CASE STUDY

Mondini Deformity in a Case of Turner Syndrome. A Radiological Finding☆

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KEYWORDS
Mondini deformity; Turner syndrome; Cochlear malformation

Abstract Turner syndrome (TS) is the human being’s most frequent sex chromosome abnormality. Progressive sensorineural hearing loss is documented in more than 50% of the women affected by this syndrome. Although Mondini defect is the cochlear congenital malformation most frequently identified in other polyomalformative syndromes, it has rarely been reported in TS. We describe the case of a 32-year-old woman with TS who presented progressive sensorineural hearing loss. The computed tomography of the ears showed bilateral Mondini deformity. © 2010 Elsevier España, S.L. All rights reserved.

PALABRAS CLAVE
Deformidad de Mondini; Síndrome de Turner; Malformación coclear

Deformidad de Mondini en un caso de síndrome de Turner. Un hallazgo radiológico

Resumen El síndrome de Turner (ST) es la anomalía cromosómica sexual más frecuente en seres humanos y en más de la mitad de las mujeres afectadas por este síndrome se ha documentado hipoacusia neurosensorial progresiva. Aunque la deformidad de Mondini es la malformación coclear más frecuentemente identificada en otros síndromes polimalformativos, raramente se ha descrito en el síndrome de Turner. Presentamos el caso de una mujer de 32 años con ST que consultó por hipoacusia neurosensorial bilateral progresiva. La tomografía computarizada de oídos demostró una deformidad de Mondini bilateral. © 2010 Elsevier España, S.L. Todos los derechos reservados.

Introduction

In 1938, Henry H. Turner first described the main physical characteristics of Turner syndrome (TS), which are female phenotype, short stature, gonadal dysgenesis and infertility, among other physical abnormalities and alterations in multiple organs.1 TS is currently the most common sex chromosome abnormality in humans. Its incidence is estimated at 1:2000 to 1:5000 of live births with female phenotype, and it represents up to 15% of all spontaneous abortions.2 Its classical and most common karyotype is X monosomy (45, XO), which represents the complete loss of the maternal X chromosome, although when the deficit of this chromosome is partial we refer to mosaicism.3

Otolological and hearing problems in TS are well documented. From 1969 to date, there is abundant literature demonstrating a high incidence of otological and hearing

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diseases in TS. Progressive sensorineural hearing loss (SNHL) has been described in up to 58% of women with TS, but the origin of this hearing loss is still unknown.

Although cochleovestibular malformations, especially Mondini deformity, are a relatively frequent finding in other polyomalformation syndromes, they have rarely been reported in TS.

In this work we present the case of a 32-year-old female patient affected by TS who attended consultation due to progressive hearing loss. A computed tomography (CT) of the ears revealed the presence of bilateral Mondini deformity. This is the third reported case describing a Mondini defect in TS and the first in which a diagnosis has been obtained radiologically.

Case Report

We present the case of a 32-year-old female patient who attended consultation due to progressive hearing loss. She suffered TS with 46,Xi (Xq) mosaicism, diagnosed at 5 years of age by a cytogenetic study. She had also undergone placement of transstympanic ventilation tubes at 2 years of age, due to repeated otitis media.

She provided a tone audiometry conducted 5 years earlier, which revealed bilateral SNHL with a right deficit of 25% and a left deficit of 30% (percentages calculated by the Fowler-Sabine method, as proposed by the American Medical Association [AMA]).

Otoscopy was normal, as was the rest of the ENT examination. We conducted an audiometry which confirmed the existence of bilateral, symmetrical SNHL, with a U-shaped audiometric profile, which mainly involved midrange frequencies. We also observed a 45% loss in the right ear and a 41% loss in the left ear (percentages calculated as recommended by the AMA).

The early onset of hearing loss and its rapid progression led us to perform a CT imaging study of the ears. The images obtained revealed the presence of a bilateral cochleovestibular malformation. We observed incomplete partition of the cochlea, with modiolus present only in the basal turn and coalescence of the middle and apical turns, as well as dilatation of the vestibule and semicircular canals (Figs. 1–3). These observations were consistent with Mondini deformity, according to the classification of cochleovestibular malformations proposed by Jackler et al. in 1987.

The patient was proposed the adaptation of a bilateral hearing aid and periodic audiometric monitoring as a treatment for hearing loss.

Discussion

As described in the introduction, the incidence of SNHL has been estimated at over half of patients affected by TS. However, this hearing loss can be manifested in different forms according to the karyotype. For Hultcrantz et al., hearing impairment occurs at an earlier age and progresses more rapidly in patients who present 45,X and 46,Xi (Xq) mosaisms. This idea matches the audiometric findings observed in our patient, who presented the 46,Xi (Xq) type.

Figure 1  Axial CT image of the left inner ear showing an incomplete partition of the cochlea, with modiolus present only in the basal turn and coalescence of the middle and apical turns. Co: cochlea.

The medical literature contains the following hypotheses which attempt to explain the origin of this hearing loss: involvement of the inner ear secondary to middle ear inflammatory disease, negative effects on inner ear maturation caused by a lack of oestrogen, a theoretical influence on the inner ear exerted by the final expression of a single X chromosome from the father and the rarely described presence of Mondini deformity in TS. However, none of these hypotheses has proven definitive and the true origin of this SNHL remains unknown. Hultcrantz et al. speculate that there may be more than one cause, thus justifying the presence of different SNHL patterns in TS.

Cochleovestibular malformations, especially Mondini deformity, are a relatively common finding in other polyomalformation syndromes, such as Pendred, Alport or Down syndromes, but have rarely been described in TS.

To date, PubMed only contains two references reporting the presence of Mondini deformity in TS. The first,
published by Windle-Taylor et al. in 1982, described the finding of Mondini deformity during the autopsy of a 47-year-old female suffering TS. The second reference, published by Fish et al. in 2009, reported a new case of Mondini deformity in an autopsy series of nine foetuses also affected by TS. In both these works, Mondini defect was a histopathological finding obtained while performing autopsies on studied subjects.

Our work presents the third reported case associating Mondini deformity with TS. Our main contribution is that this is the first case in which the diagnosis was obtained from a living patient by radiological study.

Figure 3 3D reconstruction of a CT image of the left inner ear showing an incomplete partition of the cochlea, accompanied by dilated vestibule and semicircular canals. These findings are compatible with Mondini deformity. CAI: internal auditory canal; Co: cochlea, CSH: horizontal semicircular canal; CSP: posterior semicircular canal; CSS superior semicircular canal; Ve: vestibule.

References