

Ophthalmic Images

Mud-splattered fundus

A 12-month-old girl was referred for leukocoria, left eye. The right eye showed [Fig. 1a] radial streaking of retinal pigment epithelium (RPE) and a small 3 mm × 3 mm retinoblastoma (arrow) [Fig. 1b]. Fluorescein angiography demonstrated prominent blockage from the streaks and slight hyperfluorescence of the mass. The left eye had extensive retinoblastoma with total retinal detachment and no view of underlying RPE. The mother demonstrated similar RPE changes without retinoblastoma. Genetic testing confirmed mutations of X-linked OA1 gene and chromosome 13 RB1 gene. The “mud-splattered fundus” occurs due to lyonization of X-chromosome and morphogenesis of RPE precursors with “pinwheeling” and “spreading” during embryogenesis, observed in carrier states of X-linked ocular albinism.^[1]

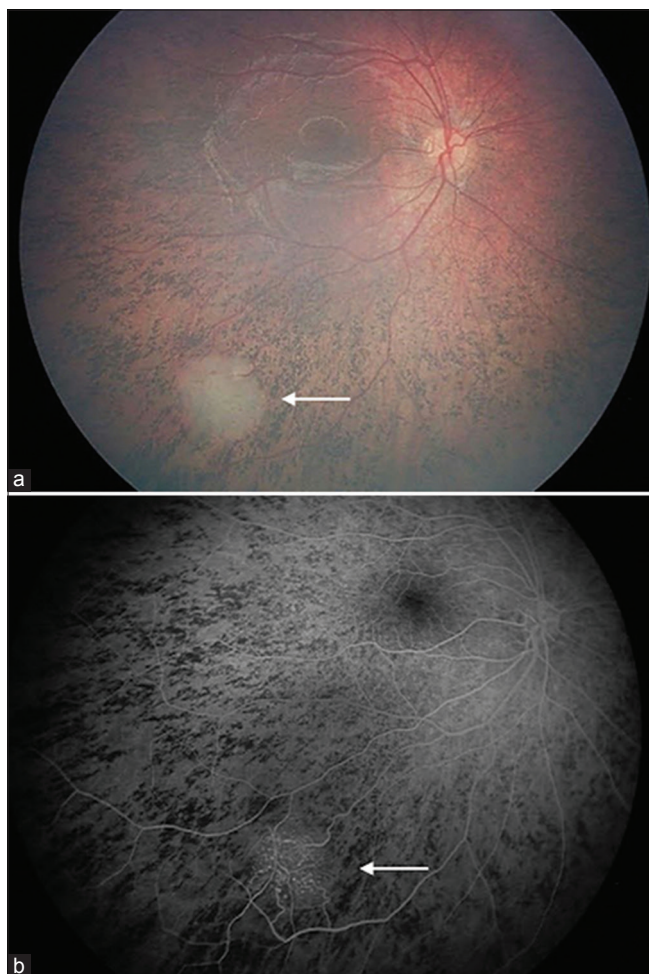


Figure 1: RetCam fundus photograph (a) and fluorescein angiography (b) demonstrating radial streaking of the retinal pigment epithelium and a small retinoblastoma of 3 mm diameter with slight hyperfluorescence (arrow)

Conclusion

This case represents a rare combination of X-linked ocular albinism carrier state with characteristic “mud-splattered fundus” and unrelated germ-line retinoblastoma.

Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent forms. In the form the patient(s) has/have given his/her/their consent for his/her/their images and other clinical information to be reported in the journal. The patients understand that their names and initials will not be published and due efforts will be made to conceal their identity, but anonymity cannot be guaranteed.

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Conflicts of interest

There are no conflicts of interest.

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Reference

1. Moshiri A, Scholl HP, Canto-Soler MV, Goldberg MF. Morphogenetic model for radial streaking in the fundus of the carrier state of X-linked albinism. *JAMA Ophthalmol* 2013;131:691-3.

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