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ORIGINAL ARTICLE

# Audiologic evaluations of children with mucopolysaccharidosis☆

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**KEYWORDS**
Mucopolysaccharidosis;
Hearing loss;
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**Abstract**
*Introduction:* Mucopolysaccharidosis is a hereditary lysosomal storage disease, which develops due to a deficiency in the enzymes that play a role in the metabolism of glycosaminoglycans (GAG). The incidence of mucopolysaccharidosis is 1/25,000, with autosomal recessive inheritance (except for MPS II). Mucopolysaccharidosis occurs in seven different types, each with a different congenital deficiency of lysosomal enzymes. In mucopolysaccharidosis patients, even though progression of clinical findings is not prominent, the disease advances and causes death at early ages. Facial dysmorphism, growth retardation, mental retardation, and skeletal or joint dysplasia are the most frequently found symptoms in these patients.
*Objective:* The purpose of our study is to present the types of hearing loss types and tympanometric findings of patients with mucopolysaccharidosis referred to our clinic with suspicion of hearing loss.
*Methods:* After otorhinolaryngological examination, 9 patients with different types of mucopolysaccharidosis, underwent to immittance and audiometric evaluations, performed according to their physical and mental abilities, and ages, in order to determine their hearing thresholds.
*Results:* The audiometric findings of the 9 patients followed with mucopolysaccharidosis were reported separately for each case.

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*Conclusion:* Based on the high frequency of hearing loss in mucopolysaccharidosis patients, early and detailed audiological evaluations are highly desirable. Therefore, regular and systematic multidisciplinary evaluations are very important.


**PALAVRAS-CHAVE**
Mucopolissacaridose;
Perda auditiva;
Avaliação audiológica

**Avaliação audiológica em crianças com mucopolissacaridose**

**Resumo**
*Introdução:* Mucopolissacaridose (MPS) é uma doença hereditária de depósito lisossômico, decorrente da deficiência das enzimas que influenciam o metabolismo dos glicosaminoglicanos (GAGs). A incidência de MPS é de 1/25.000 habitantes, resultante de herança autossômica recessiva (exceto no caso de MPS II). MPS se apresenta na forma de sete tipos diferentes e, em cada tipo, ocorre uma deficiência congênita distinta de enzimas lisossômicas. Embora em pacientes com MPS os achados clínicos não sejam geralmente observados, a doença progride em seu curso natural e costuma levar a óbito pacientes muito jovens. Dismorfismo facial, retardo de crescimento, retardo mental e displasia esquelética ou articular são os sinais e sintomas mais frequentemente observados nesses pacientes.
*Objetivo:* A finalidade do presente estudo foi apresentar os tipos de perda auditiva e os achados timpanométricos de pacientes com MPS encaminhados para nossa clínica com suspeita de perda auditiva.
*Método:* Em seguida ao exame otorrinolaringológico, nove pacientes com diferentes tipos de MPS, foram submetidos a avaliações imitanciométricas e testes audiométricos de acordo com sua faixa etária eficiência física e mental, no intuito de identificar seus limiares auditivos.
*Resultados:* Os achados audiométricos dos nove pacientes acompanhados por MPS foram descritos separadamente para cada caso.
*Conclusão:* Baseado na alta frequência de perda auditiva em pacientes com MPS, avaliação audiológica precoce e detalhada é altamente desejável. Para tanto, é importante que sejam realizados avaliações multidisciplinares periódicas e sistemáticas.


## Introduction

Mucopolysaccharidosis (MPS) is a hereditary lysosomal storage disease, which develops due to the deficiency in the enzymes that play a role in the metabolism of glycosaminoglycans (GAG). Progressive GAG accumulation causes advanced growth retardation, skeletal deformities, hearing loss, weak joint movement, and facial dysmorphism. In accordance with the deficiency of the known 11 enzymes, MPS has seven sub-types (MPS I, MPS II, MPS III, MPS IV, MPS VI, MPS VII, MPS IX). Although the incidence of MPS in accordance with its sub-types varies, a general incidence of 1/25,000 has been reported.[1–3]

In patients with MPS, hearing loss is observed due to various reasons. Conductive hearing loss (CHL) may develop due to recurrent upper respiratory tract infections and serous otitis media or bone chain deformities. Sensorineural hearing loss (SHL) is thought to be caused due to the accumulation of GAG in the cochlea, auditory nerve, and brain stem. In many patients, mixed-type hearing loss, along with the symptoms of conductive and sensorineural hearing loss, can also be seen. In CHL related to middle ear effusion, while the ventilation tube implementation is frequently used as treatment, hearing aids are advised in SHL.[4–7] Early diagnosis of hearing losses carries great importance in terms of early intervention in MPS patients, as well as in all other patients.[8,9]

The purpose of the present study was to present the hearing loss type, degree, and tympanometric findings of patients with MPS, who are directed to this clinic with the suspicion of hearing loss.

## Methods

The nine patients who had been directed to this department with the suspicion of hearing loss, whose follow-up had been conducted after they have been diagnosed with MPS, were included in this study (Ethical Committee approval number: GÜKAEK-461). The otological examinations of the patients were performed by the otorhinolaryngologist. Information related to each patient's otorhinolaryngological complaints and treatments were compiled and middle ear infection, ear discharge, tinnitus, dizziness, tympanum problems, adenoidectomy, ear tube placement, and hearing aid usage were determined. After the otorhinolaryngological examination, the immitancemetric evaluations were conducted and