Clinical Neurology and Neurosurgery 115 (2013) 1701–1708

Contents lists available at ScienceDirect

Clinical Neurology and Neurosurgery

journal homepage: www.elsevier.com/locate/clineuro

Neurophysiologic, audiometric and vestibular function tests in patients with hyperostosis cranialis interna

J.J. Waterval[a,b,*,1], M.P.H. Bischoff[a,1], R.J. Stokroos[a,b], L.J. Anteunis[a,b], D.M.W. Hilkman[c], H. Kingma[a,b], J.J. Manni[a]

[a] Department of Otorhinolaryngology – Head & Neck Surgery, Maastricht University Medical Center, P.O. 5800, 6202 AZ Maastricht, The Netherlands
[b] School for Mental Health and Neuroscience, University of Maastricht, Universiteitssingel 40, 6229 ER Maastricht, The Netherlands
[c] Department of Neurophysiology, Maastricht University Medical Center, P.O. 5800, 6202 AZ Maastricht, The Netherlands

ARTICLE INFO

Article history:
Received 2 September 2012
Received in revised form 18 March 2013
Accepted 25 March 2013
Available online 23 April 2013

Keywords:
Hyperostosis
Cranial nerve palsies
Facial nerve
Vestibulocochlear nerve
Trigeminal nerve
Hyperostosis cranialis interna
Leave hyperostosis

ABSTRACT

Objective: Hyperostosis cranialis interna (HCI) is an autosomal dominant sclerosing bone dysplasia affecting the skull base and the calvaria, characterized by cranial nerve deficits due to stenosis of neuroforamina. The aim of this study is to describe the value of several neurophysiological, audiometric and vestibular tests related to the clinical course of the disorder.
Methods: Ten affected subjects and 13 unaffected family members were recruited and tested with visual evoked potentials, masseter reflex, blink reflex, pure tone and speech audiometry, stapedial reflexes, otoacoustic emissions, brainstem evoked response audiometry and electronystagmography.
Results: Due to the symmetrical bilateral nature of this disease, the sensitivity of visual evoked potentials (VEPs), masseter reflex and blink reflex is decreased (25–37.5%), therefore reducing the value of single registration. Increased hearing thresholds and increased BERA latency times were found in 60–70%. The inter-peak latency I–V parameter in BERA has the ability to determine nerve encroachment reliably. 50% of the patients had vestibular abnormalities. No patient had disease-related absence of otoacoustic emissions, because the cochlea is not affected.
Conclusion: In patients with HCI and similar craniofacial sclerosing bone dysplasias we advise monitoring of vestibulocochlear nerve function with tone and speech audiometry, BERA and vestibular tests. VEPs are important to monitor optic nerve function in combination with radiological and ophthalmologic examination. We do not advise the routine use of blink and masseter reflex.

1. Introduction

Hyperostosis cranialis interna (HCI) is a hereditary bone dysplasia (Online Mendelian Inheritance in Man 144755) first described in 1990 [1] and is characterized by hyperostosis and osteosclerosis of the calvaria and skull base. This process leads to narrowing of various neuroforamina, causing cranial nerve entrapment and dysfunction of cranial nerves I, II, V, VII and VIII. Only one large Dutch family has been diagnosed with HCI to date, a pedigree is depicted in Fig. 1. HCI has an autosomal dominant transmission pattern.

To evaluate the function of the involved cranial nerves, patients underwent a series of function tests. The goal of this paper is to correlate symptomatology to various cranial nerve function tests. In this descriptive report, we will present an overview of the results of the tests of the affected individuals (see Fig. 2).

The test battery consisted of clinical neurophysiologic testing: visual evoked potentials (VEP); blink reflex and masseter reflex; audiometric testing: pure tone and speech audiometry, stapedial reflexes, otoacoustic emissions (OAE) and brainstem evoked response audiometry (BERA); and vestibular testing: electronystagmography (ENG).

Correspondence between objective cranial nerve tests and symptomatology or disease stage could be of prognostic value, could help for optimizing treatment planning and improve counseling.

2. Materials and methods

2.1. Subjects

Twenty-two family members were examined, 10 of which are affected. The same patients were examined as in the earlier study about the course of the disease and the symptomatology, except the

* Corresponding author at: Department of Otorhinolaryngology – Head & Neck Surgery, Maastricht University Medical Center, P.O. 5800, 6202 AZ Maastricht, The Netherlands. Tel.: +31 642250894; fax: +31 43 3875580.
E-mail address: J.Waterval@gmail.com (J.J. Waterval).
[1] Share first authorship.

0303-8467/$ – see front matter © 2013 Elsevier B.V. All rights reserved.
http://dx.doi.org/10.1016/j.clineuro.2013.03.008

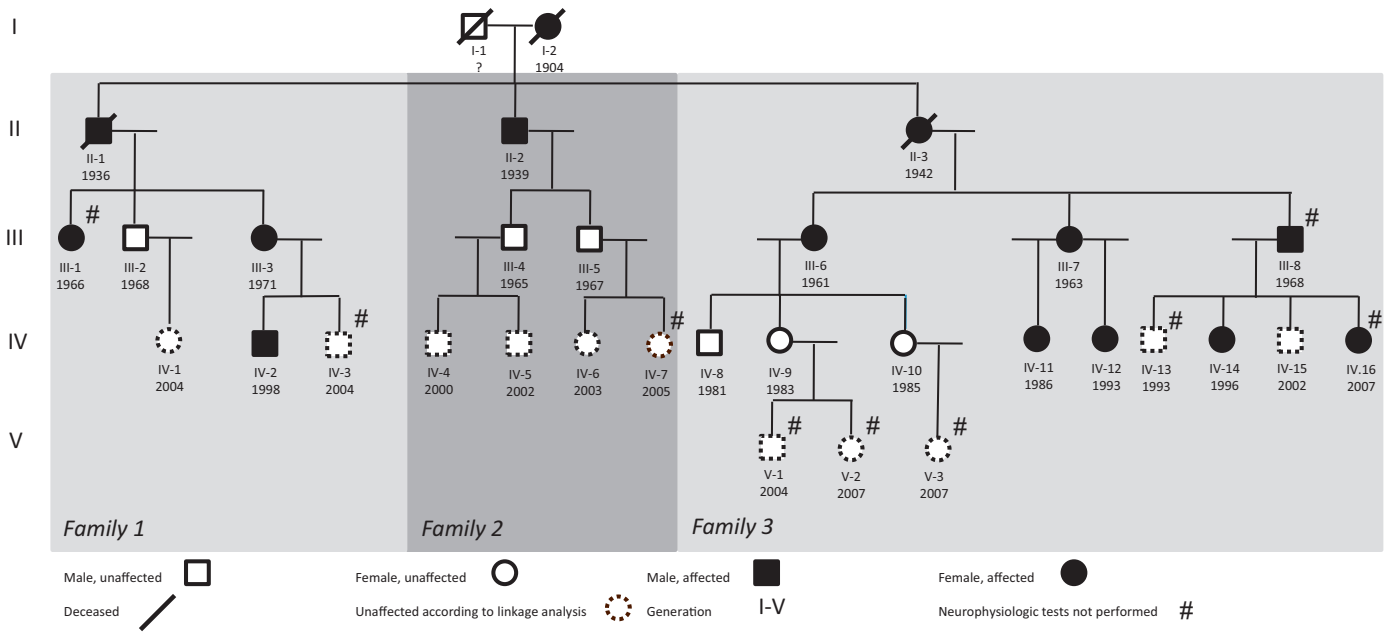

Fig. 1. Pedigree of the affected family.

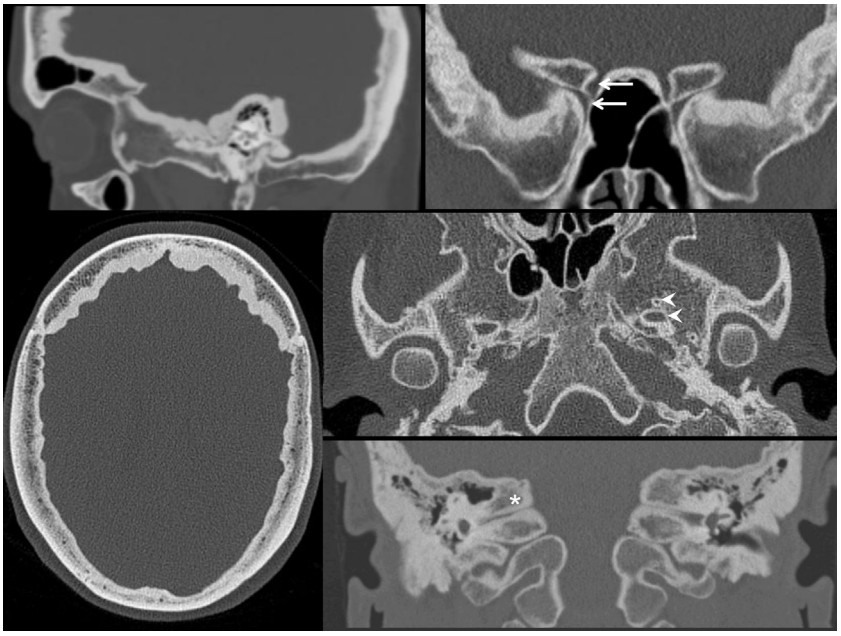

Fig. 2. Multiple CT images of patients affected with hyperostosis cranialis interna. Upper left: sagittal image of the skull base and calvaria at the level of the inner ear. An hyperostotic endosteal layer can be seen (female mid-thirties). Lower left: axial image of a markedly thickened skull. Upper right: coronal image (23-year-old female) of the greater and lesser sphenoid wings. Note narrowing of the optic canal (*upper arrow*) and the superior orbital fissure (*lower arrow*). Middle right: axial image showing the foramen rotundum (*upper arrowhead*) and the foramen ovale (*lower arrowhead*) in an 18-year-old female. Lower right: bilateral narrowing of the internal auditory canal in a 16-year-old patient (*asterisk*, coronal reconstruction).

individuals indicated with # in Fig. 1 [2]. The main reason was that the concerning children were considered to be too young. Function tests were performed in research setting. The individuals were aged 5–68 years at the time of examination. Written informed consent was obtained for all tested individuals.

2.2. Optic nerve

Optic nerve function was tested with pattern reversal VEP (ISCEV standard for clinical visual evoked potentials) [3,4], each eye stimulated individually. Seven electrodes were placed on the scalp to register potentials. Peak latencies N75, P100 and N145 were recorded. The P100 latencies were compared to laboratory reference values and classified accordingly as normal or as possible optic neuropathy and VEP outcome was correlated to clinical ophthalmologic examination.

2.3. Trigeminal and facial nerve

Trigeminal nerve function was assessed using both the masseter reflex and the blink reflex (afferent limb). To assess the masseter reflex, an active electrode was placed bilaterally on