months old. 14.48% of scheduled patients did not attend the appointment. Conclusion: In this sample, the most prevalent reason for referral was evaluation for cochlear implant surgery. Overall, children were, on average, 42.3 months at referral. Patients with a neonatal screening alteration were the youngest group at referral (14.5 months), highlighting the importance of this test; children with suspected ASD were the oldest (88.9 months).

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MEAN AGE AT COCHLEAR IMPLANTATION IN CHILDREN WITH PRELINGUAL DEAFNESS ACCORDING TO ETIOLOGIES

CATEGORIA DO TRABALHO: PESQUISA

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Introduction: Timely diagnosis and treatment of hearing loss in children is imperative. When cochlear implantation (CI) is indicated, it must be performed as early as possible for adequate language development. Some etiologies of hearing loss allow for a diagnosis earlier than others. Objective: To evaluate the mean age of children with prelingual deafness who underwent CI at Porto Alegres Clinical Hospital (HCPA), and compare mean age between different etiologies of hearing loss. Methods: The medical records of 212 pediatric patients who underwent CI at HCPA January 2009 to December 2020 were analyzed. Results: The overall mean age at CI was 43.2 months (± 26.6). In 53.77% of patients the etiology was indeterminate, and the mean age at CI for this group was 42.4 months. Patients with hearing loss due to perinatal complications were the second largest subgroup (17.92%); mean age at CI was 37 months. The remaining etiologies and mean ages at CI were: meningitis (6.13%), 48.7 months; GJB2 or GJB6 mutation (5.66%), 37.4 months; genetic syndrome (5.19%), 37.8 months; inner ear malformation (4.25%), 71.1 months; neonatal infection (2.36%), 33.8 months; auditory neuropathy (2.36%), 40.8 months; and central etiology (2.36%), 39.4 months. Conclusion: The overwhelming majority of patients underwent CI between the first and sixth year of life. Three patients (two with undetermined etiology, one with an inner ear malformation) underwent CI at a very late age (17 - 18 years). Removing these outliers, the mean ages at CI were very similar between subgroups.

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ANÁLISE DA PREVALÊNCIA DA MUTAÇÃO 35DELG DA CONEXINA 26, FATORES DE RISCO E GRAU DE SURDEZ EM PACIENTES PEDIÁTRICOS DO AMBULATÓRIO DE SURDEZ INFANTIL DO HOSPITAL DE CLÍNICAS DE PORTO ALEGRE

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Introdução: As mutações no gene da Conexina 26 (GJB2) determinam deficiência auditiva. Isso ocorre porque a estrutura proteica das junções intracelulares das células ciliadas sensoriais está alterada, prejudicando a remoção dos íons potássio e, dessa forma, impossibilitando a excitação a novos estímulos sonoros. Objetivo: O presente trabalho visou analisar a prevalência da mutação 35delG no gene GJB2, os fatores de risco e o grau de surdez em pacientes encaminhados ao ambulatório de surdez infantil do HCPA (Hospital de Clínicas de Porto Alegre). Métodos: Foram analisados os prontuários de 220 pacientes que realizaram o estudo molecular da Conexina 26 pelo método PCR (reação em cadeia da polimerase). Resultados: 26 pacientes (11,82%) apresentaram a mutação 35delG, 16 (7,27%) eram homozigotos e 10 (4,55%) heterozigotos. A partir dessa amostra que continha a mutação, 22 pacientes apresentavam informações de grau e fatores de risco para surdez infantil. 20 (90,91%) apresentavam grau profundo e 2 (9,09%) grau severo. Acerca dos fatores de risco, 10 (45,45%) não apresentavam nenhum fator; 5 (22,73%) relataram histórico familiar de surdez; 3 (13,64%) múltiplos fatores; 3 (13,64%) otite média aguda recorrente; 1 (4,55%) consanguinidade. Conclusão: Os achados do estudo corroboram que o grau de perda auditiva profunda é a intensidade mais comum encontrada em pacientes com a mutação do GJB2, bem como a sua associação à história