A case of Revesz syndrome

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Introduction
Classical features of Revesz syndrome (RS) include skin pigmentation, nail dystrophy, oral leukoplakia, cerebellar hypoplasia, growth retardation and delayed psychomotor development[1]. 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).

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 the neutropenia. He was commenced on Dapsone as a treatment for underlying haematological abnormality.

Discussion
RS was first described in 1992 by Revesz T, *et al* in a male infant from Sudan[1]. The second description was in 1994 by Kajtar P, *et al* in a 2-year-old Hungarian girl[2]. 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.

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 length[3]. Dyskeratosis congenita (DC) consists of a triad of nail dystrophy, pigmented skin and leukoplakia[4]. 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 RS[1]. 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 described[5]. Similar neuroimaging findings were observed in this patient, but he did not have brain lesions.

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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[3]. 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.

Figure 1: ***Ten year old boy with Revesz syndrome***

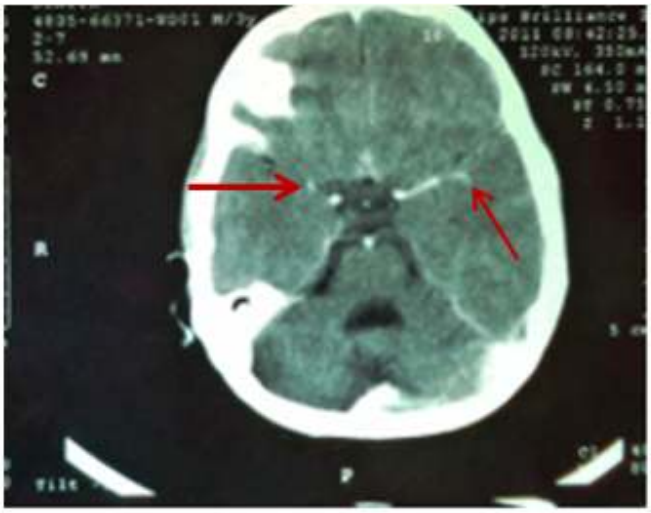

Figure 2: CT scan of brain showing calcificaion

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