ORIGINAL ARTICLE Egypt. J. Hum. Genet. Vol. 8, No. 2, Nov. 2007

Prevalence of Renal Anomalies in Children with Auricular Malformations among Attendance of Genetics Clinic in Ain Shams University

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ABSTRACT

Objective: Although many pediatricians pursue renal ultrasonography when patients are noted to have external ear malformations, there is much confusion over which specific ear malformations do and do not require imaging. The objective of this study was to delineate characteristics of a child with external ear malformations that suggest a greater risk of renal anomalies. We highlight several multiple congenital anomaly (MCA) syndromes that should be considered in a patient who has both ear and renal anomalies.

Patients and Methods: From September 2004 to April 2006, 50 patients with external ear anomalies (Preauricular pits and tags, low set ears, microtia, anotia, cup, phone, bat, and other forms of dysplastic ears) were consecutively recruited from the Genetics and Outpatient Clinic of Children's Hospital, Ain Shams University. Abdominal and pelvic ultrasound was done to each patient to screen for any renal anomaly.

Results: 4 patients (8%) with external ear anomalies as a part of MCA syndrome had renal anomalies, in the form of left hydronephrotic changes (n=1; 2%), absent left kidney and bifid right renal pelvis (n=1; 2%), and bilateral reflux and hydronephrotic changes (n=2; 4%).

Conclusions: Ear malformations are associated with an increased frequency of structural renal anomalies compared with the general population. This is because auricular malformations often are associated with specific MCA syndromes that have high incidences of renal anomalies. A renal ultrasound is useful not only in discovering renal anomalies, but also in the diagnosis and management of MCA syndromes themselves; it should be performed without delay in patients with ear anomalies, especially if those anomalies are part of MCA syndromes, in order to start conservative management or corrective surgery before irreversible kidney damage.

Key Words:

Renal anomalies, auricular malformations, multiple congenital anomaly, syndromes.

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INTRODUCTION

The association between external ear abnormalities and renal malformations has been reported previously. There is a general consensus on the need to rule out urinary tract malformation in a child with external ear malformation or when the isolated preauricular tag/pit is accompanied by other dysmorphic features.[1]

The aim of the present work was to study the prevalence of renal malformations among children with external ear anomalies attending the Genetics Clinic, Ain Shams University, and to identify the types of auricular anomalies that are associated most with renal malformations, in order to be screened for in our practice.

PATIENTS AND METHODS AND RESULTS

A total of 50 patients with ear anomalies (Table 1) received a renal ultrasound; their ages ranged between 1 month and 19 years with a mean of 4.9±5.57 years. 38 (76%) were males and 12 (24%) were females. Of the 50 studied cases, 4 (8%) displayed renal anomalies. All the 4 patients with renal anomalies received a diagnosis of MCA (Multiple congenital anomalies) syndrome.

The clinical characteristics of the 4 patients are summarized in (Table2) and (Figures1-4).

Table 1: External ear anomalies in the 50 studied cases.

Ear anomaly	n	%	Associated congenital anomalies
Cup ears (Fig. 5)	22	44	
Phone ears	8	16	
Large abnormal ear	1	2	
Bat ears	1	2	
Rudimentary auricles	4	8	Mild deafness (n=1) Lipodermoid cyst in left eye (n=1)
Raised lobule of the ear and abnormal shape	8	18	
Low set ears with no structural ear anomaly	4	8	Abnormal shape of the eye and rocker bottom heel (n=1)
Bat ears	2	4	
Total	50	100	

Case 1:

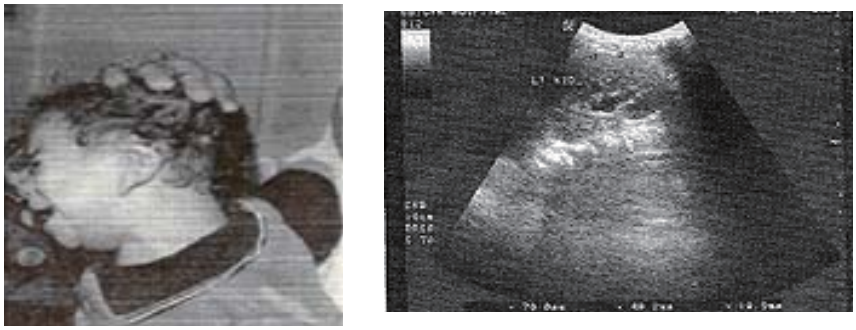

Fig. 1: A 5 years old female patient, 9th in order of birth of a non consanguineous marriage, presenting with bilateral abnormal shaped external ears and raised ear lobules, diagnosed as Cornelia de Lange syndrome. Renal ultrasound showing reflux and hydronephrotic changes.

Case 2:

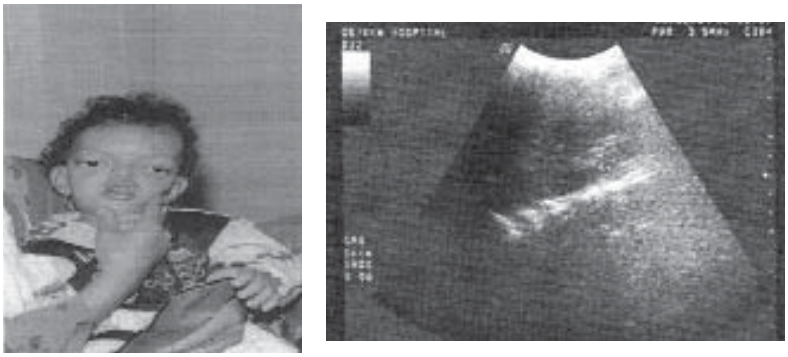

Fig. 2: A 2 years old female patient, 3rd in order of birth of a non consanguineous marriage, delivered by forceps and presenting with low set ears. she had rocker bottom heels and bilateral proptosis as a part of Crouzon syndrome. Renal ultrasound showing reflux and hydronephrotic changes.

Case 3:

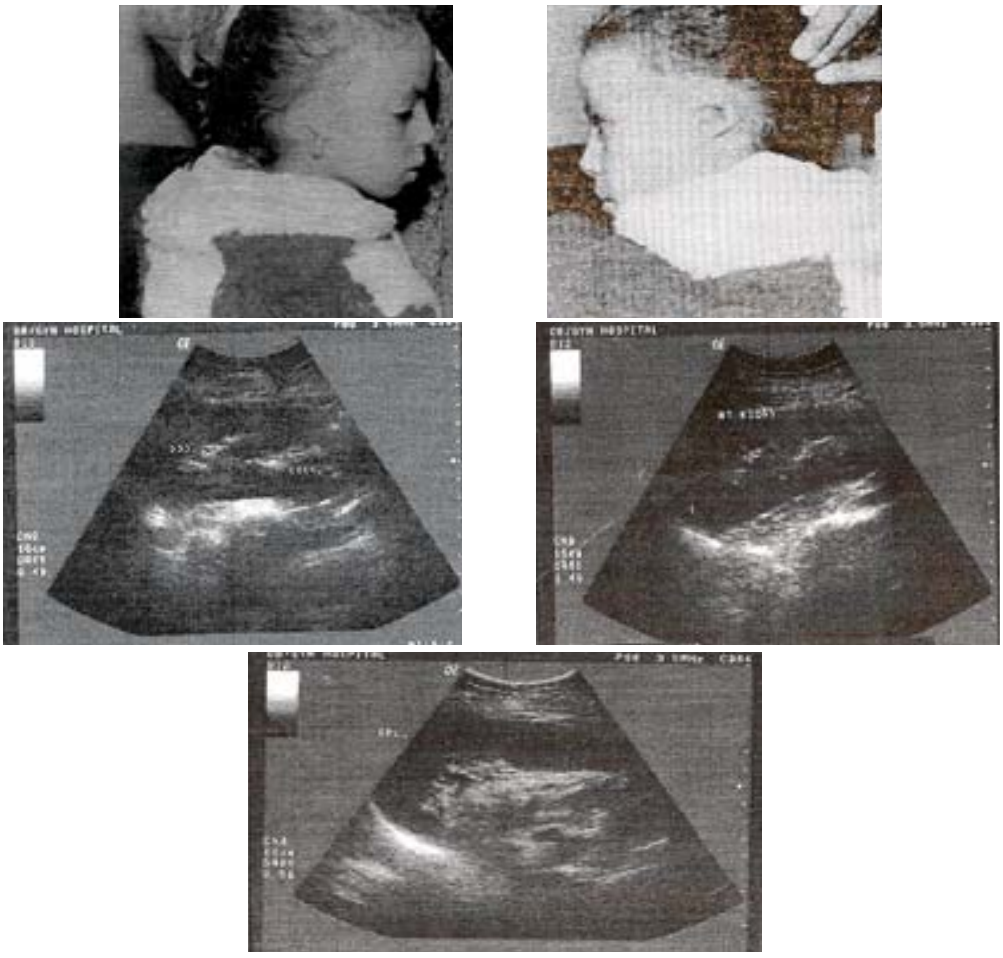

Fig. 3: A 2 years old female patient, 3rd in order of birth of a non consanguineous marriage. Mother had maternal gestational diabetes. She presented with bilateral microtia & rudimentary auricles. She was diagnosed to have Goldenhar syndrome and had a lipodermoid cyst in the left eye.

Case 4:

Fig. 4: A 6 years old male patient, 3rd in order of birth of a 1st cousin marriage, with a positive family history of external ear anomaly. He presented with cup ears as a part of MCA syndrome. He had left sided hydronephrotic changes on ultrasound.

Table 2: Renal ultrasound & clinical description of the 4 cases with renal anomalies.

Renal anomaly	n	%	Age (years)	Sex	Order of birth	Consan-guinity	Family history	Obstetric & drug history	Associated ear anomaly	Associated anomalies
Bilateral reflux & hydronephrotic changes(Fig.1)	1	2	5	♀	9th	N	N	N	Abnormal shape and raised ear lobules	Cornelia de Lange syndrome
Bilateral reflux & hydronephrotic changes(Fig.2)	1	2	2	♀	3rd	N	N	Forceps delivery	Low set ears	Rocker bottom heels, bilateral proptosis, Crouzon syndrome
Absent left kidney and bifid right renal pelvis (Fig.4)	1	2	2	♀	3rd	N	N	Maternal gestational diabetes	Bilateral microtia & rudimentary auricles	Goldenhar syndrome and lipodermoid cyst in left eye
Left hydronephrotic changes	1	2	6	♂	3rd	P	Ear anomalies	N	Cup ears (Fig. 3)	MCA syndrome
Total	**4**	**8**								

Of the 4 patients, 3 were females and one was male. Their ages ranged between 2 and 6 years. The 3 female children were product of non consanguineous marriages and had negative family history of external ear malformations or MCA syndrome, while the male child had 1st cousin parents and had family history of similar external ear anomaly in the father. Associated congenital anomalies were present in the 4 cases with the diagnosis of Cornelia de Lange syndrome in one female patient, Crouzon syndrome in one patient, Goldenhar syndrome with unilateral lipodermoid cyst of the eye in one patient and dysmorphic features with depressed nasal bridge, midfacial hypoplasia, polydactyly and low anterior hair line in one patient.

The remaining 46 cases had normal renal ultrasound (Figure 5).

Case 5:

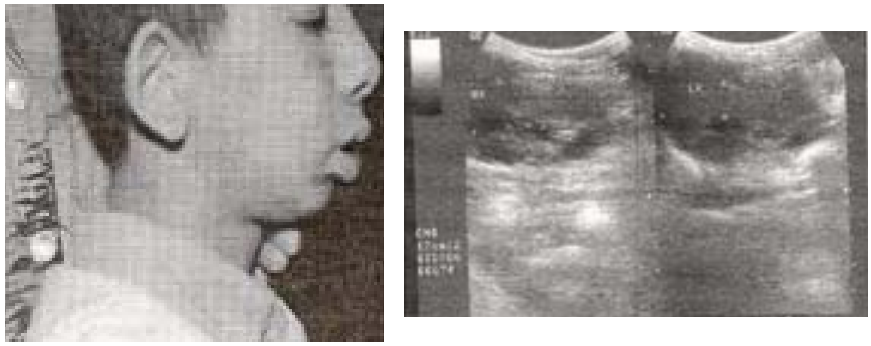

Fig. 5: An 8 years old patient, 2nd in order of birth of a 1st cousin marriage, he had phone ears and spastic cerebral palsy. Renal ultrasound revealed no abnormality.

DISCUSSION

In 1946, Edith Potter's association of crumpled, flattened ears with bilateral kidney agenesis[2] led pediatricians to order renal ultrasounds in virtually any type of ear anomaly. Many studies in the literature have noted a significant association between renal anomalies and various ear anomalies.[3] The objective of this study was to delineate characteristics of a child with external ear malformations that suggest a greater risk of renal anomalies.

In our study, a total of 50 patients with external ear anomalies had renal ultrasound. It revealed renal anomalies in 4 patients (8%). This percent of renal malformations is high when compared to the pediatric population (Structural renal anomalies occur in 1-3 per 100 live births)[4-6], and when compared to the general population.[7,8]

In the study, all the patients who had renal malformations on ultrasound (n=4; 100%) were diagnosed as multiple congenital anomalies (MCA) syndrome. This corroborates with the results of Wang et al.[3] who found 92% of the patients with renal anomalies to be a part of MCA syndrome. In another study by Leung and Robson, renal anomalies were described on ultrasound in 33% of patients with MCA syndrome.[9] Moreover, all our patients with isolated ear anomalies had normal renal ultrasound.

Ear and kidney development have been characterized in great detail, and it is well known that embryologically, ear and kidney primordia arise at different times and develop at different rates. Therefore the association between ear and kidney anomalies usually is not due to an isolated insult to the embryo that affects both developing structures at the same time. Prolonged embryonic insults may cause defects not just in ears and kidneys, but also in many other organ systems. This reflects the ongoing teratogenesis of toxic metabolites on all developing structures of the embryo.[3]

However, in some studies, isolated ear anomalies in the form of preauricular pits, sinuses and tags were associated with significant renal anomalies, ranging from hypoplasia to aplasia, either unilaterally or bilaterally, or anomalies of the urinary collecting system.[3,9,10,11]

In our study, one patient (2%) with rudimentary auricles had the diagnosis of Goldenhar syndrome and her ultrasound showed absent left kidney with bifid right renal pelvis. We reviewed the literature for the association between rudimentary auricles and renal anomalies; in the study of Bosco et al. patients with OAVS (Oculoauriculovertebral spectrum) had abnormal renal ultrasound result requiring at least follow-up according to an urologist.[12]

Because the incidence of ear malformations is relatively rare (Almost 1.3 per 10 000 live births)[3], it is difficult for 1 center to conduct a prospective population study large enough to accumulate sufficient numbers of children with ear anomalies to analyze. Our study was limited by review of only our clinic patients with ear anomalies, which introduces selection bias in that their ears (Or other organ systems) had to be anomalous enough to have been referred for a clinical genetics evaluation. Furthermore, our study enrolled patients who attended the Genetics Clinic and had ear anomalies in a consecutive manner. This led to the fact that some rare anomalies such as preauricular tags were not described among our patients. Further studies are recommended to focus on isolated rare anomalies in Egyptian children and it is also recommended to use other investigative tools to detect genitourinary anomalies on a structural as well as on a functional basis.

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