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Renal ultrasound abnormalities in children with syndromic and non-syndromic microtia

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ABSTRACT

Objective: Renal abnormalities are commonly considered in the work up of pediatric patients with external ear malformations. However, there is little consensus regarding an appropriate renal screening protocol for patients with microtia. We sought to characterize renal abnormalities detected on ultrasonography in pediatric patients with microtia.

Methods: We conducted a retrospective cohort study of pediatric patients diagnosed with microtia who underwent renal ultrasound from 1991 to 2014 at a single tertiary academic institution. Renal ultrasound reports and medical records were reviewed to assess for renal abnormalities and to determine whether patients required specialist follow-up or interventions. Audiograms and otolaryngology notes were used to determine patterns of hearing loss. The following additional information was recorded from the electronic medical records: patient sex, microtia grade (I-IV), microtia laterality, and known associated syndromes. Characteristics were compared between those who did and did not have renal ultrasound findings using Fisher's exact test. Univariate logistic regression analysis was performed to determine factors associated with renal ultrasound findings.

Results: The majority of patients in this cohort were syndromic (n = 51, 64%) with grade III microtia (n = 46, 58%) and conductive hearing loss (n = 58, 72%). Syndromic children with microtia demonstrated a higher crude rate of renal ultrasound abnormalities (22%) than children with isolated microtia (7%). Of these patients, 69% required specialist follow-up. Univariate logistic regression analysis did not identify predictors that were significantly associated with renal ultrasound findings.

Conclusion: Fairly high rates of abnormalities in syndromic and non-syndromic patients may warrant screening renal ultrasound in all patients with microtia, especially given the high percentage of findings requiring renal follow-up. A prospective study to formally evaluate screening efficacy is needed.

1. Introduction

Microtia is a spectrum of congenital auricular abnormalities often accompanied by hearing loss, with prevalence ranging between 0.83 and 17.4 per 10,000 births depending on region and ethnicity [1]. The presentation can range from mild structural deviations to complete absence of the ear (anotia), commonly classified from grade I to grade IV [2,3]. Microtia may exist in isolation or arise as part of a genetic syndrome in 20–60% of cases [4–7], necessitating careful clinical assessment. Many of these multiple congenital anomaly syndromes (e.g. Oculo-auriculo-vertebral, CHARGE, Townes-Brocks, Nager, and Branchio-oto-renal) have high incidences of renal abnormalities alongside other organ defects and dysmorphic features [8]. Thus, children with microtia may demonstrate a variety of renal abnormalities [8] ranging from renal agenesis and horseshoe kidney to vesicoureteral reflux.

Although there is some published data describing an association between microtia and renal abnormalities, there remains a paucity of empirical evidence to inform best practices for utilizing renal ultrasound in the evaluation of patients with microtia. Some authors suggest screening all patients with microtia for renal abnormalities [9,10]. Others recommend microtia patients undergo a routine screening renal ultrasound at the time of diagnosis and cervical spine X-rays at 3 years of age [10,11]. On the other hand, recent studies demonstrate that renal ultrasonography is not indicated in children with minor ear defects unless accompanied by other dysmorphic features, suggestive family history, or known risk factors such as gestational diabetes [8,12].

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However, there is a lack of clarity on whether the presence of microtia specifically, rather than other external ear anomalies, is enough to warrant sonographic investigation.

Given the known association of microtia and renal abnormalities, limited description of demographic predictors, and potential for screening utility and earlier diagnosis of associated syndromes, we sought to retrospectively characterize the renal ultrasound results of children with both syndromic and isolated microtia at our academic institution.

2. Methods

With institutional review board approval, a retrospective chart review was conducted of all patients: 1) under 18 years of age, 2) with documented microtia, and 3) who underwent renal ultrasound between 1991 and 2014 at a tertiary academic medical center. The following information was recorded from the electronic medical records of eligible patients: patient sex, microtia grade (I-IV), microtia laterality, known associated syndromes, presence of sensorineural or conductive hearing loss, renal ultrasound findings, and whether patients underwent renal or urological follow-up. The majority of non-syndromic (n = 27, 93%) and syndromic (n = 46, 90%) patients had their diagnosis made by a medical geneticist at our institution. Patients without genetics consultation within our system were categorized based on the presence or absence of a specific diagnosis in the treating physician's notes (an otolaryngologist with or without a plastic surgeon). In most cases, patients without specific genetics consultation in our medical record were seen in craniofacial clinic, which is supervised by a geneticist. Characteristics were compared between those who did and did not have renal ultrasound findings using Fisher's exact test. Univariate logistic regression analysis was performed to determine factors associated with renal ultrasound findings. Significance was defined as $P < 0.05$. Statistical analyses were performed using STATA/SE software (version 14.0, StataCorp, College Station, TX).

3. Results

A total of 80 patients were included in this study: 42 boys (52%) and 38 girls (48%). Demographics, microtia type, renal ultrasound findings, and syndrome-related characteristics are shown in Table 1 for all patients and separately for those with and without renal ultrasound findings. The majority of patients in this cohort were syndromic (n = 51, 64%) with grade III microtia (n = 46, 58%) and conductive hearing loss (n = 58, 72%). 7 patients had mixed hearing loss and all of them were syndromic. Oculo-auriculo-vertebral spectrum (OAVS) was the most common syndrome identified with 34 patients having this diagnosis. CHARGE was the second most common with 5 patients. Among the patients with isolated microtia, 5 (17%) had other comorbidities including Tetralogy of Fallot, pulmonary atresia, polydactyly, and developmental delay.

Overall, 13 patients (16%) had abnormal renal ultrasound findings and the majority of these patients (69%) required renal follow up. Patient gender, microtia side, and microtia grade were not significantly different between patients with and without renal ultrasound findings ($p > 0.05$). The presence of an associated syndrome also was not significantly associated with having a renal ultrasound finding ($p = 0.119$). Renal findings included duplicated collecting systems, ectopic kidneys, dysmorphic kidneys, solitary kidneys, and hydronephrosis/pelviectasis (Table 2). These occurred with grades I, II, and III microtia. Patients were followed by nephrology and/or urology for medical problems including acute renal failure, chronic kidney disease, vesicoureteral reflux (VUR), and recurrent urinary tract infections with management including avoidance of nephrotoxic drugs, antibiotic prophylaxis, and reconstruction of ectopic/dysmorphic kidneys. Of the 4 patients without follow-up, only one was deemed a variation of normal while the other three were not evaluated within our system.

Table 1
Cohort characteristics.

Characteristic		Renal Anomalies	
	All	No	Yes
	No. (%)	No. (%)	No. (%)
Total	80 (100%)	67 (84%)	13 (16%)
Gender			
Male	42 (52%)	35 (52%)	7 (54%)
Female	38 (48%)	32 (48%)	6 (46%)
Microtia side			
Bilateral	17 (21%)	12 (18%)	5 (38%)
Left	23 (29%)	19 (28%)	4 (31%)
Right	40 (50%)	36 (54%)	4 (31%)
Microtia grade			
I	17 (21%)	13 (19%)	4 (31%)
II	16 (20%)	13 (19%)	3 (23%)
III	46 (58%)	40 (60%)	6 (46%)
IV	1 (1%)	1 (1%)	0 (0%)
Hearing Loss			
None	2 (2%)	1 (1%)	1 (8%)
CHL only	58 (72%)	52 (78%)	6 (46%)
Mixed, SNHL and CHL	7 (9%)	6 (9%)	1 (8%)
No audiology report	13 (16%)	8 (12%)	5 (38%)
Renal follow-up			
Yes	9 (11%)	0 (0%)	9 (69%)
No	68 (85%)	67 (100%)	1 (8%)
Unknown	3 (4%)	0 (0%)	3 (23%)
Syndromic			
Yes	51 (64%)	40 (60%)	11 (85%)
No, isolated microtia	29 (36%)	27 (40%)	2 (15%)
Specific Syndrome			
Branchio-oto-renal	2 (2%)	2 (3%)	0 (0%)
CHARGE	5 (6%)	3 (4%)	2 (15%)
DiGeorge	2 (2%)	1 (1%)	1 (8%)
Nager	1 (1%)	1 (1%)	0 (0%)
Noonan	1 (1%)	0 (0%)	1 (8%)
OAVS	33 (41%)	28 (42%)	5 (36%)
OAVS and Vater	1 (1%)	0 (0%)	1 (8%)
Townes-Brock	1 (1%)	0 (0%)	1 (8%)
Treacher Collins	2 (2%)	2 (3%)	0 (0%)
Trisomy 13	1 (1%)	1 (1%)	0 (0%)
VACTERL	1 (1%)	1 (1%)	0 (0%)
Unknown	1 (1%)	1 (1%)	0 (0%)
Not applicable	29 (36%)	27 (40%)	2 (15%)

Abbreviations: conductive hearing loss (CHL), sensorineural hearing loss (SNHL), Oculo-Auriculo-Vertebral Spectrum (OAVS).

Table 2
Renal ultrasound findings for patients with isolated microtia and associated syndromes.

Renal Ultrasound Findings	Total	Isolated Microtia	Syndromic
	No. (%)	No. (%)	No. (%)
Total	80 (100%)	29 (36%)	51 (64%)
Renal anomalies	13 (16%)	2 (7%)	11 (22%)
Duplicated collecting system	2 (2%)	1 (3%)	1 (2%)
Ectopic kidney	1 (1%)	1 (3%)	–
Dysmorphic and ectopic kidney	2 (2%)	–	2 (4%)
Hydronephrosis/Pelviectasis	6 (8%)	–	6 (12%)
Solitary kidney	2 (2%)	–	2 (4%)
Temporary, resolved findings	4 (5%)	1 (3%)	3 (6%)
No anomalies	63 (79%)	26 (90%)	37 (73%)

Among those with ultrasound abnormalities, 11 (85%) had an associated syndrome with OAVS being the most common (n = 6). Of 29 patients with non-syndromic, isolated microtia, 2 (7%) had renal abnormalities while 1 (3%) had temporary hydronephrosis that resolved during the study period. None of these patients had other comorbidities. Of the two non-syndromic patients with renal ultrasound findings, one with an ectopic kidney pursued follow-up outside of our institution and