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Rothmund-Thomson Syndrome - A Rare Case Report

KEYWORDS: genodermatosis, poikiloderma, shortstature, skeletal abnormalities

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ABSTRACT *we here with report a case of 4 yr old male child with bow legs, mild developmental delay, sparse hair, peg shaped teeth, ricketty features, and hyper and hypo pigmentation. All these findings are consistent with the diagnosis of Rothmund-Thomson syndrome. We are presenting this case because of its rarity.*

Introduction:
Rothmund-Thomson syndrome (RTS) is a rare condition the prevalence of which is unknown.It is a genodermatosis presenting with a characteristic facial rash (poikiloderma) associated with short stature, sparse scalp hair, sparse or absent eyelashes and/or eyebrows,skeletal abnormalities, radial ray defects, premature ageing and a predisposition to cancer.we are herby presenting this case inview of its rarity.

Case report:
A 4 year old male child,of 1st birth order,born to a 2nd degree consanguinous couple, who is developmentally normal presented to our hospital with complaints of not growing well since birth and recurrent episodes of loose stools,bow legs since late infancy.Perinatal history was uneventful.He was stunted and wasted his weight was 10 kgs(<-3SD),height 77 cms(<-3SD).On examination child had no dysmorphic features,he had sparse and hypopigmented hair,peg shaped teeth,widening of wrist and bow legs suggestive of rickets and hyper pigmentation of the trunk and limbs with hypo pigmented macular lesions of palms and soles.On investigations X-ray wrist joint showed widening and fraying of lower end of radius and ulna and x ray skull-open anterior fontanelle.His Hb was 6.2gm/dl (anemic),TLC 7200/mm,TPC 6.6lakh/mm,and peripheral smear suggestive of hypochromic microcytic anemia with polychromasia. Serum Ca-(9.3),Po4(4.2),ALP(124),serum cortisol(200ng/dl), are all within normal limits.Opthalmological examination revealed corneal xerosis and positive Schirmer's test.Skin biopsy was done which was s/o Hypohydrotic Ectodermal Dysplasia,On dental examination child was diagnosed to have Amelogenesis Imperfecta,CT scan brain was normal.

Based on the compatible clustering of all clinical features diagnosis of Rothmund thomson syndrome was made,which requires genetic testing of RECQL4 gene for confirmation.In our case we could not do genetic testing. Child was started on 6lakh IU vit d followed by maintenance of 400IU daily along with iron supplementation for anemia.On follow up X-ray wrist revealed healing rickets.We are planning for 6 monthly evaluation for cataracts,and annual evaluation for malignancies.

Fig (A)

Fig (B)

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Fig (C)

Fig (D)

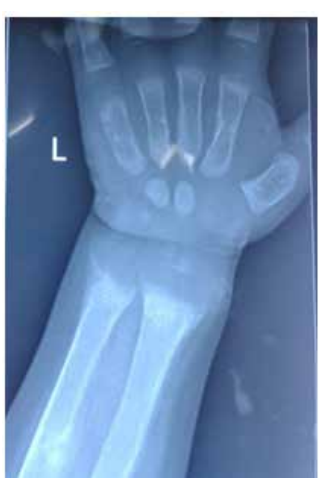

Fig (E)

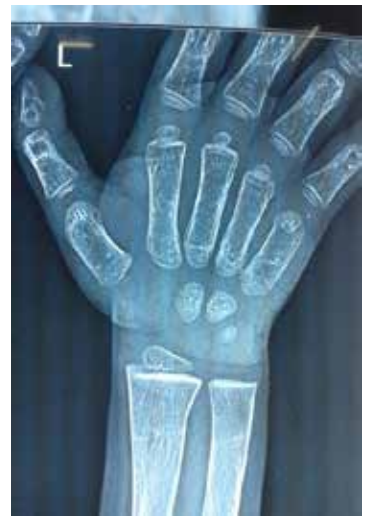

Fig (F)

Description for Images:

A) Hyperpigmentation of skin involving trunk and limbs with bowing of legs.
B) Peg shaped teeth with cheilitis.
C) Hypopigmented sparse hairs.
D) Healed poikilodermal lesions showing hypopigmented macular areas in the palms.
E) Xray wrist joint showing cupping and fraying of distal ends of radius and ulna before vit D therapy.
F) Xray showing white line with zone of provisional calcification after 6 months of vit D therapy suggestive of healing rickets.

Discussion:

The diagnostic hallmark is facial erythema, which spreads to the extremities but spares the trunk, which manifests itself within the first year and then develops into poikiloderma. Two clinical subforms of RTS have been defined: RTSI is characterised by poikiloderma, ectodermal dysplasia and juvenile cataracts, and RTSII characterised by poikiloderma, congenital bone defects and an increased risk of osteosarcoma in childhood and skin cancer later in life. Gastrointestinal, respiratory and haematological signs have been reported. RTSII has autosomal recessive inheritance, and is caused by homozygous or compound heterozygous mutations in the RECQL4 helicase gene, whereas the aetiology in RTSI remains unknown. Diagnosis is based on clinical findings (primarily on the age of onset, spreading and appearance of the poikiloderma) and molecular analysis for RECQL4 mutations.In our case there is poikiloderma,hypohydrotic ectodermal dysplaia,hence clinical diagnosis of Rothmund thomson syndrome type I was done. In RTS I,the skeletal abnormalities may be overt (frontal bossing, saddle nose and congenital radial ray defects),or subtle (visible only by radiographic analysis).in our case child is having rickets as the prominent skeletal manifestation.

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