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ORIGINAL ARTICLE

The impact of laronidase treatment in otolaryngological manifestations of patients with mucopolysaccharidosis☆

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KEYWORDS
Mucopolysaccharidosis I;
Laronidase;
Enzymatic replacement therapy;
URT infections;
Sleep apnea;
Hearing loss

Abstract
Introduction: Mucopolysaccharidosis (MPS) is a lysosomal storage disease caused by deficiency of α-L-iduronidase. The otolaryngological findings include hearing loss, otorrhea, recurrent otitis, hypertrophy of tonsils and adenoid, recurrent rhinosinusitis, speech disorders, snoring, oral breathing and nasal obstruction.
Objective: To evaluate the impact of enzymatic replacement therapy with laronidase (Aldurazyme®) in patients with mucopolysaccharidosis (MPS I), regarding sleep and hearing disorders, and clinical manifestations in the upper respiratory tract (URT).
Methods: Nine patients with MPS I (8 Hurler-Scheie, and 1 Scheie phenotypes) of both sexes, ages ranging between 3 and 20 years, were included in this study. Patients were evaluated between seven and 11 months before the treatment and between 16 and 22 months after the onset of the enzymatic replacement. They were all submitted to a clinical and otolaryngological evaluation, including nasofibroscopical, polysomnographic and audiologic exams.
Results: The results' data showed decreasing of the frequency of ear, nose and throat infections, with improvement of the rhinorrhea and respiratory quality. No remarkable changes were observed regarding macroglossia and tonsil and adenoid hypertrophy. Audiometric and polysomnographic evaluations did not show statistical significance.

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Conclusion: Enzymatic replacement therapy in patients with mucopolysaccharidosis I provides control of recurrent URT infections, rhinorrhea and respiratory quality, however it is does not seem to improve audiologic and polisomnographic parameters, with no effect on adenoid and tonsils hypertrophy and macroglossia.
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PALAVRAS-CHAVE
Mucopolissacaridose I;
Laronidase;
Terapia de reposição enzimática;
Infecções do TRS;
Apneia do sono;
Perda auditiva

Impacto do tratamento com laronidase nas manifestações otorrinolaringológicas de pacientes com mucopolissacaridose

Resumo
Introdução: Mucopolissacaridose (MPS) é uma doença de depósito lisossômico causada pela deficiência de α-L-iduronidase. Os achados otorrinolaringológicos incluem perda auditiva, otorreia, otites de repetição, hipertrofia adenotonsilar, rinossinusite recorrente, distúrbios da fala, roncos, respiração bucal e obstrução nasal.
Objetivo: Avaliar o impacto da terapia de reposição enzimática com laronidase (Aldurazyme®) em pacientes com mucopolissacaridose I (MPS I) em relação ao sono, distúrbios auditivos e manifestações clínicas do trato respiratório superior (TRS).
Método: Nove pacientes com MPS I (oito com fenótipo Hurler-Scheie e um com fenótipo Scheie), de ambos os sexos, com idades variando entre 3 e 20 anos, foram incluídos neste estudo. Os pacientes foram avaliados entre 7 e 11 meses antes do tratamento e entre 16 e 22 meses após o início da reposição enzimática. Todos foram submetidos a uma avaliação clínica e otorrinolaringológica, incluindo nasofibroscopia, polissonografia e exames radiológicos.
Resultados: Os dados dos resultados mostraram diminuição da frequência de infecções de orelha, nariz e garganta, com melhora da rinorreia e da qualidade respiratória. Mudanças significativas não foram observadas em relação à macroglossia e à hipertrofia adenotonsilar. Avaliações audiométricas e polissonográficas não apresentaram significância estatística.
Conclusão: A terapia de reposição enzimática em pacientes com mucopolissacaridose I fornece controle de infecções recorrentes do TRS, rinorreia e qualidade respiratória, porém, não parece melhorar os parâmetros audiológicos e polissonográficos, ou exercer efeito sobre a hipertrofia adenotonsilar e macroglossia.
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Introduction

Mucopolysaccharidosis (MPS) is a lysosomal storage disease caused by deficiency of an enzyme involved in the degradation of glycosaminoglycans (GAGs). They are classified according to the involved enzyme and GAG in seven types: I (Hurler, Hurler Scheie and Scheie), II (Hunter), III (Sanfillipo), IV (Morquio), VI (Maroteaux Lamy), VII (Sly) e IX (Natowicz).[1–3]

MPS I is caused by deficiency of α-L-iduronidase, which leads to intralysosomal deposits of dermatan and heparan sulfate. It is an autosomic recessive genetic disease, with estimated incidence varying from 1:100,000 for severe cases to 1:800,000 for cases with mild manifestations.[4]

Clinical manifestations of MPS I are extremely heterogeneous, with symptoms that evolve in many ways, from very mild manifestations of late development, without cognitive disorders, and high life time expectation (Scheie), to very severe cases of early onset, rapidly progressive, with neural degeneration and limited capabilities in life, usually manifested by the first decade (Hurler), and passing through an intermediary level of severity (Hurler–Scheie).[1,2,5]

The disease may involve nervous, skeleton, digestive, cardiac, superior and inferior respiratory systems presenting different levels of severity in an independent manner. Regarding the otolaryngological findings, the most frequent symptoms include hearing loss, otorrhea, recurrent otitis, hypertrophy of tonsils and adenoid, recurrent rhinosinusitis, speech disorders, snoring, oral breathing and nasal obstruction.[1,2,6]

Obstructive sleep apnea and hypopnea syndrome (OSAHS) is frequently diagnosed in MPS I patients. Obstructive and restrictive factors such as reduction of the thoracic volume (musculoskeletal alterations), restriction of the diaphragmatic movement due to hepatosplenomegaly, presence of atelectasis secondary to the reduction of the lung volume, deposit of GAGs into the pulmonary interstitial tissue, tracheal stenosis, vocal cord thickening, adenoid and tonsil hypertrophy, macroglossia, short neck, thickened high

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