A case of Revesz syndrome

*Chamal Imalke Kankananarachchi1, Nuwan Dilanke Wickramasinghe2, Sadani Rajika Vithana2, Thilina Madushanka Munasinghe2, Sujeewa Deshapriya Amarasena1

DOI: http://dx.doi.org/10.4038/sljch.v47i2.8492
(Key words: Revesz syndrome, dyskeratosis congenita, pancytopenia, cerebellar hypoplasia, nail dystrophy)

Introduction
Classical features of Revesz syndrome (RS) include skin pigmentation, nail dystrophy, oral leukoplakia, cerebellar hypoplasia, growth retardation and delayed psychomotor development1. We report a child with RS with classical multisystem involvement.

Case Report
A 10-year-old boy was investigated for recurrent oral ulcers since late infancy. He was the first child born to non-consanguineous healthy parents following an uneventful antenatal and perinatal period. He had several febrile illnesses in the past requiring oral antibiotics and global developmental delay. He had short stature, microcephaly, reticulate skin pigmentation of upper body, nail dystrophy, alopecia and leukoplakia of oral mucosa (Figure 1).

Neurological examination revealed ataxia without other cerebellar signs. There were no changes in conjunctiva and the anal, urethral, or genital mucosa. He was pale but had no bleeding manifestations. He had no gastrointestinal or pulmonary involvement.

A haemoglobin level of 7.5g/dl, a white blood cell count of 450 per cu mm and a platelet count of 30,000 per cu mm indicated pancytopenia. Bone marrow biopsy was in favour of aplastic anaemia. A computed tomography (CT) scan of the brain showed cerebellar hypoplasia and a few cerebral calcifications (Figure 2).

Discussion
RS was first described in 1992 by Revesz T, et al in a male infant from Sudan1. The second description was in 1994 by Kajtar P, et al in a 2-year-old Hungarian girl2. Since then several case reports have been published in the literature but no cases were reported from Sri Lanka. Though retinal involvement was a consistent finding in many of the published cases, it was not evident in this patient.

Serum calcium, phosphorus and alkaline phosphatase were within normal limits. Dual energy x-ray absorptiometry (DXA) showed <-3SD bone mineral density but he never had fractures.

Diagnosis of RS was made on the clinical criteria. Specific genetic mutation analysis was not carried out due to lack of facilities. He was given one blood transfusions at the age of 10 years due to symptomatic anaemia. From time to time he was treated with granulocyte colony stimulating factor for severe neutropenia. He was commenced on Dapsone as a treatment for underlying haematological abnormality.

In RS there is a heterozygous mutation in TRF1-interacting nuclear factor-2 part of the shelterin telomere protection complex resulting in a short telomere length3. Dyskeratosis congenita (DC) consists of a triad of nail dystrophy, pigmented skin and leukoplakia4. Since RS shares some of the features of DC, it is considered as an overlap syndrome of it. In addition to features seen in DC, growth retardation, microcephaly, cerebellar hypoplasia and bilateral exudative retinopathy are evident in RS1. All these features were seen in this patient except the ophthalmological manifestations. Scheinfeld MH et al presented the neuro-radiological findings of a patient with RS in which intracranial calcification, cerebellar hypoplasia and unusual brain lesions were described5. Similar neuroimaging findings were observed in this patient, but he did not have brain lesions.

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1Faculty of Medicine, University of Ruhuna, Sri Lanka, 2Teaching Hospital Karapitiya, Sri Lanka

*Correspondence: imalke462@gmail.com

(Received on 01 August 2017; Accepted after revision on 15 September 2017)

The authors declare that there are no conflicts of interest.

Personal funding was used for the project.

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Management of both DC and RS is similar. Androgens, granulocytes colony stimulating factor and bone marrow transplantation are the available treatment modalities. There are no published data on usage of immunosuppressive in DC. Despite appropriate interventions, majority of the previously described patients died before they reach the second decade. Complications related to bone marrow suppression were the main reason for mortality.
References


