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OTOLOGY

Hearing disorders in Turner's syndrome: a survey from Iran

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Abstract Turner syndrome (TS) is one of the most frequently encountered sex-linked chromosomal abnormalities, occurring in one per 2,000 female births. These patients present with short stature and failure to begin puberty. In this syndrome, there are multiple organ abnormalities, including auditory disorders. TS patients were referred to the ENT clinic by a pediatric endocrinologist. A questionnaire was filled out and the patients went through a complete otologic examination. They were then referred to the audiology clinic to undergo audiologic test battery plus high-frequency pure tone audiometry. From a total of 48 ears examined, 11 (22.9 %) had a normal audiometry. Mid-frequency sensorineural hearing loss (SNHL), high-frequency SNHL, combined and mixed hearing loss were diagnosed in 6 (12/5 %), 20 (41/7 %), 6 (12/5 %) and 1 (2/1 %) ear, respectively. Tympanogram results showed normal compliance (A, As, Ad) in the majority of cases. B and C patterns were found in a few cases. Speech discrimination score was normal in all patients whereas speech reception threshold was normal in 92 % of the ears. Audiometry abnormality especially SNHL is common in TS patients, with the high-frequency pattern being the most frequent.

Keywords Ear · Turner syndrome · Hearing loss · Auditory disorders

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Introduction

Turner syndrome is a sex-linked chromosomal abnormality which involves 1 out of 2,000 live female births [1]. It was initially diagnosed in 1938 by an American physician named Henry H. Turner and described by two main findings including a short stature and absent secondary sexual characteristics due to ovarian failure resulting from gonadal dysgenesis, while hearing disorders were overlooked [2]. This syndrome is caused by the absence of two complete copies of the X chromosome in some or all the cells. The affected cells may have only one X (monosomy) (45,X) or they may be classified in one of the several types of partial monosomy like a short (p) arm deletion of one X chromosome (46,XdelXp) or the presence of an isochromosome with two q arms (46XiXq). In the case of mosaicism, cells with X monosomy (45, XO) may occur along with cells that are normal (46,XX), cells that have partial monosomies, or cells that have a Y chromosome (46,XY) [3].

Recent reports have suggested a significant incidence of sensorineural hearing loss in children with Turner syndrome. Yet, the incidence, diversity and even analysis of such otological signs and symptoms in patients with this syndrome have been difficult to evaluate because of relatively small numbers of reported surveys, different criteria used as hearing loss threshold and its amount and the overlap between conductive and sensorineural hearing loss

in some ear infections [4]. Anyhow, these patients may suffer from conductive, sensorineural and mixed hearing loss [5]. Conductive hearing loss usually occurs as a result of recurrent middle ear infections which are common during the childhood of such kids [1]. In the available literature the rate of conductive hearing loss has ranged from 6 to 44 % [6]. Although higher rates reaching up to 58 % of middle ear infections requiring treatment were also reported in some studies [7]. The prevalence of sensorineural hearing loss has been reported as high as 9–83 % in other studies [6] and has two common patterns of mid (dip) and high-frequency loss. The dip type begins at early age and most probably has a genetic basis; it is also a strong predictor of progressive hearing loss in the future [8]. High-frequency hearing loss is most commonly seen in older women [9] and is almost equal to presbycusis, a disorder which is seen in a younger age in the general population resulting from estrogen insufficiency [8].

The simultaneous existence of these two types of hearing loss has been also diagnosed (the mixed pattern). Hultcranz and Sylven [10] believe that with aging, high-frequency hearing loss is added to the dip pattern which results in serious hearing problems in Turner cases in comparison to their peers.

Regarding the prevalence of hearing disorders in Turner patients, no similar study having been conducted in the Middle East so far, and considering the wide variety between different communities in terms of environmental, socioeconomic, climate and health aspects that affect the hearing disorders in such patients, we aimed at studying these disorders in Turner patients and reporting their prevalence in our region. Speech reception thresholds and speech discrimination score were also measured to evaluate the communication disabilities among Turner Syndrome patients. The small sample size hampered the karyotype and age specific categorization in this survey.

Materials and methods

This cross-sectional study was performed from summer 2010 to autumn 2012 on 25 patients with Turner syndrome.

All patients had been referred by a pediatric endocrinologist to the ENT clinic of Qaem Educational Hospital, Mashhad, Iran. The patients were fully examined by an otolaryngologist, microscopically. In addition, the patients were referred to an audiologist to undergo high-frequency pure tone audiometry and tympanometry as well as speech discrimination score and speech recognition threshold. Normal pure tone audiometry (PTA) or speech reception threshold (SRT) level is defined as PTA or SRT level $\leq$25 dB HL.

In the case of any abnormalities, the appropriate treatment was recommended and initiated accordingly. Patients with a history of trauma, otologic surgery or ototoxic drug consumption were excluded from the study. An informed consent was obtained from each patient or their guardian prior to study entrance.

The collected data were analyzed by the SPSS software version 11.5. Independent t test was used for data analyses and a $P \leq 0.05$ was considered as statistically significant.

Results

Our studied subjects were aged from 4 to 37 years (mean age 14.6 $\pm$ 6.93 years). Among the 25 cases, 22 karyotypes were available from which 12 (55 %), 8 (36 %) and 2 (9 %) had 45xo, isochromosome or mosaicism karyotype, respectively.

Some audiologic tests could not be performed in one patient due to very young age. In total, among the 48 studied ears, 11 (22.9 %) had normal hearing; 6 (12.5 %) and 20 (41.7 %) had mid- and high-frequency sensorineural hearing loss, respectively, whereas 4 (8.3 %) had a combination of high- and mid-frequency hearing loss. Conductive and mixed hearing loss were detected in 6 (12.5 %) and 1 (2.1 %) case, respectively.

Speech discrimination score (SDS) values were between 92 and 100 %. Twenty-three (92 %) patients had a normal SRT in both ears whereas it was out of normal range in 2 (8 %). In total, type A tympanogram was found in 84 % (21 cases) and 88 % (22 cases) of right and left ears, respectively. While these figures were 16 and 12 % for the type B in a same order.

The frequency of different types of hearing loss in the studied subjects is shown in Table 1.

In this study no meaningful relationship was found between the right and left ear tympanogram with age (P value in the right ear = 0.148 and in the left ear = 0.343).

Table 1 Frequency of different types of hearing loss in the studied cases

Type of hearing loss	Frequency (%)
Normal	11 (22.9)
Mid-frequency SNHL	6 (12.5)
High-frequency SNHL	20 (41.7)
CHL	6 (12.5)
Mixed	1 (2.1)
Combined SNHL	4 (8.3)
Total	48 (100)

Discussion

Turner syndrome is an X-linked chromosomal abnormality which results in a short stature and abnormal expression of secondary sexual characteristics. Moreover, a wide range of various organ disorders such as heart, kidney, endocrine system and especially otologic disorders are developed [1].

Hearing disorders were first described by Anderson in Turner's patients [7]. From then the psychosocial impact of this hearing impairment has increasingly become the issue of concern. This is even more important in sensorineural hearing loss as the presence of a mid-frequency dip is specifically a strong predictor of future hearing status deterioration with subsequent social consequences [8].

Regarding the possible role of racial and environmental factors in the incidence of various disorders and as no such study has been conducted on hearing disorders in Turner patients in the Middle East, we aimed at investigating its prevalence in Iran.

The level of normal hearing in our study was 22.9 % which has ranged from 17 to 71.9 % in other studies [4, 6, 8, 11].

High-frequency sensorineural hearing loss was found in 41.7 % of our patients in agreement with the study by Chan in 2012 [6] and Beckman in 2004 [4]. In 1994, Hultcranz et al. [10] reported that this type of hearing loss is added to the dip type in older ages. However, Hederstierna et al. [8] believed that it is actually presbycusis which has occurred early in Turner patients.

The mid-frequency pattern of sensorineural hearing loss (dip) was reported as 12.5 % in the present study which has ranged between 4 and 68 % in similar researches [12, 13].

In Stenberg study, the latter pattern (dip) was the dominant pattern of hearing loss in the studied patients [13]. Hultcranz and Hederstierna in 2009 [8] stated that this kind of hearing loss as a genetic origin and is a strong predictor of rapidly progressive hearing loss in the near future. This type of hearing loss usually manifests at an early age which justifies the high rate of this kind of hearing loss in Stenberg's study [13].

The simultaneous existence of both types of hearing loss in high and mid frequency (combined) was seen in 8.3 % of the cases which is in consistent with Makishima and Barrenas studies [4, 14]. In Makishima study among the 57 cases with sensorineural hearing loss, 60 % had the high- and mid-frequency pattern [14].

Conductive hearing loss was diagnosed in 12.5 % which is similar to the result reported by Chan in 2012 as 11 % [6]. This type of hearing loss is due to recurrent infections in the middle ear and the resulting sequel in the middle ear and the tympanic membrane. Regarding the high rate of infections in the early ages, this type of hearing loss usually develops at childhood, as in Verver (age range 1.7–21.2 years) [12], Stenberg (age range 4–15 years) [13] and Watkin (mean age 10.5 years) [15] studies, the reported rate of conductive hearing loss was high. However, in Han study which was conducted on Turner adult cases (age >19 years), this type of hearing loss was diagnosed in 18 % of the patients [16]. Another explanation might be the environmental effect, as we can see most of the aforementioned studies with a high-prevalence rate were from Nordic or high altitude countries. Although the climate changes may play an important role in this field, we should not overlook the socio-economic aspect of the Middle Eastern developing countries and its effect on middle ear infections and its complications.

The mixed pattern of hearing loss was found in only one (2.1 %) patient. The same pattern was also reported in Verver and Dhooge study but with a lower rate [11, 12].

In tympanometric studies 84 and 88 % were reported as type A in the right and left ear, respectively. The same pattern was dominant in other studies as well [11, 17].

There are few reported studies that measure speech discrimination score. In our study the SDS ranges between 92 and 100 %. Of note, this percentage was obtained in a silent environment. The report by Hederstierna et al. [8] involved speech comprehension measurement in noise, a more difficult task for hearing-impaired people.

Limitations

Lack of sufficient data to relate the patients' age and karyotype to the type and amount of hearing loss was an important limitation of this study.

Conclusion

This study shows that audiometric abnormalities are frequent in Turner syndrome patients, though not significant. It seems that hearing and audiometric investigations are important and Turner syndrome patients should be visited by otolaryngologist and audiologist as complimentary physical examination.

Considering the difference with similar studies performed in other parts of the world, the role of racial and environmental factors is further verified in Turner syndrome patients. Careful follow-up during early childhood of children with Turner syndrome is necessary to detect middle ear disease and prevent sequel.

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Conflict of interest None to declare.

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