the

mandibular left first primary molar with follow-up of 2 years.

CASE PRESENTATION

A 7-year-old boy with Rothmund-Thomson syndrome reported to the department of pedodontics and preventive dentistry with decayed anterior and posterior teeth. Telangiectatic lesions with hypo and hyperpigmentation started at the age of 8 months on the face of child and progressed gradually to the extremities. In his family history, a younger brother also suffers from the same condition (RTS). Dermatological lesions in the younger brother were similar to those seen in the elder sibling, but his periodontal and caries status was comparatively better than that of the elder sibling. Skeletal defects were not visible in X-rays of the patient and his younger brother. On inspection, there was symmetrical hyper pigmentation of the skin of the face (figure 1), hands and legs (figure 2), which on closer examination appeared dry and atrophic with poikilodermatous changes. The patient's scalp hair was sparse and thin. An ophthalmological examination was negative for the development of cataract. The child was shy and withdrawn. His general health was good; he was born from a full term, normal delivery.

Figure 1 Clinical photograph showing hyper pigmentation of the facial skin.

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Figure 2 Clinical photograph showing hyper pigmentation of hands and legs.

Intraoral examination showed a set of mixed dentition ([figure 3](#)) where, in the maxillary arch, the first primary molar on the right (Tooth 55), and primary central incisor (Tooth 61), primary lateral incisor (Tooth 62), primary canine (Tooth 63), primary first molar (Tooth 64) and primary second molar (Tooth 65) on the left, were affected with dental caries ([figure 4](#)). In the mandibular arch, the primary central incisor (Tooth 71), primary canine (Tooth 73), primary first molar (Tooth 74) and primary second molar (Tooth 75) on the left were affected with dental caries. On the right side of the mandibular arch, the primary central incisor (Tooth 81), primary canine (Tooth 83), primary first molar (Tooth 84) and primary second molar (Tooth 85) were affected with caries ([figure 5](#)). The maxillary left first primary molar (Tooth 64; [figure 4](#)) and mandibular right first primary molar (Tooth 84; [figure 5](#)) were grossly decayed and the maxillary left primary central incisor (Tooth 61), maxillary left primary lateral incisor (Tooth 62), and mandibular left and right primary incisors (Teeth 71, 81), had proximal decay with grade I mobility. All the first permanent molars in the maxillary and mandibular arch (Teeth 16, 26, 36, 46) were stained and had carious fissures.

INVESTIGATIONS

Intraoral periapical radiographs were used to visualise the tooth and its surrounding structures. Orthopantomograms (OPGs) can help in the visualisation of the maxilla and mandible together

Figure 3 Intraoral photograph showing a set of mixed dentition.

with their associated structures in a single film, with considerably less radiation exposure. Intraoral periapical radiograph revealed pulp involvement in the mandibular first primary molar (Tooth 74) and OPG X-ray revealed that the maxillary permanent central incisor (Tooth 11), lateral incisor (Tooth 12), first premolar (Tooth 14) and second premolar (Tooth 15) on the right; maxillary permanent lateral incisor (Tooth 22), first premolar (Tooth 24) and second premolar (Tooth 25) on the left; mandibular permanent lateral incisor (Tooth 32), first premolar (Tooth 34) and second premolar (Tooth 35) on the left; and mandibular permanent lateral incisor (Tooth 42), first premolar (Tooth 44) and second premolar (Tooth 45) on the right, were congenitally missing ([figure 6](#)). Skeletal radiographical examination was normal and routine blood laboratory tests showed no abnormal values.

DIFFERENTIAL DIAGNOSIS

The differential diagnosis of RTS are other poikilodermatous conditions such as Clericuzio-type poikiloderma with neutropenia,⁹ Werner syndrome, Kindler syndrome, dyskeratosis congenita and Bloom syndrome.^{10 11} Clericuzio-type poikiloderma with neutropenia (PN (MIM %604173)) is a rare autosomal-recessive genodermatosis characterised by poikiloderma, pachyonychia and chronic neutropenia, and considered to be a rare cancer predisposing genodermatosis; partial clinical overlap has caused misdiagnosis of this condition with RTS until identification of the causative gene.⁹ In Werner syndrome, the poikilodermatous changes start, approximately, at the time of puberty,

Figure 4 Intraoral photograph showing right and left maxillary primary teeth affected with caries.

Figure 5 Intraoral photograph showing right and left mandibular primary teeth affected with caries.

with early greying and loss of hair.¹⁰ In Kindler syndrome, patients develop sacral bullae at birth and if it gets traumatised, they then develop diffuse poikiloderma and widespread dermatitis with no increased risk for malignancy.¹¹ In dyskeratosis congenita, there is a male predominance with onset of poikiloderma in late childhood, presence of mucosal and oral leucoplakia, and mental retardation.¹⁰ Bloom syndrome is characterised by extremely short stature, abnormal immunity, neonatal blistering and an increased risk for all types of malignancy at an early age.¹⁰

A genetic test for RECQ4 was not performed for our patient. As most of the features such as early age onset of poikiloderma, photosensitivity and normal intelligence were evident in this case, the clinical diagnosis of RTS could be made.

TREATMENT

Preliminary management involved appropriate preventive procedures, including oral prophylaxis and fluoride application. Preventive restorations were carried out with glass ionomer cements on all the first permanent molars (Teeth 16, 26, 36, 46). Extraction of maxillary left primary central and lateral incisors (Teeth 61, 62); first primary molar (Tooth 64); mandibular left primary central incisor (Tooth 71); and mandibular right central incisor and first molar (Teeth 81, 84) were performed followed by restorations in relation to the maxillary and mandibular left primary second molars (Teeth 65, 75) with glass ionomer cement. After pulpectomy of mandibular left first molar (Tooth 74), canals were filled with MTA as an obturating material followed by stainless steel crown with mandibular left first primary molar (Tooth 74), second primary molar (Tooth 75; figures 7 and 8) and maxillary right first primary molar (Tooth 54; figure 9). Composite restorations were carried out in relation to right and left maxillary primary canines and

Figure 6 Orthopantomograph radiograph showing multiple missing permanent teeth of maxillary and mandibular arch.

Figure 7 Intraoral photograph showing stainless steel crowns with mandibular left first and second primary molars.

mandibular right primary canine (Teeth 63, 73, 83), and final occlusion was checked after the removal of interference, and finishing and polishing of the restoration using sofex tips (figure 10). Band and loop space maintainer in relation to mandibular right first primary molar (Tooth 84; figure 7) and Mayne's space maintainer with respect to maxillary left first primary molar (Tooth 64; figure 9) were given. Strict oral hygiene instruction for tooth brushing and flossing was provided.

OUTCOME AND FOLLOW-UP

The patient was advised to attend regular check-ups every 3 months. Follow-up after 2 years revealed successful obturation of the mandibular left primary first molar (Tooth 74) with MTA.

DISCUSSION

RTS is genodermatosis presenting at an early age with a characteristic facial rash (poikiloderma) as the diagnostic hallmark and heterogeneous clinical features. The RTS, in fact, belongs to a range of hereditary disorders that have common increased cancer vulnerability. Mutations in the RECQ4 gene on

Figure 8 Intraoral periapical photograph showing mandibular left first primary molar obturated with mineral trioxide aggregate.

Figure 9 Intraoral photograph showing stainless steel crown with maxillary right first primary molar.

chromosome 8 programming a DNA helicase of the RECQ family have been recognised in patients with RTS.¹² RECQ mutations are found in about two-thirds of patients with Rubinstein-Taybi syndrome, mainly those with skeletal defects and without cataract, who are classified as RTS type II.²

Dental malformation associated with RTS that included a large diversity of malformations such as abnormal crown formations, microdontia, rudimentary or hypoplastic teeth,¹ abnormal short roots, mostly affecting the maxillary teeth, and multiple missing teeth, especially premolars and third molars, was first described by Rothmund.¹³ These malformations mainly affected the permanent dentition, and were detected in radiographical examination. An increased occurrence of caries has been observed in RTS patients¹⁴ and the overall incidence of dental abnormalities has been estimated to be between 27% and 59% of cases.^{4 13 15}

A short root anomaly and atypical pulp chambers of the permanent incisors have been reported in the mixed dentition period of our patient with RTS, and dental panoramic examination of the parents of the patient excluded a familial pattern of short root anomaly, suggesting the occurrence of this condition in the context of RTS. In the present case, short roots were observed with all the first molars of the maxillary and mandibular arches (Teeth 16, 26, 36 and 46).

The major component, Portland cement, is a mixture of dicalcium silicate, tricalcium silicate, tricalcium aluminate and tetracalcium aluminoferrite. MTA is prepared as a mixture of powder and water in a slurry form, which gradually hardens in the oral environment.⁸ MTA is osteoinductive material and its placement in the root canal causes dissolution to form

hydroxyapatite crystal growth filling the microscopic spaces between MTA and dentinal wall. It is bonded mechanically initially and leads to chemical bonding in later on due to diffusion of ions. In the present case, MTA was obturated with the help of lentulospiral at slow speed and condensed with moist paper points. The release of alkaline phosphatase and osteocalcium, interleukin production, periodontal attachment, cementum growth and dentinal bridge formation will result in a good apical seal and delayed root resorption.⁸

MTA can be used to obturate the root canals of a retained primary tooth where the succedaneous permanent tooth is absent.^{16 17} This technique is not recommended for the obturation of primary teeth that are expected to exfoliate, since it is anticipated that MTA would not resorb, or resorb very slowly. In the present case, the mandibular left first primary molar (Tooth 74) was obturated using MTA expecting that it would remain in the oral cavity for a longer duration. Also, band and loop space maintainers were placed in relation to the maxillary left first primary molar (Tooth 64) and mandibular right first primary molar (Tooth 84), to maintain the space for future option of implant placement.

Although the microbial causation of destructive periodontal diseases is well documented in the literature, genetic disorders can also influence the host susceptibility by altering the immune response, which is also considered as an aetiological factor in periodontal diseases. Haytac C Malso showed that periodontopathogens were absent in RTS cases, which also eliminates the possibility of periodontal disease due to microbial origin. Although antibiotic use during childhood may have altered oral flora, the patient's history revealed that he had not used antibiotics during the past 5 years. Therefore, it is very likely that the periodontal condition of this patient is closely related to the structural defects of the gingiva rather than of microbial origin.

In conclusion, patients with RTS quality of life after dental treatment improves physical, psychological and social aspects and it also has a positive effect on the patient and the patient's family. The long-term effects of dental treatment in such patient, by periodic recall to maintain periodontal status and to permit early detection of oral neoplastic disease, need to be assessed.

Learning points

- ▶ Rothmund-Thomson syndrome (RTS) is a rare autosomal recessive trait disease characterised by skin, eye and skeletal abnormalities, and other abnormalities such as juvenile cataract, short stature, abnormal teeth, congenital bone defects and soft tissue contractures.
- ▶ Despite the patient not being processed for RECQ4, his clinical presentation is suggestive of RTS type II. The main concern for patients with RTS type II is the increased risk of osteosarcoma, which needs careful oncological surveillance.
- ▶ Primary teeth that remain in the oral cavity for a longer duration can be obturated with mineral trioxide aggregate.

Figure 10 Intraoral photograph showing final occlusion after complete treatment.

Contributors NVR and MSB treated the patient. NT wrote the manuscript. SB edited the manuscript.

Competing interests None declared.

Patient consent Obtained.

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