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ORIGINAL ARTICLE

Evaluation of Family History of Permanent Hearing Loss in Childhood as a Risk Indicator in Universal Screening☆

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KEYWORDS
Risk factor;
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Abstract
Introduction and objective: Sixty percent of prelingual hearing loss is of genetic origin. A family history of permanent childhood hearing loss is a risk factor. The objective of the study is to determine the relationship between this risk factor and hearing loss. We have evaluated clinical and epidemiological characteristics and related nonsyndromic genetic variation.
Material and method: This was a retrospective, descriptive and observational study of newborns between January 2007 and December 2010 with family history as risk factor for hearing loss using transient evoked otoacoustic emissions and auditory brainstem response.
Results: A total of 26,717 children were born. Eight hundred and fifty-seven (3.2%) had family history. Fifty-seven (0.21%) failed to pass the second test. A percentage of 29.1 (n=16) had another risk factor, and 17.8% (n=9) had no classical risk factor. No risk factor was related to the hearing loss except heart disease. Seventy-six point four percent had normal hearing and 23.6% hearing loss. The mean of family members with hearing loss was 1.25. On genetic testing, 82.86% of homozygotes was normal, 11.43% heterozygosity in Connexin 26 gene (35delG), 2.86% R143W heterozygosity in the same gene and 2.86% mutant homozygotes (35delG). We found no relationship between hearing loss and mutated allele.
Conclusions: The percentage of children with a family history and hearing loss is higher than expected in the general population. The genetic profile requires updating to clarify the relationship between hearing loss and heart disease, family history and the low prevalence in the mutations analyzed.

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PALABRAS CLAVE
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Detección precoz;
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Evaluación de la historia familiar de hipoacusia permanente en la infancia como factor de riesgo en el cribado universal

Resumen
Introducción y objetivo: El 60% de las hipoacusias prelinguales tienen un origen genético. Entre los factores de riesgo se encuentra el antecedente familiar de hipoacusia permanente en la infancia. El objetivo del estudio es conocer la relación entre este factor de riesgo y la hipoacusia, evaluándose características clínico-epidemiológicas y si existe variación genética no sindrómica relacionada.
Material y método: Estudio retrospectivo, transversal, descriptivo y observacional de los recién nacidos entre enero de 2007 y diciembre de 2010 con factor de riesgo de antecedente familiar de hipoacusia mediante otoemisiones acústicas provocadas transitorias y potenciales evocados auditivos de tronco cerebral.
Resultados: Nacieron 26.717 niños. Ochocientos cincuenta y siete (3,2%) tenían antecedente familiar. Cincuenta y siete (0,21%) no pasan segundas otoemisiones. Un 29,1% (n = 16) tenían otro antecedente de riesgo añadido. Un 17,8% (n = 9) tenían factor de riesgo no clásico. Ningún factor de riesgo tenía relación con la hipoacusia, excepto la cardiopatía. Según potenciales, el 76,4% tenían normoaudición y el 23,6%, hipoacusia. La media de familiares hipoacúsicos es de 1,25. En test genéticos el 82,86% son homocigosis normal, el 11,43% heterocigosis para mutación 35delG del gen de la Conexina 26, el 2,86% heterocigosis R143W del mismo gen y el 2,86% homocigosis mutante 35delG. No se encuentra relación entre hipoacusia y tener un alelo mutado.
Conclusiones: El porcentaje de niños con antecedente familiar diagnosticado de hipoacusia es superior a lo esperado en la población general. Es necesaria la actualización del perfil genético para esclarecer la relación encontrada entre hipoacusia con cardiopatía, el número de familiares afectos y la baja prevalencia en las mutaciones analizadas.

Introduction

Hearing is the natural way of acquiring language. Consequently, deafness is a serious impediment whose effects go far beyond the impossibility of speaking. Child hearing loss is a condition clearly differentiated from that of adult hearing loss because of everything it implies in social and intellectual development, and whose disabling and incapacitating potential can be resolved in terms of how early diagnosis is reached and appropriate treatment and rehabilitation are started.[1,2]

Hearing loss can arise from genetic (50%), environmental (25%) or unknown (25%) origins. Ones caused by heredity represent 20%–25% of the syndromic cases, while they are some 75% of the nonsyndromic cases. Within the latter, 50% are due to a single gene, the Connexin 26 gene.[3] Inherited hearing loss can be transmitted as autosomal dominant (10%–20%), autosomal recessive (70%–80%), X-linked inheritance (1%–5%) or inheritance through mutations in mitochondrial DNA (3%). More than 100 genes responsible for nonsyndromic hearing loss that codify products with highly varied functions have been identified.

Approximately 60% of hearing loss before the acquisition of language is of genetic origin and a great percentage of hearing loss in general probably has undemonstrated genetic influence in normal healthcare practice. The contribution of the *GJB2* gene (Connexin 26) has modified the assessment of children with hearing loss.[3] As a group, the mutations in the *GJB2*, *GJB3* and *GJB6* genes constitute the most frequent cause of nonsyndromic hereditary hearing loss in our milieu. The *GJB2* gene mutations are the most frequent cause of autosomal recessive nonsyndromic hearing loss.[4] More than 100 pathogenic variations of this gene have been identified, the most common of the recessive hearing loss cases (more than 80%) being the 35delG mutation.[3–5] About 1%–3% of the general population are carriers.[4]

The Joint Committee on Infant Hearing (JCIH) establishes criteria for risk of hearing loss validated by the Spanish Commission for Early Detection of Hearing Loss (Spanish acronym: *CODEPEH*). Before universal screening was instituted, auditory tests focused on children having these risk factors (RFs), given that the incidence of hearing loss was 40–50 times greater in this collective.[6] Family history of permanent hearing loss in infants (FHPHLI) falls among these RFs.

The objective of this study was to determine the relationship between the RF for FHPHLI and sensorineural hearing loss in the children born in the 2007–2010 period. The clinical-epidemiological characteristics of the children who went through a universal hearing loss screening program and those who failed it were analyzed, establishing the presence and degree of hearing loss, as well as whether any nonsyndromic genetic variation existed.