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Service (sector) Retina and Vitreous Nº CEP

OCULAR ABNORMALITIES AND GENETIC FINDINGS IN THE MARFAN SYNDROME

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PURPOSE: Identify the ocular abnormalities in Marfan Syndrome patients. METHODS: Prospective study of 46 Marfan patients with complete ophthalmologic evaluation. Seventeen patients also underwent clinical genetic and molecular analysis. RESULTS: Among 46 patients included in this study, the following ocular abnormalities were found most frequently: ectopia lentis (67,3%), hypoplastic irides (67,3%), retinal detachment (7,6%), flattened cornea (2,2%), megalocornea (2,2%) and myopia (34,8%). Five patients (10,9%) presented with a normal ocular evaluation in both eyes. A pathogenic mutation different from those published in other studies was found in one patient. CONCLUSIONS: Ocular abnormalities in the Marfan Syndrome are frequent. The better understanding of the FBN-1 gene and its expression in the eye are very helpful tools to the diagnosis and treatment of the abnormalities in this condition.

Keywords: ocular abnormalities / Marfan Syndrome / genetics