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Inner ear anatomy in Waardenburg syndrome: Radiological assessment and comparison with normative data

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ABSTRACT

Objective: As patients with Waardenburg syndrome (WS) represent potential candidates for cochlear implantation, their inner ear anatomy is of high significance. There is an ongoing debate whether WS is related to any inner ear dysplasias. Our objective was to evaluate radiologically the inner ear anatomy in patients with WS and identify any temporal bone malformations.
Methods: A retrospective case review was carried out in a tertiary, referral center. The high resolution computed tomography (HRCT) scans of the temporal bone from 20 patients (40 ears) with WS who were managed for deafness in a tertiary referral center from 1995 to 2012 were retrospectively examined. Measurements of 15 different inner ear dimensions, involving the cochlea, the vestibule, the semicircular canals and the internal auditory meatus, as well as measurements of the vestibular aqueduct, were performed independently by two neuroradiologists. Finally, we compared the results from the WS group with a control group consisting of 50 normal hearing subjects (100 ears) and with previously reported normative values.
Results: Inner ear malformations were not found in any of the patients with WS. All measured inner ear dimensions were within the normative values compiled by our study group as well as by others.
Conclusions: Inner ear malformations are not characteristic for all types of WS; however, certain rare subtypes might be related to inner ear deformities. Normative cochleovestibular dimensions that can help in assessing the temporal bone anatomy are provided.

1. Introduction

Waardenburg syndrome (WS) is an autosomal dominant disease, characterised by dystopia canthorum, hyperplasia of the eyebrows, heterochromia iridis, white forelock and congenital sensorineural hearing loss [1,2]. Although WS was initially linked to genetic mutations in the gene PAX3, nowadays, six different genes, including MITF (microphthalmia-associated transcription factor), EDN3 (endothelin 3), EDNRB (endothelin receptor type B), SOX10 (encoding the Sry bOX10 transcription factor), and SNAI2 have been involved [1,2]. Based on the genetic background and the clinical characteristics, WS has been divided into four types. The characteristic feature for type I is dystopia canthorum, while in type II dystopia canthorum is missing. In type III patients additionally have upper limp anomalies and more coarse facial characteristics and, finally, type IV involves Hirschsprung disease [1–4]. Despite the differences between the four types, sensorineural hearing loss represents a common feature of all patients with WS [2–4]. Hearing loss may vary in severity; most patients with WS, however, suffer from profound sensorineural deafness and therefore are candidates for cochlear implantation [5–10].

As the inner ear anatomy is of high significance for cochlear implantation, imaging studies of the temporal bones are crucial for the preoperative assessment of patients with WS [11–13]. There is an ongoing debate whether WS is related to any anatomical inner ear anomalies. The results from previous studies are very heterogeneous with the incidence of temporal bone anomalies varying from 0 to 100% among patients with WS [9,14–16]. In particular, two studies focusing explicit on the temporal bone imaging and involving six and eight patients showed inner ear

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malformations in 50% and 17% of the cases, respectively [15,16]. However, the number of temporal bones evaluated in both studies was small. On the other hand, recent work on the outcome of cochlear implantation, involving 25 patients with WS did not note any inner ear anomalies, which could affect the implantation, without, however, having performed any detailed radiological measurements of the inner ear dimensions [5].

As sufficient evidence on the inner ear malformations in cases with WS is missing, our objective was to evaluate the temporal bone anatomy of such patients using appropriate imaging studies and measurements and comparing the obtained data with normative cochlear and vestibular dimensions.

2. Materials and methods

2.1. Study settings and patients

This retrospective study was carried out in a tertiary referral center. Approval from the Ethical Committee of the University was obtained.

Patients with WS who had been referred to our department from 1995 to 2012 for hearing loss assessment and management were identified through the electronic patients database and the coding system used by the medical staff. The notes of all patients with WS were found and retrospectively reviewed; the severity of hearing loss was documented.

2.2. HRCT settings

The CT scans were performed with three different types of scanners over the lengthy period covered by our study:

Two patients were examined using a HiSpeed Advantage RP CT scanner (GE, Milwaukee, WI, USA): helical CT examinations were performed at 140 kV and 80 mA s, with a section thickness of 1 mm and a pitch of 1.

Six patients were examined using a HiSpeed Advantage CT scanner (GE, Milwaukee, WI, USA): helical CT examinations were performed at 120 kV and 80 mA s, with a section thickness of 1 mm and a pitch of 1. The field of view was 16 cm using a 512 × 512 matrix.

Twelve patients were examined using a Light–Speed16 CT scanner (GE, Milwaukee, WI, USA): helical CT examinations were performed at 120 kV, auto mA s with a maximum up to 100 mA s, a speed of 5.62 s, a thickness of 0.625 mm, and a pitch of 0.562:1.

The CT scans were uploaded directly to our current picture archiving and communications system (PACS) (GE, Milwaukee, WI, USA): except for the eight patients in whom only the plain films were available. These were scanned in order to perform the measurements at the workstation. Reconstructions in the coronal plane were available in 12 cases uploaded primarily to the PACS.

2.3. Measurements

All measurements were performed using PACS and the electronic calipers and were taken in millimeters. A modified measurement system of the ones mainly proposed by Purcell et al. [17] as well as by Krombach et al. [18] was employed. In particular, 15 different inner ear dimensions were measured on the coronal and axial plane. These dimensions involved:

- Vestibule (axial): width and height (Fig. 1).
- Cochlea (axial): length of basal turn, width of the basal turn lumen, length of the apical turn and height of the apical turn (Fig. 2).
- Cochlea (coronal): height of the cochlea (Fig. 2).
- Semicircular canals: the width of the superior and the posterior semicircular canals, the width of the bony island (Fig. 3) and the ampulla (Fig. 4) of all three semicircular canals. Canal lumen and bony island measured at maximum diameter of the turn, most often two sections below the first view of the superior semicircular canal. Ampulla identified at sections between canal lumen and vestibule, most often one section below the section used for the lumen measurements. Regarding the posterior semicircular canal, it was measured one to two sections above the inferior limb.
- Internal auditory meatus: length was measured at the longest section and width close to the cochlear aperture (Fig. 5).

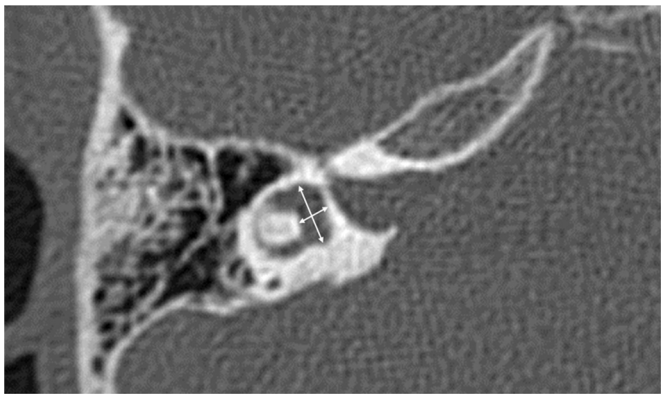

Fig. 1. Measurements of the vestibule in axial HRCT scans.

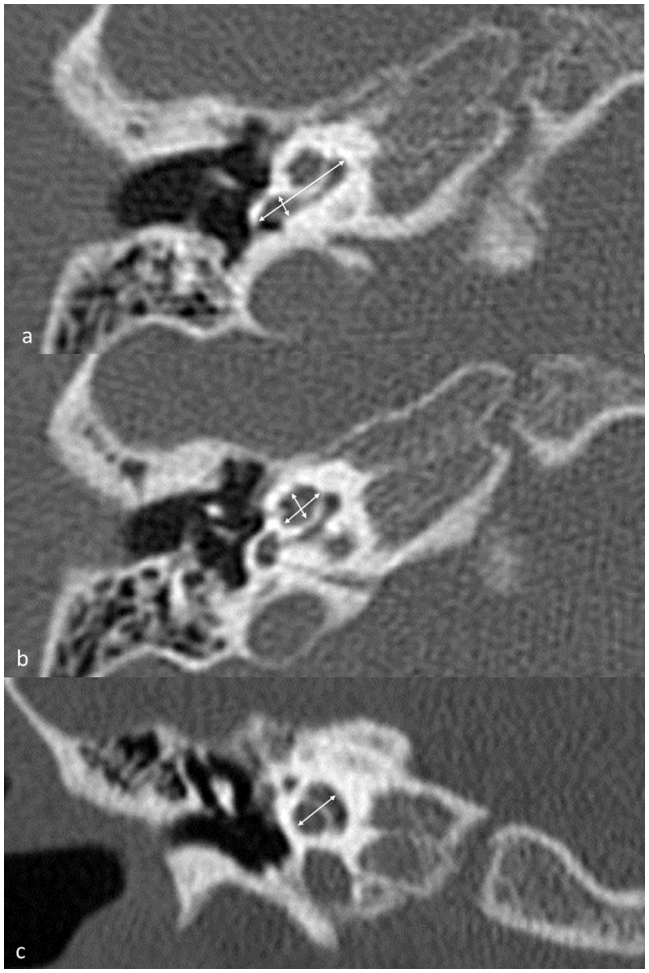

Fig. 2. Measurements of the cochlea on the axial plane: length of basal turn (a), width of the basal turn lumen (a), length of the apical turn (b) and height of the apical turn (b) and on the coronal plane (height of the cochlea, (c).