Case report

Rothmund - Thomson Syndrome Associated With Myelodysplastic Syndrome

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Abstract: Rothmund-Thomson syndrome (RTS) is a rare genodermatoses inherited by autosomal recessive mode. Though data on prevalence of RTS is not available around 300 cases been reported worldwide. RTS mainly manifests with abnormalities in skin, skeleton and eyes. Hematological abnormalities ranging from anemia to leukemia have been reported in few cases. Myelodysplatic syndrome(MDS) presents as acquired pancytopenia caused by bone marrow infiltration. We report such a rare association of MDS in RTS. Key words: Rothmund-Thomson syndrome, poikiloderma, genodermatoses, myelodysplatic syndrome, pancytopenia.

Case report: An male adolescent child aged 13 years brought with history of fever and occasional pain abdomen of 2 weeks duration. On Examination vitals were stable, pallor was present. He had Short stature, hypopigmented and hyperpigmented skin lesions over face, trunk and extremities, sparse eye lashes and eyebrows, dystrophic nails, skeletal deformity in the form of anterior bowing of tibia bilaterally (left more than right). Normal on systemic examination and ophthalmic examination. Investigations revealed pancytopenia, ultrasonogram of abdomen revealed Liver abcess which responded well to medical treatment with intravenous antibiotics given for 14 days. Skin Biopsy histology suggestive of poikiloderma. Bone marrow picture was consistent with MDS. Currently child is under hematological surveillance and being planned for bone marrow transplantation. Genetic studies were not performed on account of availability and affordability concerns.

Discussion: Rothmund- Thomson syndrome (RTS) is a rare genodermatoses 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. Only two thirds of patients with a clinical diagnosis of RTS carry RECQL4 mutations. Diagnosis is based on presentation of variable clinical feautures and poikiloderma. There is no diagnostic test or cytogenetic finding which is specific to RTS. Two forms of RTS described type I RTS, characterised by poikiloderma, ectodermal dysplasia and juvenile cataracts, negative for the RECQL mutation scan and type II RTS, with poikiloderma, congenital bone defects and an increased risk of osteosarcoma related to deleterious RECQL mutations. Although initially the prevalence of cataracts was reported to be as high as 50% in some series, it was subsequently found to be much lower. In our case there was no cataract. The myelodysplastic syndromes are a group of hematologic disorders defined by morphologic abnormalities of the three cell lines. Myelodysplasia in the young and RTS are both rare conditions which are both likely to be due to a common etiologic cause of non repair of stem cell DNA damage. Few cases been reported such association of MDS with RTS. MDS can arise in a previously healthy child and is conformingly named de novo or primary. It may also develop in a child with a known predisposing condition, when it is referred to as secondary. Haematopoietic stem cell transplantation (HSCT) is the treatment of choice and results in cure rates of around 60%. Although some clinical signs suggest precocious aging, the patient’s lifespan is not altered, provided that the neoplastic disease is diagnosed and treated in time. Patients should be managed by a multidisciplinary team and should be offered long term follow-up.
REFERENCES


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