Revesz syndrome masquerading as bilateral cicatricial retinopathy of prematurity

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Purpose: We describe for the first time cicatricial retinopathy as the presenting feature in a case of Revesz syndrome. We wish to highlight this syndrome as a rare but important cause of bilateral retinopathy in childhood which may masquerade as retinopathy of prematurity.

Methods: The defining features of Revesz syndrome are retinopathy, aplastic anaemia, cerebellar hypoplasia and nail dystrophy. It is a variant of Dyskeratosis Congenita, a heterogeneous, heritable spectrum of primarily haematological disorders characterised by shortened telomere length.

Results: We outline the clinical features of a case of Revesz Syndrome, comment on its pathophysiology, and review the limited published literature.

Conclusions: Revesz Syndrome should be considered in all children presenting with bilateral retinal detachment. Early diagnosis allows for rapid, appropriate management of the associated haematological disease, in particular, timely hematopoietic stem cell transplant. Ophthalmic review of children suspected of having Dyskeratosis Congenita is also required. This will provide for prompt treatment of the sight threatening retinopathy found in some cases.