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Original article

Otologic disorders in Turner syndrome

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ABSTRACT

Introduction: Patients with Turner syndrome (TS) have craniofacial malformations, such as Eustachian tube hypoplasia and dysfunction and velar dysfunction, which foster acute otitis media. The aim of this study was to inventory pediatric otologic disorders in patients with TS at their first ENT consultation in our center.
Patients and methods: We reviewed the ENT consultation data of pediatric TS patients followed in our center between 2005 and 2015: otoscopy, hearing threshold, and history of acute otitis media or ENT surgery. Data were compared according to karyotype: X monosomy (45,X), mosaic (45,X/46,XX), isochromosome (46,Xi [Xq]), X ring chromosome X (XrX), with Y material, and "other".
Results: Ninety patients, with mean age 11.9 years (± 4.8 years) at first ENT consultation, were included: 29% showed tympanic abnormality on otoscopy, 21% had hearing loss, 24% had history of recurrent acute otitis media; 18% had undergone adenoidectomy, 24% T-tube insertion, and 5.6% tympanoplasty. No particular karyotype was associated with higher risk of hearing loss or acute otitis media.
Conclusion: Patients with TS showed high prevalence of pediatric otologic disorders; they therefore require close and prolonged ENT follow-up.

1. Introduction

Turner syndrome (TS) is a rare chromosome pathology with prevalence of 1/2500 female neonates, involving total or partial loss of an X-chromosome. The most frequent karyotype is 45,X monosomy (40–50%), but others are also reported, and notably mosaic karyotypes (30–40%); X-chromosome structure abnormality, such as duplication (isochromosome Xp or Xq), deletion or X ring chromosome, are rarer [1].

The phenotype almost systematically comprises retarded growth and ovarian insufficiency and, less systematically, other particularities of variable severity and increased risk of congenital cardiac and renal deformity and acquired pathology: cardiovascular, ENT, metabolic, autoimmune, etc.

In the ENT sphere, TS is associated with several abnormalities: in the outer ear, low-set ears, cupped auricles, narrow external auditory canal, or protruding ears [2]; and in the inner ear, Mondini dysplasia or maturation disorder related to estrogen deficiency [3]. Pterygium colli is also found in case of cervicofacial dysmorphia.

The literature reports that girls with TS frequently suffer from recurrent acute otitis media between the ages of 1 and 6 years, with a peak around 3 years [2]. This is exacerbated by hypoplasia and dysfunction of the soft palate and Eustachian tube and by shortened skull base [4], and contributes to the conductive or mixed hearing loss frequently found in TS adults [5].

Current French health authority guidelines (HAS 2008) recommend regular ENT follow-up and care, at a rhythm corresponding to the disorders diagnosed.

The aim of the present study was to report the pediatric specificities found at first ENT consultation in our center in TS patients, and to analyze them by karyotype, so as best to screen for and prevent ENT sequelae in adulthood.

2. Patients and methods

A retrospective study included patients followed for TS in the pediatric ENT department of the Robert Debré Hospital (Paris, France), between January 2005 and June 2015. Anonymized data were retrieved from medical files: admission and consultation reports and complementary examination results. All TS patients were referred for ENT consultation as part of the initial workup. A dedicated TS consultation form that has been used in the

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department for several years facilitated retrospective data harvesting. Patient age in the present study was that at this first ENT consultation.

ENT disorders comprised: tympanic abnormality on otoscopy (serous otitis media, myringosclerosis, retraction pocket, etc.), history of ENT surgery (adenoidectomy, T-tube insertion, tympanoplasty), hearing loss (on air- and bone-conduction audiogram) and recurrent acute otitis media (classically defined as ≥ 5 episodes per year). Hearing loss was defined as air-conduction hearing threshold ≥ 25 dB (averaging thresholds at 500, 1000, 2000 and 4000 Hz) in at least one ear, and was classified as conductive (or mixed) for Rinne > 15 dB.

Six karyotype subgroups were distinguished: monosomy X (45,X), mosaic (45,X/46,XX), isochromosome (46,Xi [Xq]), X ring chromosome (XrX), containing Y material, and "other".

Statistical analysis was performed with alpha risk = 0.05, on chi^2 test for qualitative variables.

The study had local institutional review board approval.

3. Results

3.1. General cohort characteristics

Ninety TS patients were included, with mean age 11.9 ± 4.8 years at first ENT consultation. Diagnosis was antenatal in 18 cases, at birth in 7 and postnatal in 65. Mean age at diagnosis was 7.8 ± 4.9 years, excluding antenatal diagnoses and counting birth as 0 years. The difference between age at diagnosis and at consultation was due to certain patients having been initially followed up elsewhere. All patients with diagnosis of TS were seen in ENT consultation in our center.

3.1.1. Data at first ENT consultation (Table 1)

Twenty-two patients (24%) presented with recurrent acute otitis media.

Twenty-six (29%) showed tympanic abnormality on otoscopy: unilateral (n = 4) or bilateral (n = 10) serous aspect, unilateral (n = 3) or bilateral (n = 5) tympanic retraction, or unilateral (n = 2) or bilateral (n = 2) myringosclerosis; 3 had T-tubes fitted, with normal tympanic membrane.

Nineteen (21%) had hearing loss: 10 pure conductive (9 bilateral, 1 unilateral), with mean threshold 39 dB (±9.2 dB); 3 bilateral sensorineural, with mean threshold 35 dB (±5.7 dB); and 6 mixed (3 bilateral, 3 unilateral), with mean threshold 40 dB (±9.8 dB).

Four patients with conductive hearing loss had associated serous otitis.

Sixteen (18%) had had adenoidectomy; 22 (24%) had had T-tube insertion (including one with several T-tubes); 5 (5.6%) had had tympanoplasty, including 4 for cholesteatoma, between the ages of 5 and 15 years.

3.1.2. Data from first ENT consultation according to karyotype (Table 1)

Twenty-nine patients (32%) had 45,X karyotype. Ten of these (34%) showed abnormality on otoscopy, 8 (27%) had hearing loss (6 conductive, 2 mixed), 8 (28%) had recurrent acute otitis media, 7 (24%) had had adenoidectomy, 10 (34%) had T-tube insertion (including 4 multiple) and 3 (10%) had had tympanoplasty for cholesteatoma.

Eleven patients had 45,X/46,XX karyotype. Two of these (18%), none had hearing loss, 3 had recurrent acute otitis media, 2 had had adenoidectomy and multiple T-tube insertion, and none had had tympanoplasty.

Six patients had 46,Xi (Xq) karyotype. Two of these (33%) showed abnormality on otoscopy, 2 had hearing loss (1 mixed, 1 sensorineural), none had recurrent acute otitis media, 1 had a T-tube insertion, and 1 had had tympanoplasty for uncontrollable retraction.

Eleven patients had XrX karyotype. Three of these (27%) showed abnormality on otoscopy, 3 had hearing loss (1 sensorineural, 2 mixed), 3 had recurrent acute otitis media, 2 had had adenoidectomy, 3 had a T-tube insertion: (1 multiple), and had had tympanoplasty for cholesteatoma.

Five patients had a karyotype containing Y chromosome. One of these showed tympanic abnormality, none had hearing loss or history of otitis, and 1 had had adenoidectomy.

Twenty-eight patients had "other" karyotypes. Eight of these (29%) showed abnormality on otoscopy, 6 (21%) had hearing loss (4 conductive, 1 sensorineural and 1 mixed), 8 (29%) had recurrent acute otitis media, 4 (14%) had had adenoidectomy, 6 (21%) had T-tubes (4 multiple), and none had had tympanoplasty for cholesteatoma.

Table 1 shows first consultation data.

The most frequent karyotype was 45,X, and it was in this subgroup that otologic disorders were most common. We therefore compared the 45,X subgroup versus the other subgroups taken together, but found no significant differences on chi^2 test for frequency of abnormality on otoscopy, adenoidectomy, T-tube, tympanoplasty, hearing loss of all types or recurrent acute otitis media.

4. Discussion

The present study showed increased risk of inner ear pathology in girls with Turner syndrome. Karyotype analysis did not

Table 1
Otologic and audiologic characteristics of karyotype subgroups at first ENT consultation.

Characteristics n (%)	45,X 29 (32%)	45,X/46,XX 11 (12%)	46,Xi (Xq) 6 (7%)	XrX 11 (12%)	Y 5 (6%)	Other 28 (31%)	Whole cohort n = 90
Age (yrs) at diagnosis	6.6	8.1	6.6	6.3	8.0	9.7	7.8
Age (yrs) at first ENT consultation	11.6	12.4	13.5	10.8	11.0	12.2	11.9
Abnormality on otoscopy	10 (34.4%)	2 (18.2%)	2 (33.3%)	3 (27.3%)	1 (20.0%)	8 (28.6%)	26 (28.9%)
Adenoidectomy	7 (24.1%)	2 (18.2%)	0	2 (18.2%)	1 (20.0%)	4 (14.3%)	16 (17.8%)
T-tube	10 (34.5%)	2 (18.2%)	1 (16.7%)	3 (27.3%)	0	6 (21.4%)	22 (24.4%)
Single	6	0	1	2	0	2	11 (12.2%)
Several	4	2	0	1	0	4	11 (12.2%)
Tympanoplasty	3 (10.3%)	0	1 (16.7%)	1 (14.3%)	0	0	5 (5.6%)
Hearing loss	8 (27.6%)	0	2 (33.3%)	3 (30.0%)	0	6 (21.4%)	19 (21.1%)
Conductive Sensorineural	6	0	0	0	0	4	10
Mixed	0	0	1	1	0	1	3
	2	0	1	2	0	1	6
Recurrent AOM	8 (27.6%)	3 (27.3%)	0	3 (27.3%)	0	8 (28.6%)	22 (24.4%)

AOM: acute otitis media.

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