A Rare Case Report of Rothmund Thomson Syndrome

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Abstracts: Rothmund-Thomson syndrome (RTS) is a genodermatosis presenting with a characteristic facial rash (poikiloderma) associated with short stature, sparse scalp hair, sparse or absent eyelashes and/or eyebrows, juvenile cataracts, skeletal abnormalities, radial ray defects, premature aging and a predisposition to cancer. RTS also present dental abnormalities like malformation: microdontia and failure of eruption. We report an unusual patient with Rothmund-Thomson syndrome (RTS), a genodermatosis. The patient is 19 year old with poikiloderma, skeletal abnormalities and dental abnormalities. In this case report evaluation of the various abnormality of the patient has been reviewed. [Payal A NJIRM 2017; 8(5):105-107]

Key Words: Poikiloderma, skeletal abnormalities, Failure of eruption of teeth.

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Introduction: Rothmund Thomson Syndrome (RTS) is a rare genetic autosomal recessive disorder characterized by genodermatosis. Genodermatosis are inherited genetic skin conditions often grouped into three categories: chromosomal, single gene, and polygenetic. RTS was originally described in 1868 by the German ophthalmologist Rothmund who observed poikiloderma, growth retardation and rapidly progressive bilateral juvenile cataracts in 10 children in a Bavarian village. In 1936, the English dermatologist Thomson reported three similar patients with "Poikiloderma congenitale" and growth retardation who displayed skeletal defects, including bilateral thumb aplasia and hypoplastic radii and ulnae, but no cataracts. [1] The syndrome presents in early childhood with characteristic facial rash (poikiloderma), which is the diagnostic hallmark for RTS. Heterogeneous clinical features including short stature, sparse scalp hair, sparse or absent eyelashes and/or eyebrows, juvenile cataracts, skeletal abnormalities, radial ray defects, premature aging and a predisposition to osteosarcoma. The eponym Rothmund- Thomson syndrome was coined by Taylor in 1957 to describe a group of patients with the above-mentioned disorders.[1, 2] Till date, approximately 300 patients have been recorded in the medical literature. At present the rationale for such grouping awaits further knowledge on the genetic basis of the syndrome.

RTS is a very rare disease and reliable data on its prevalence are not available. Due to the highly variable clinical spectrum, this brings together patients with shared and unique developmental defects. Patients who display an atypical/ borderline clinical presentation may be overlooked. Consistent with autosomal recessive transmission, most patients appear as isolated cases but few siblings, mostly from consanguineous families or from small close communities, have been reported.[1, 2]

Case Report: A 19 year old male patient visited our Department of Oral Medicine and Radiology with the chief complaint of pain in upper right and lower left back teeth region accompanied by difficulty in chewing food due to missing teeth since one month. Patient was asymptomatic before 1 month. Gradually he noticed discoloration and pain in upper right and lower left back teeth. He further stated that he experienced mild pain which aggravated on chewing food and relieved by itself.

On general examination, he presented with poikiloderma [figure1] and skeletal abnormality like hypoplasia of finger and thumb of hands since childhood [figure 2]. His height was 152 cm, and he weighed 48 kg. He had tendency of developing facial erythema and oedema after a brief sun exposure, subsiding with hyperpigmentation in 2-3 days. Poikiloderma had developed over years involving the face, neck and back [figure1]. Dental abnormalities like hypoplastic teeth, unusual crown formation, multiple caries and failure of eruption were noticed on intra oral examination [figure 3 & 4]. Restoration was advised in all carious teeth. Extraction of grossly carious 36 (mandibular left molar) was advised followed by oral prophylaxis as well as prosthetic rehabilitation after satisfactory healing. With the diagnosis of RTS, genetic counselling, photo protection, prosthetic correction and long-term follow-up were advised.

Figure 1: Showing Poikiloderma involving the neck and back.

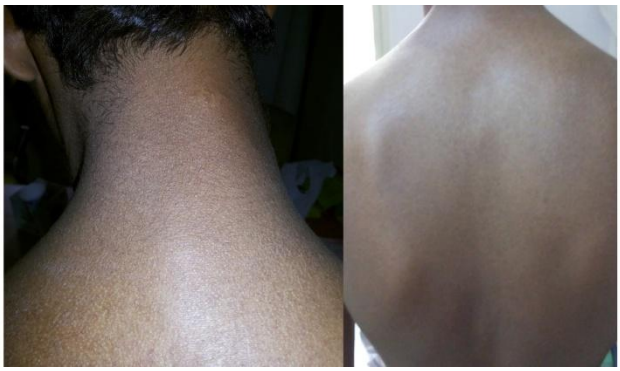


Figure 2: Showing skeletal abnormality like hypoplasia of finger and thumb of hands.

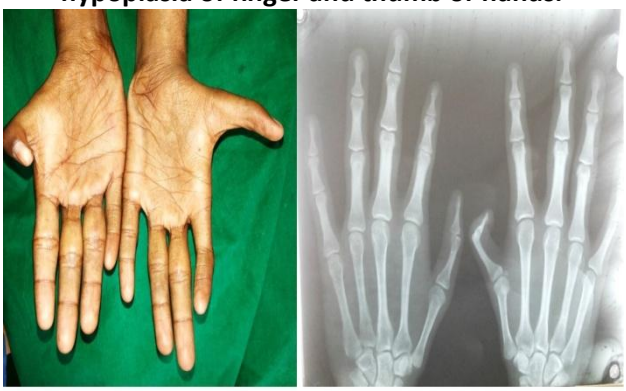
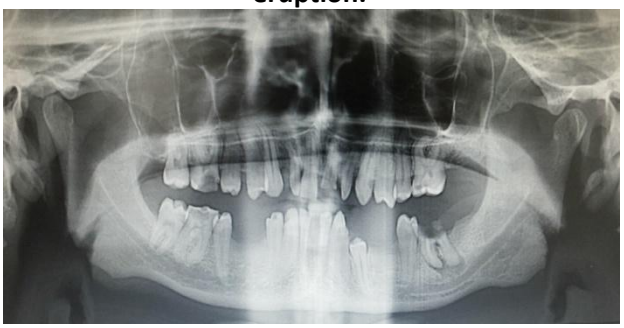


Figure 3: Showing Dental abnormalities like multiple caries and failure of eruption.

Figure 4: Showing radiographic evident of dental abnormalities like multiple caries and failure of eruption.



Discussion: Rothmund described patients who had a rash and juvenile cataracts, but unlike those patients, the patients described by Thomson had osseous abnormalities, including bilateral thumb aplasia and hypoplastic radii and ulnae. In a literature review by Vennos[4] et al. in 1992, 68% of patients were found to have skeletal abnormalities, including frontal bossing, small hands and feet, and long-bone abnormalities, saddle nose, including radial ray defects.[3]

Abnormalities like short stature, hypogonadysm, dystrophic teeth nails, congenital bone defects, soft tissue contractures, mental retardation can also be present. It has been observed that most of the times patients developed the initial skin manifestations during the first year of life, usually within 6 months postnatally. The acute phase can begin in first year of life as red patches or oedematous plaques. Sometimes blistering may be present. Usually, the cheeks are first involved and later it spreads to other areas of the face, the extremities, and the buttocks. Over months to years, the rash enters a chronic stage characterized by poikiloderma (atrophy, telangiectasia, and pigmentary changes).In more than 30% of cases photosensitivity is present. The characteristic skin findings are the most consistent feature of the syndrome. Irregular erythema and oedema of the skin are replaced by reticulated red-brown patches associated with punctate atrophy and telangiectasia (poikiloderma). These characteristic skin changes are typically seen on the face, extensor extremities, and buttocks with sparing of the chest, abdomen, and back.[1, 5]

Acral hyperkeratotic lesions on the elbows, hands, knees and feet can be seen at puberty. Palmar keratoderma has been reported. Patients may have sparse hair, premature greying, and dystrophic or atrophic nails.[5]

Dental abnormalities include malformation: failure of eruption, microdontia, hypoplastic teeth. Dental (microdontia, early caries), gastrointestinal (oesophageal/pyloric stenosis, annular pancreas, chronic emesis, diarrhoea), and haematological (microcytic hypochromic anaemia, leukopenia) abnormalities occur infrequently.[1, 5, 6]

About one half of patients have skeletal abnormalities, most frequently a characteristic face with frontal bossing, saddle nose, and micrognathia. In 20% of

patients small hands and feet disproportionate to the patient's body size are observed. Approximately 10% of patients have absent or malformed radii, and 5% of patients have absent or partially formed thumbs.[5, 6]

Although physical development is frequently retarded (short stature, slender and delicate limbs, small hands, bird like skull), mental development remains normal. 10-40% patients may develop bilateral juvenile cataract by 4-7 years of age and about 30% may develop hypogonadism.[1, 6, 7]

Nails may be dystrophic and hair (scalp, beard, pubic, axillary) may be sparse or absent in 50% cases while eyelashes/eyebrows are sparse/absent in 70% cases.[6, 7]

Patients are at higher risk for extra cutaneous malignancies (osteogenic sarcoma, myelodysplastic syndrome) and skin malignancies (squamous cell or basal cell carcinomas, malignant fibrous histiocytoma) ascribed to defective DNA repair.[6, 7]

Although life expectancy remains unaffected, ultimate prognosis depends upon development / type of malignancy. The diagnosis is determined by characteristic clinical features such as photosensitivity in early infancy, poikiloderma, skeletal deformity and dental abnormalities.[6]

Ectodermal dysplasia is a differential diagnosis of RTS. Ectodermal dysplasia includes abnormalities like hypodontia, hypotrichosis and hyperhidrosis. In this case, hypodontia was present but other features like hypotrichosis and hyperhidrosis were absent.[9] Differentiation of RTS from ectodermal dysplasia is not difficult as poikiloderma is a main characteristic of RTS which is not seen in ectodermal dysplasia. Differentiation from other syndromes of DNA defect repair such as Bloom's syndrome or Cockayne syndrome is not difficult. Development of erythema with cafe-au-lait spots and not pokiloderma, slender built, narrow delicate face, prominent nose and high-pitched voice in Bloom's syndrome are essential differences while poikiloderma, mental retardation and dwarfism, conductive deafness, pigmentary retinopathy/blindness are hallmark features of Cockayne syndrome.[6,8]

Conclusion: Rothmund Thomson syndrome is a very rare genetic disorder. This syndrome resembles ectodermal dysplasia. However the diagnostic hallmark such as poikiloderma and skeletal abnormalities make it different from other developmental disorders. Early diagnosis and appropriate treatment plan leads to improvement in quality of life.

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