



Phenotypic characterization and risk factors for microtia in East China, a case–control study



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ABSTRACT

Objective: Although congenital microtia has been reported in various studies, little is known about the etiology of isolated and sporadic cases. The aim was to analyze potential risk factors for isolated and sporadic microtia using case–control study in East China.

Methods: The study analyzed data from the hospital-based recruitment for deliveries between 2007 and 2013. Nine hundred eleven patients with microtia enrolled in the phenotypic characterization analysis, and then were adjusted by sex, age, region, syndrome and family history to compare with 562 random normal controls for potential risk factors.

Results: Microtia is observed more often in males (69.7%), and the cases were typically unilateral (74.0%), right-sided (57.2%), sporadic (92.0%) and isolated (69.5%). Mothers of children with microtia were more likely to have suffered a periconceptional cold-like syndrome as well as to have had a history of previous spontaneous abortion. Inflammatory infection (aOR, 3.56; 95% CI, 2.07–6.13) and chemical exposure (aOR, 2.77; 95% CI, 1.78–4.32) was associated with a higher risk of microtia. However, threatened abortion was not the risk factor (aOR, 1.14; 95% CI, 0.78–1.67), using progesterone may increase the risk (aOR, 1.92; 95% CI, 1.03–3.59).

Conclusion: The results of phenotypic characterization analysis were similar to other studies. By controlling the effects of potential confounders, some risk factors could be teratogens of isolated and sporadic microtia in East China.

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1. Introduction

Microtia is defined as a malformation of the auricle with varying severity ranging from minimal structural abnormalities to a total absence of the pinna, the latter is always called anotia [1]. The prevalence of microtia is reported to range from 0.83 to 17.4 per 10,000 births [2]. Microtia is known to be more prevalent in

males, and unilateral microtia is more common than bilateral microtia, with the right side being more commonly affected [3].

There are more than 18 different microtia-associated syndromes for which single-gene defects or chromosomal aberrations have been reported [4]. However, microtia often occurs as isolated and sporadic form, little is known about the etiology of isolated and sporadic cases. A number of studies have found that gestational exposure to alcohol, cigarette, influenza virus or medications can be associated with isolated microtia [5–7]. But the following reasons lead us to do this work. First, China, especially East China, has its own cultures and policies. Most women are primipara, and they do not smoke, drink alcohol or take medicine during pregnancy; then, the common potential risk factors in East China have been addressed in this work; next, some

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studies distinguished between syndromic and isolated cases, but not familial and sporadic cases; finally, we did a large sample case–control study and adjusted by sex, age, region, syndrome and family history.

The objective of this study is to describe the phenotypic characterization of microtia with our study, and analyze potential risk factors for isolated and sporadic microtia using case–control study in East China.

2. Methods

The study analyzed data from the hospital-based recruitment for deliveries. A detailed medical examination and retrospective chart review was performed for 911 patients with congenital microtia for phenotypic characterization study, who presented to our Eye & ENT Hospital of Fudan University between 2007 and 2013. Patients with microtia were considered isolated if they did not present any other malformation or if they only presented selected minor congenital malformation, like pre-auricular pit or pre-auricular tag [5]. Cases with microtia on one side and another selected minor ear malformation on the other side were categorized as unilateral. The patients whose relatives had any type of auricle malformation, such as microtia, pre-auricular tags or tragus defects, were categorized as having familial microtia.

All the patients and their parents received detailed medical examinations performed by one clinician to classify the type of microtia according to the classifications of Marx [8] and to determine whether other congenital malformation were present in this study. Each patient's clinical data was recorded in a questionnaire. Then, the patient's parents were asked to fill out the other part of this detailed questionnaire regarding their particulars and health histories, including name, sex, age, region, living environment, periconceptional cold-like syndrome, folic acid or medications use, history of spontaneous abortion, threatened abortion and prevention treatments, family history related to microtia or other congenital malformations. In this process, we explain and provide illustrations of the different types of auricle malformations.

All 911 microtia cases come from China, and nearly 80% from East China. Then were adjusted by sex, age, region, syndrome and family history. The case and control group had the same sex proportion; all participants were less than 18 years old and come from the same district, East China; the syndrome type and/or family history of microtia were excluded. Finally, 460 isolated and sporadic microtia fit these inclusion criteria for risk factor study, and were compared with 562 random normal controls. The normal controls were enrolled by field investigation. All controls and their parents without any birth defects, especially pre-auricular fistula

or tags. The controls had the same questionnaire, and the sex, age and region had been matched with 460 isolated and sporadic microtia in East China. These data were gathered by personal interviews and detailed questionnaires filled out by their parents. The data were analyzed using SPSS 16.0 (IBM, USA) by the chi square test, in which a *p* value less than 0.05 were considered statistically significant.

3. Results

A total of 911 patients were enrolled in this study, including 635 males (69.7%) and 276 females (30.3%), and 237 patients (26.0%) were affected on both sides. The majority of the microtia cases developed sporadically (838 cases; 92.0%), but 73 (8.01%) cases of familial microtia were discovered. Isolated microtia with no other malformation was observed in 633 cases (69.5%), and 278 cases (30.5%) demonstrated microtia in association with some other abnormality, including 168 cases (18.4%) with mandibular dysplasia, 80 cases (8.78%) with pre-auricular fistula or tags, 23 cases (2.52%) with eye defects, 17 cases (1.87%) with tragus defects, 10 cases with cleft lip and palate, 4 cases with cardiac defects and 9 cases with other defects, such as polydactyly, spinal defects and anosmia (Table 1).

There were a total of 1148 malformed ears; 657 ears (57.2%) were affected on the right side, and 491 ears (42.8%) were affected on the left side. Marx I microtia was observed in 205 ears (17.8%), Marx II microtia in 118 ears (10.3%), Marx III microtia in 814 ears (70.9%), and anotia in 11 ears (0.96%)(Table 2). There was significant difference between males and females for the degree of microtia: Marx I degree, $\chi^2 = 18.21$, $p < 0.01$; Marx III degree, $\chi^2 = 11.75$, $p < 0.01$. Boys have a greater auricle malformation degree than girls. The association of another abnormality with microtia significantly differed between males and females ($\chi^2 = 19.15$, $p < 0.01$), with girls having a greater number of associated abnormalities.

Table 3 shows mothers of children with microtia were more likely to have suffered a periconceptional cold-like syndrome as well as to have had a history of previous spontaneous abortion. Inflammatory infection (aOR, 3.56; 95% CI, 2.07–6.13) and chemical exposure (aOR, 2.77; 95% CI, 1.78–4.32) was associated with a higher risk of microtia. However, threatened abortion in the first trimester of pregnancy was not the risk factor (aOR, 1.14; 95% CI, 0.78–1.67); using progesterone may increase the risk (aOR, 1.92; 95% CI, 1.03–3.59). By contrast, using folic acid during the first trimester of pregnancy reduced the risk of microtia (aOR, 0.35; 95% CI, 0.27–0.47).

Some pregnant women would like to use traditional Chinese medicine, like radix isatidis, pseudo-ginseng or goldthread root to

Table 1
Characteristics of the 911 patients with microtia.

Characteristic	Total		Male		Female	
	Cases	%	Cases	%	Cases	%
Total Cases	911	100	635	100	276	100
Bilateral	237	26.0	173	27.2	64	23.2
Isolated microtia	633	69.5	452	71.2	181	65.6
Familial cases	73	8.01	–	–	–	–
Total defects	311	34.1	188	29.6	123	44.6
Pre-auricular fistula or tags	80	8.78	47	7.40	33	12.0
Mandibular dysplasia	168	18.4	108	17.0	60	21.7
Tragus defects	17	1.87	10	1.57	7	2.54
Eye defects	23	2.52	13	2.05	10	3.62
Cardiac defects	4	0.44	2	0.31	2	0.72
Cleft lip and palate	10	1.10	6	0.94	4	1.45
Other defects	9	0.99	2	0.31	7	2.54

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