Acrorenal Syndrome: Unusual Association of Limb and Renal Anomaly

Acrorenal syndrome is a spectrum of congenital disorders characterized by the co-occurrence of distal limb anomalies (usually bilateral cleft feet and/or hands) and renal defects (e.g., unilateral, or bilateral agenesis) that can be associated with a variety of other anomalies such as those of genitourinary tract (genital anomalies, ureteral hypoplasias, vesicoureteral reflux), abdominal wall defects, intestinal atresias, and lung malformations. Familial cases have been reported which may have autosomal recessive or dominant inheritance (1-3).

Our patient, a 6-year-old boy, first product of second-degree consanguineous marriage, was noticed by his parents to have an abnormal right hand since birth. No medical attention was sought till 3 months of age when he developed high-grade fever. On examination, the child was febrile but hemodynamically stable. Examination revealed atypical cleft right hand with absent second, third, and fourth digits and deformed fifth digit with preserved thumb opposition and adequate grasp [Figure 1a]. There was no other skeletal abnormality or dysmorphism. Systemic examination was essentially normal, and the patient had normal hearing and vision. Investigations showed culture-positive urinary tract infection (UTI) with normal renal function tests (RFTs). X-ray of the right hand and wrist revealed absent third and fourth ray and absent phalanges of second finger with four malformed carpals with carpometacarpal dislocation [Figure 1b]. Ultrasound KUB (kidneys, ureters, and bladder) revealed hydroureteronephrosis of right kidney with left renal agenesis. Voiding cystourethrography showed Grade IV reflux on the right side. Dimercaptosuccinic acid (DMSA) scan done 12 weeks after the episode of UTI showed non-visualization of the left kidney with scarring of the upper pole of the right kidney. The patient was started on antibiotic prophylaxis with co-trimoxazole. The child, however, had recurrent breakthrough UTIs while on antibiotic prophylaxis. The child was therefore taken up for ureteric reimplantation of the right side at 2 years of age. Post reimplantation, the child has no febrile UTIs and is on follow-up in the pediatric nephrology clinic for breakthrough UTIs, proteinuria, hypertension, and RFTs. The parents were offered genetic counseling and evaluation, but they were not willing for the same.

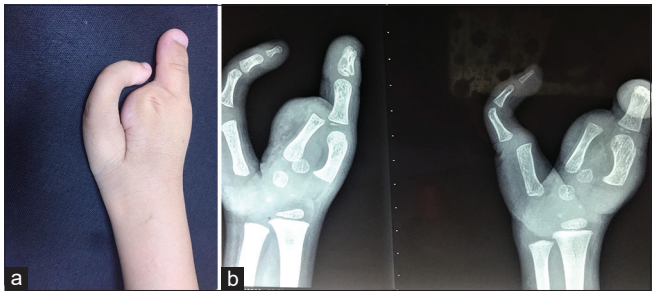

Figure 1: (a) Atypical cleft right hand with absent second, third, and fourth digits and deformed fifth digit with preserved thumb opposition and adequate grasp. (b) X-ray of the right hand and wrist revealed absent third and fourth ray and absent phalanges of the second finger with four malformed carpals with carpometacarpal dislocation

Limb and renal anomalies may occur together as isolated defects or may be a component of the malformation syndrome. The term *acrorenal syndrome* was coined by Curran *et al*. in 1972 for patients with congenital renal and limb anomalies,[1] although this syndrome was first reported by Dieker and Opitz in 1969.[2] These include longitudinal defects of the radius, ulna, tibia or fibula with characteristic renal and urinary tract abnormalities such as renal agenesis and hypoplasia.[3,4] Some children may exhibit additional malformations of the oromandibular region, trachea, lungs, eyes, and so on, and various syndromes have been described with these additional characteristic anomalies, including acro-renal-ocular syndrome, which has additional ocular anomalies such as optic nerve coloboma.[5]

Thus, the affected patients display huge phenotypic heterogeneity and variability in mode of inheritance.Thus, it is very important to evaluate children with congenital bone anomalies for other systemic anomalies, especially renal anomalies, as they may not be evident on physical examination. This will go a long way in minimizing/preventing morbidity in these children due to the timely institution of medical and/or surgical management.

Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent forms. In the form the patient(s) has/have given his/her/their consent for his/her/their images and other clinical information to be reported in the journal. The patients understand that their names and initials will not be published and due efforts will be made to conceal their identity, but anonymity cannot be guaranteed.

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Conflicts of interest

There are no conflicts of interest.

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