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Microphthalmia-retinitis pigmentosa-foveoschisis-optic disc drusen syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base.

Microphthalmia-retinitis pigmentosa-foveoschisis-optic disc drusen syndrome.

ORPHA:251279

Microphthalmia-retinitis pigmentosa-foveoschisis-optic disc drusen syndrome is a rare, genetic, non-syndromic developmental defect of the eye disorder characterized by the association of posterior microphthalmia, retinal dystrophy compatible with retinitis pigmentosa, localized foveal schisis and optic disc drusen. Patients present high hyperopia, usually adult-onset progressive nyctalopia and reduced visual acuity, and, on occasion, acute-angle glaucoma.