ROD-CONE DYSTROPHY AND THE IMPORTANCE OF GENETIC ANALYSIS FOR DIAGNOSIS

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OBJECTIVE

Report the case of a patient diagnosed with Rod Cone Dystrophy (RCD).

METHODS

Analysis of medical records and genetic exam.

RESULTS

Female, 13 years old, comes to the ophthalmology service complaining of low visual acuity in both eyes, accompanied by hemeralopia and photophobia. Denies family or personal history of eye diseases and surgeries. On examination, best corrected visual acuity of 20/200. Biomicroscopy without particularities. Wide-field colour-corrected (figure 1) imaged illustrated multiple grayish dots and areas of retinal pigment epithelium atrophy and flecks. Autofluorescence (figure 2) showed blotchy hypoautofluorescence in the posterior pole and the ring of hyperautofluorescence bounding the atrophic area. Optical coherence tomography (figure 3) showed diffuse external retinal loss and retinal thinning. A genetic test was requested, showed a heterozygous pathogenic mutation and two ABCA4 variants of undetermined significance.

IMAGES

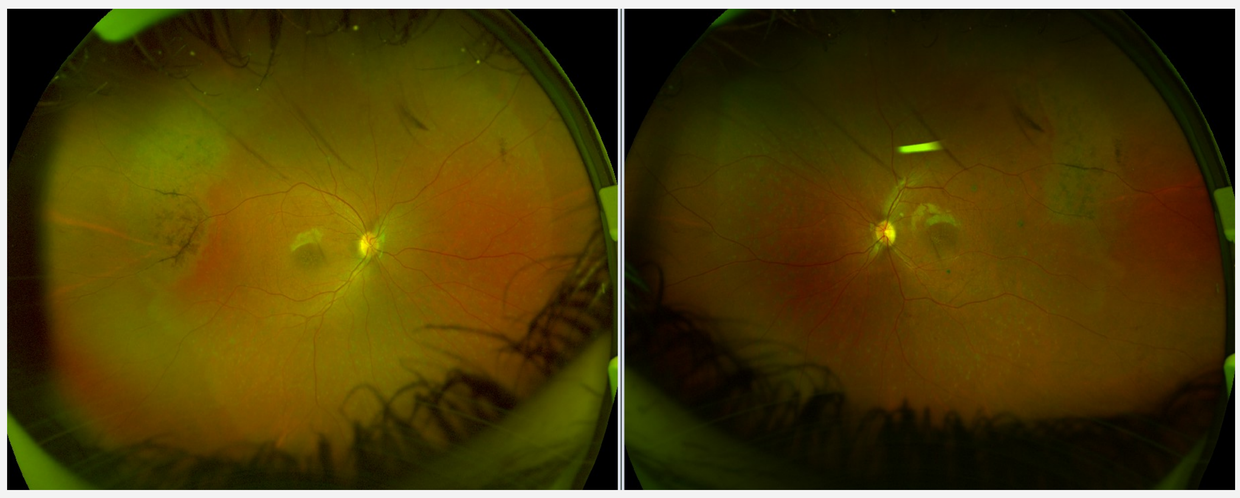

Figure 1.

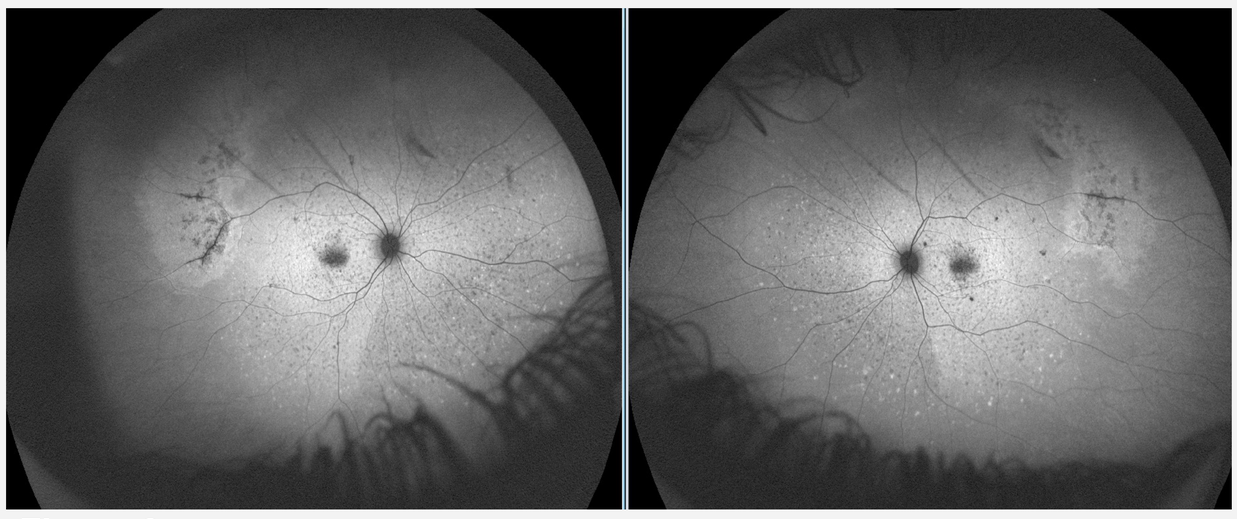

Figure 2.

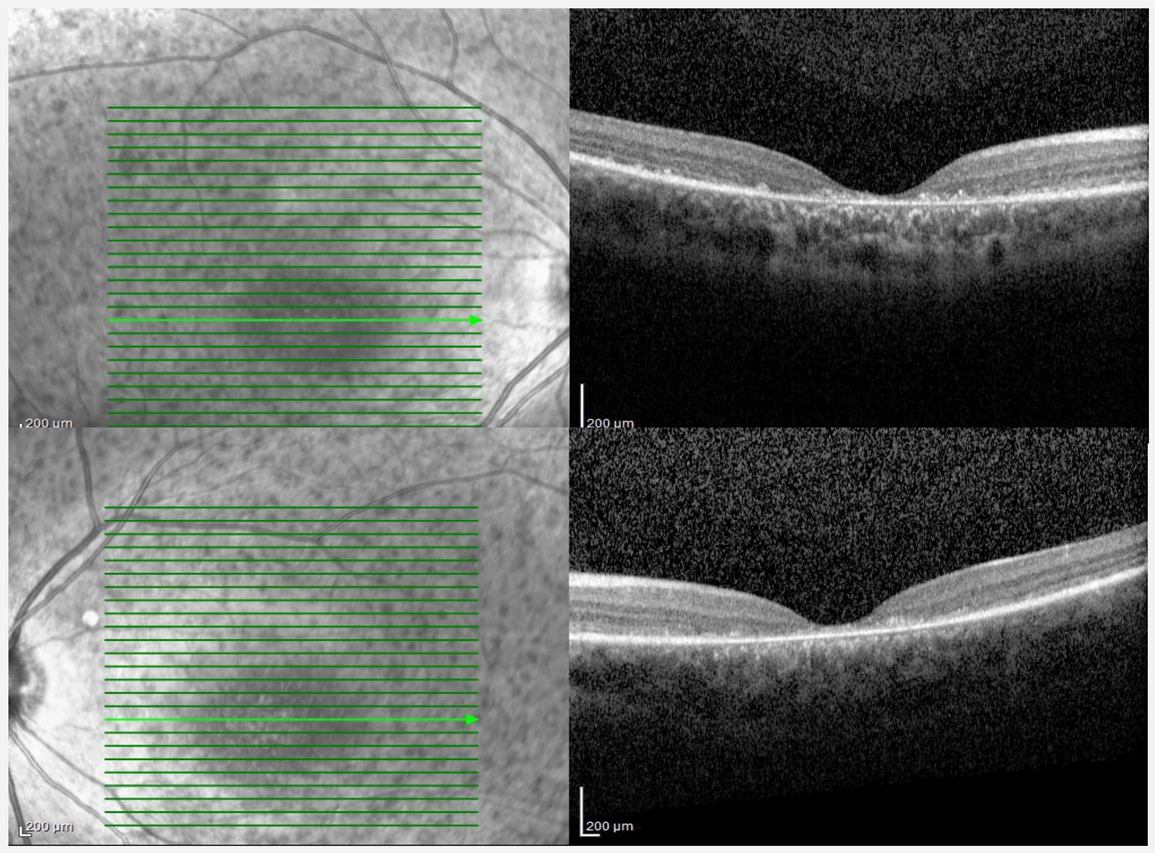

Figure 3.

DISCUSSION AND CONCLUSIONS

RCD is a group of hereditary retinal diseases with progressive degeneration of photoreceptors, with cones being more affected than rods, and may be related to an autosomal recessive, dominant or X-linked inheritance. Only 25% of cases are caused by identified genes. Mutation in the ABCA4 is the most common cause of autosomal recessive inheritance.

Symptoms commonly start in schoolchildren and young adults, with decreased night vision, photophobia, progressive loss of the visual field, low visual acuity, discromatopsia, hemeralopia, and may present with central scotoma. The diagnosis is made through clinical history, ophthalmological examination and genetic tests. The electrophysiological field test detects alterations earlier than the clinical examination.

In the reported case, visual prognosis was explained, the patient was referred to subnormal vision testing and was instructed about family planning and risk of consanguinity.

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