P-036 - OTORHINOLARYNGOLOGICAL FINDINGS OF PATIENTS OF THE COLOMBIAN SOUTHWEST WITH CLINICAL, ENZYMATIC AND MOLECULAR DIAGNOSIS OF MUCOPOLISACARIDOSIS II, IV-A AND VI.

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INTRODUCTION: Mucopolysaccharidosis (MPS) is characterized by excessive accumulation of glycosaminoglycan sulfate (GAGs) in organs and tissues. The accumulation of GAGs in the upper respiratory tract generates damage to the mucosa, produces respiratory infections and conditions such as hypertrophy of the tonsils and adenoids, macroglossia, apnea and otitis media; secondarily, it has been related to hearing loss. OBJECTIVE: To characterize the otorhinolaryngological manifestations of patients with MPS

METHODS: Thirty-five patients (18 male and 17 female) with a diagnosis of MPS type II, IV-A and VI of the Colombian southwestern region aged between 2 and 60 years were evaluated. The otorhinolaryngological results of the patients were analyzed taking into account the type of MPS, sex, age during the study, audiological tests, audiometry test according to their age, and hearing treatments.

RESULTS: 6 cases of MPS II, 24 of MPS IV-A and 5 of MPS IV were reviewed. 65.64% of the patients evaluated according to the types of MPS (II, IV-A and VI), presented mild conductive hearing loss (17.14%) and bilateral sensorineural hearing loss (47.5%). 11.3% presented hypertrophy of the tonsils, 17.10% diagnosed a short neck and 5.7% presented apnea-hypopnea syndrome and macroglossia. 47.8% of the patients presented otitis media (OM). OM was reported in patients with MPS II in 33%, and in patients with MPS IV-A and VI occurred in 12.5% and 20% respectively. 20% of patients received treatment with hearing aids. No patient reported otosclerosis or tinnitus. A direct relationship of hearing loss was found as age progressed.

CONCLUSIONS: In patients with different types of MPS, there is a high prevalence and progressive tendency to suffer audiological losses and recurrent infections, which is why a periodic follow-up is required, given the important repercussion of this pathology in the quality of life and in the development of these patients The early diagnosis of patients with MPS as well as the development of audiological studies allows us to approach a personalized medicine based on the initiation of an adequate enzyme replacement therapy or other complementary therapies at an early age.