P-037 - OPHTHALMOLOGICAL MANIFESTATIONS OF PATIENTS OF THE COLOMBIAN SOUTHWESTERN WITH CLINICAL, ENZYMATICAL AND MOLECULAR DIAGNOSIS OF MUCOPOLYSACCHARIDOSIS II, IV-A AND VI.

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**INTRODUCTION:** Mucopolysaccharidoses (MPS) are a group of hereditary metabolic diseases caused by the deficiency of lysosomal enzymes necessary to degrade glycosaminoglycans (GAG). The GAGs without degrading accumulate causing dysfunctions in cells, tissues and organs. The cornea is an organ that can be affected by the blockages of the metabolic pathways, which compromises the transparency, the optical function and its structural integrity. These accumulations of GAGs have also been optically related to papilledema, retinitis pigmentosa, optic neuropathy, glaucoma and even blindness.

**OBJECTIVE:** To characterize the ophthalmological manifestations of patients with MPS

**METHODS:** A retrospective study of the clinical, ophthalmological findings of 35 patients diagnosed with MPS type II, IV-A and VI of the Colombian southwestern region. The ocular results were analyzed, taking into account the type of MPS, sex, age during the study, ophthalmological tests and diagnoses.

**RESULTS:** Thirty-five patients diagnosed with MPS, 18 male and 17 female aged between 2 and 60 years, were evaluated. The ophthalmological results showed that for the three types of MPS studied (II, IV-A and VI) the most frequent diagnoses were the light opacity in the cornea (42%), the transparent lens (14%) and hyperopia and astigmatism (14%). Some 8.5% had refraction disorders such as anisometropia or presbyopia. 5% of the patients presented acute atopic conjunctivitis and 31.4% had a normal vision diagnosis. In the case of those affected by MPS II, the incidence of mild opacity in the cornea was reported in 66% of the cases, 25% for MPS IV-A and 60% in patients with LV. No patient presented with pigmentary retinitis or blindness. The typical ocular features derived from the MPS were more easily recognized in patients older than 6 years.

**CONCLUSION:** Ophthalmological complications that cause a significant reduction in vision are common in MPS. A large number of patients with MPS from the Colombian southwestern present corneal opacification, which can lead to difficulties in their diagnosis and follow-up. Therefore, it is necessary to promote timely diagnosis, monitoring and treatment of the disease, to recognize and evaluate the typical ocular features that appear early in patients with MPS thus ensuring a better quality of life.