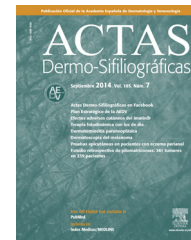




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ORIGINAL ARTICLE

Otologic Manifestations of Autosomal Recessive Congenital Ichthyosis in Children[☆]



A. Martín-Santiago,^{a,*} M. Rodríguez-Pascual,^b N. Knöpfel,^a Á. Hernández-Martín^c

^a Servicio de Dermatología, Hospital Universitario Son Espases, Palma de Mallorca, Spain

^b Servicio de Otorrinolaringología, Hospital Universitario Son Espases, Palma de Mallorca, Spain

^c Servicio de Dermatología, Hospital Infantil Universitario Niño Jesús, Madrid, Spain

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KEYWORDS

Ichthyosis;
Hearing loss;
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Abstract

Background: Few studies have investigated ear involvement in nonsyndromic autosomal recessive congenital ichthyosis (ARCI).

Objectives: To assess the type and frequency of otologic manifestations of ARCI in patients under follow-up at the pediatric dermatology department of our hospital.

Materials and methods: We prospectively studied the presence of ear pain, ear itching, tinnitus, otitis, cerumen impaction, accumulation of epithelial debris, and hearing loss. Daily hygiene measures, topical treatments, medical-surgical interventions, and frequency of visits to an ear, nose, and throat (ENT) specialist were noted in the patients' medical records. Ear examination and hearing tests were performed in all cases.

Results: Ten patients were studied: 2 had a self-healing collodion baby phenotype and 8 had ichthyosis. There was mention of otologic manifestations in the records of all 8 patients with ichthyosis (100%); 6 of these patients (75%) had abnormalities in the external auditory canal examination and 2 (25%) had conductive hearing loss. Our findings are limited by the small number of patients studied, all of whom were younger than 19 years.

Conclusions: The involvement of both dermatologists and ENT specialists in the management of patients with ichthyosis is crucial to ensure the application of the best therapeutic and preventive measures. More studies are needed to assess the prevalence and impact on quality of life of ear involvement in patients with ichthyosis and to determine the optimal interval between ENT visits for these patients.

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* Corresponding author.

E-mail address: anahsd@telefonica.net (A. Martín-Santiago).

PALABRAS CLAVE

Ictiosis;
Hipoacusia;
Sordera;
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Manifestaciones otológicas en los niños con ictiosis congénitas autosómicas recesivas**Resumen**

Introducción: Las complicaciones otológicas asociadas a las ictiosis congénitas autosómicas recesivas (ICAR) no sindrómicas, apenas han sido estudiadas en la literatura.

Objetivo: Conocer la frecuencia y el tipo de manifestaciones otológicas de los pacientes diagnosticados de ICAR, actualmente en seguimiento en la unidad de dermatología pediátrica de nuestro hospital.

Material y método: Se estudió de forma prospectiva la presencia de los siguientes parámetros: dolor, prurito ótico, acúfenos, otitis, tapón de cerumen, acúmulo de restos epiteliales y sordera. Se recogió en la anamnesis las medidas de higiene diaria, tratamientos tópicos o intervenciones médico-quirúrgicas requeridas y la periodicidad con la que los pacientes habían consultado a un especialista de otorrinolaringología (ORL). En todos los casos se realizaron otoscopia y pruebas auditivas.

Resultados: Se estudiaron 10 pacientes, 2 con fenotipo de bebé colodión autorresolutivo y 8 con ictiosis. Un 100% (8/8) de los pacientes con ictiosis referían algún síntoma o signo en la anamnesis, en el 75% (6/8) se observaron anomalías en la exploración del conducto auditivo externo y en el 25% (2/8) se objetivó sordera de conducción, que en un caso se consiguió revertir. Nuestro trabajo está limitado por el escaso número de pacientes, todos menores de 19 años.

Conclusiones: Es fundamental la participación conjunta del dermatólogo y del especialista de ORL en el manejo de los pacientes con ictiosis para establecer las mejores medidas terapéuticas y preventivas. Se precisan más estudios que determinen la frecuencia de la afectación otológica, su repercusión en la calidad de vida y la periodicidad mínima idónea de visitas al especialista de ORL.

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Introduction

Nonsyndromic autosomal recessive congenital ichthyosis (ARCI) is a rare hereditary keratinization disorder in which heat intolerance, pruritus, growth abnormalities, ocular and hearing disorders, and social communication deficits may also arise in addition to skin manifestations.^{1,2} The clinical course of ear complications has rarely been reported in the literature.^{3,4}

Objectives

The objective of this study was to determine the type of otologic manifestations present in patients diagnosed with ARCI and currently in follow-up in the Pediatric Dermatology Unit of the University Hospital Son Espases, Spain.

Methods

The presence of the following manifestations was studied prospectively: pain, ear pruritus, tinnitus, otitis, wax plug, accumulation of epithelial remnants, and deafness. The medical history included daily hygiene measures, topical treatments or medical-surgical interventions required, and frequency of visits to ear-nose-throat (ENT) specialists. All patients underwent otoscopy and hearing tests (pure tone audiometry [PTA], audiometry with visual reinforcement [AVR], and measurement of distortion products in the audiogram [DPgram]). The auditory results were calculated separately for each ear in decibels (dB). The binaural

loudness was quantified using pure tone average, which is used in children and recommended by the Bureau International d'Audiophonologie (BIAP). In children who, given their age, could not perform conventional audiometry, estimates were derived from objective tests (DPgram) and behavioral tests (AVR). In these cases, hearing was considered better than 30 dB (although the true hearing could be better) when functional hearing was observed.

Results

The same ENT specialist examined 10 patients, 8 boys and 2 girls, aged between 11 months and 18 years. The underlying molecular deficit was identified in both patients with self-healing collodion baby (*TGM1* and *ALOX12B*) but only in 2 out of 8 patients with skin manifestations of ichthyosis (*ALOXE3* and *TGM1*) (Tables 1 and 2). Detailed medical history and the examination performed by the ENT specialist did not reveal otologic symptoms or deafness in either of the 2 cases of self-healing collodion baby. Currently, both have only minimal dermatologic manifestations, although one did have tympanic membrane retraction. All of the remaining 8 children with generalized scaling had some otologic symptom: 8 out of 8 had a history of wax plugs, 6 out of 8 had accumulation of epithelial remnants, 3 out of 8 had ear pruritus, 3 out of 8 had occasional ear pain, 2 out of 8 had otitis, and 1 out of 8 had tinnitus. None of the 8 children used topical ear products daily, only 2 out of 8 cleaned the external auditory canal (EAC) with a cotton wool bud, and 2 out of 8 had been receiving systemic treatment with acitretin for several years. Examination of the EAC of these

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