ARCHIVOS DE LA SOCIEDAD ESPAÑOLA DE OFTALMOLOGÍA

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Original article

A curious fact: Photic sneeze reflex. Autosomical dominant compelling helio-ophthalmic outburst syndrome☆

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ARTICLE INFO

Article history:
Received 18 November 2015
Accepted 12 January 2016
Available online 23 April 2016

Keywords:
Photic reflex
Sneeze
Sunlight
Ocular
Dominant autosomal inheritance
Prominent corneal nerves

ABSTRACT

Objective: To assess ocular involvement in the pathophysiology of autosomal dominant compelling helio-ophthalmic outburst syndrome (ACHOOs).
Methods: An interview was conducted with a Caucasian family that showed clinical features of ACHOOs. Twelve of them had photic reflex and were recruited. A complete eye evaluation was made.
Results: A dominant autosomal inheritance with mild penetrance was demonstrated, with 67% of the studied subjects showing some degree of prominent corneal nerves. No other eye changes were found.
Conclusions: Prominent corneal nerves may be associated with ACHOOs. The other eye structures studied do not seem to play a role in ACHOOs. Further studies are needed to understand the physiology of the ACHOOs.

Curiosidad: reflejo de estornudo fótico. Síndrome helio-oftálmico de estornudos compulsivos autosómico dominante

Palabras clave:
Reflejo fótico
Luz solar
Estornudo
Ocular

RESUMEN

Objetivo: Evaluar la implicación ocular en la fisiopatología del síndrome helio-oftálmico de estornudos compulsivos autosómico dominante (ACHOOs).
Métodos: Una familia de raza caucásica, que muestra las características clínicas de ACHOOs, fue interrogada. De toda la familia, 12 pacientes presentan reflejo fótico y fueron seleccionados. Se realiza una evaluación oftalmológica completa.

☆ Please cite this article as: Sevillano C, Parafita-Fernández A, Rodriguez-Lopez V, Sampil M, Moraña N, Viso E, et al. Curiosidad: reflejo de estornudo fótico. Síndrome helio-oftálmico de estornudos compulsivos autosómico dominante. Arch Soc Esp Oftalmol. 2016;91:305–309.

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Herencia autosómica dominante
Nervios corneales prominentes

Resultados: Se encuentra una herencia autosómica dominante con penetrancia parcial. El 67% de los sujetos estudiados mostró algún grado de prominencia en los nervios corneales. No se encontraron otras alteraciones oculares.
Conclusiones: Los nervios corneales prominentes pueden tener asociación con el ACHOOs. Las otras estructuras del ojo estudiados no parecen desempeñar un papel en el ACHOOs. Se necesitan más estudios para comprender la fisiología del ACHOOs.

Introduction

Autosomal Dominant Compelling Helio-ophthalmic Outburst Syndrome (ACHOO) is very widespread, but few published reports discuss the pathophysiology of the syndrome.[1–17] The first reference is attributed to Aristotle.[18,24] It consists of a variable amount of sneezing in uncontrollable outbursts after sudden exposure to bright light, usually sunlight.[1–13,15,16] Inheritance is autosomal dominant,[1,2,8–13,18] with high penetrance, without preference for gender or iris colour,[8,11,12] but it is more common in Caucasians.[11,15] Prevalence ranges from 11% to 35%.[1,2,8–12,15] The aetiology is not fully understood but a number of theories have been suggested.[1,2,4,7,9,11–13,18]

Method

We studied a Spanish family that suffered from photic reflex, constructing the family tree to verify the number of patients affected and the pattern of transmission (Fig. 1). Like that published by Peroutka and Peroutka,[10] our case is that of the author's own family (C. Sevillano). All the patients (30) were questioned about their sneezing, and only 12 people showed clear signs of ACHOO. Two more patients showed partial symptoms compatible with ACHOO (not included in the study), and another had pathological sneezing with alcohol consumption. A complete eye examination was performed looking for alterations that might be related to the syndrome in some way. When assessing the corneal nerve plexus, the classification proposed by Takai et al.[14] was considered, where grade 0 is "invisible", grade 1 is "visible in slit-lamp but not in photograph ×25" and grade 2 is "visible in both cases" (Fig. 2).

The anterior segment images were taken with an ATE 600 DC1 camera (Topcon®), the optical coherence tomography scans with HD-OCT Cirrus (Carl Zeiss Meditec, Inc., Dublin, CA, USA), pachymetry measured with Pachette 2 model 500 SN-100096 (DHG Technology Inc., Exton, PA, USA) and pupillometry with the CA-100F topography corneal analyser (Topcon Medical Systems, Oakland, NJ, USA). All measurements were performed by the same person under the same circumstances (time, lighting conditions, eye drops, etc.).

Results

Twelve out of 30 members (40%) of the family clearly had the syndrome. The autosomal dominant inheritance pattern with partial penetrance was confirmed in the family tree (Fig. 1). The age range of the patients was broad (10–84 years), although onset of the syndrome is always before the age of 30. Of the patients in the sample, 76% had blue eyes and the ratio of male to female was 7:6. Four subjects (33%) had chronic sinusitis and only one of them also had a deviated septum. Two subjects had some type of allergy. One patient had Thygeson's superficial keratitis (Table 1).

The clinical features of the sneezes are shown in Table 2. The threshold[13] is a particular feature that consists of looking into sunlight when they feel the need to sneeze in order to trigger the sneeze; that is, providing the sneeze reflex with a minimal light stimulus to trigger the response; 75% of the subjects had this feature.

The frequency was graded as I (sometimes, <25%), II (often, 25–50%), III (nearly always, 50–75%), IV (always, >75%). In our study, 70% had grades III or IV.

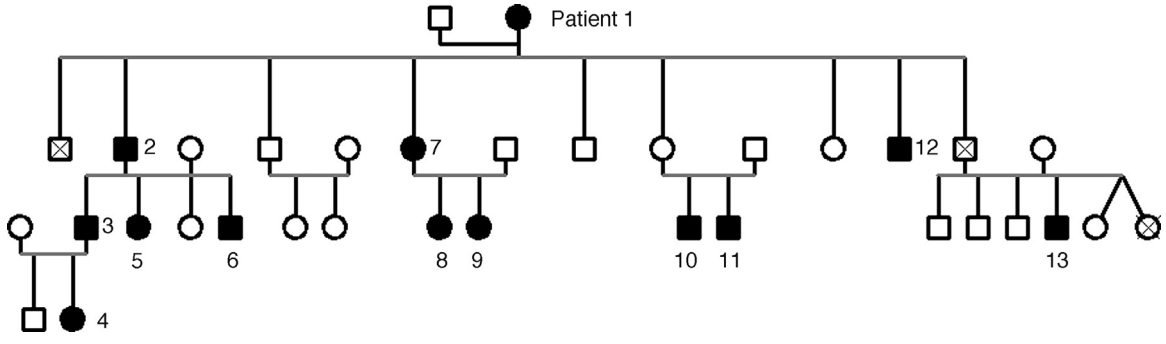

Fig. 1 – Family tree for the family studied.