Polycystic kidney disease in neonate with acrorenal mandibular syndrome

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Clinical Dysmorphology 2014, **23**:133–137

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Received 4 June 2013 **Accepted** 17 June 2014

List of key features

Split foot
Polycystic kidney disease – neonate
Mandibular hypoplasia
Bronchogenic cyst
Uterus unicornis
Hydrosalpinx

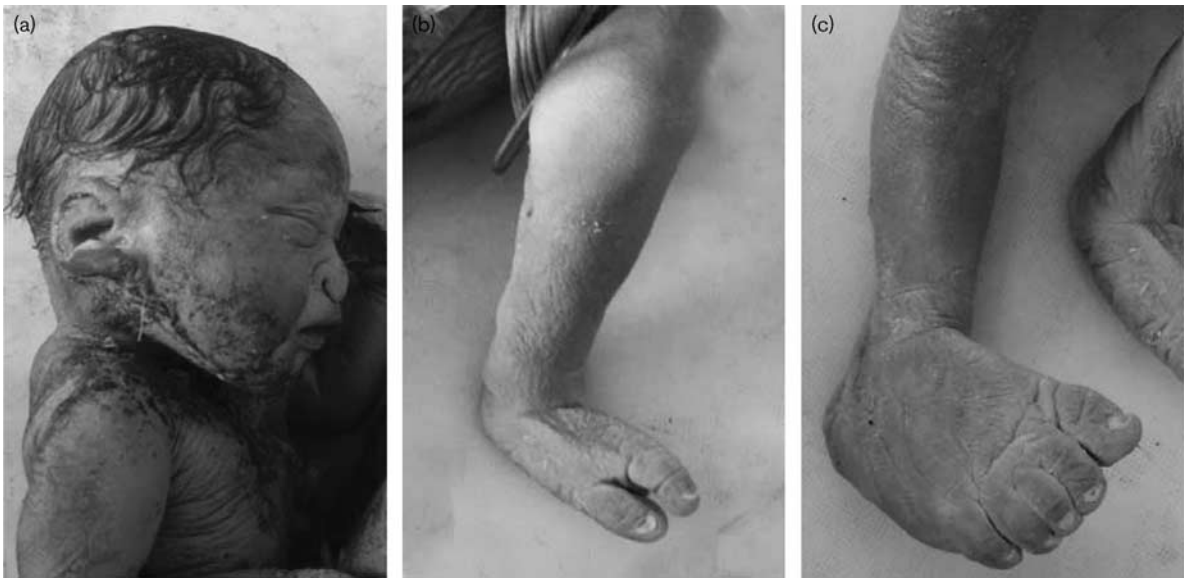
Introduction

A full-term female neonate with ectrodactyly, polycystic kidney disease, mandibular hypoplasia, uterus unicornis, bronchogenic cyst and spina bifida is reported. The features are compared with other cases of limb and renal abnormalities reported in the literature. This case along with similar cases reported by Halal *et al.* (1980), Evans *et al.* (2000); Tobias *et al.* (2001); Phadke and Manisha (2006); John (2007); and Girisha *et al.* (2012) delineates acrorenal mandibular syndrome as a distinct entity.

Case report

The proposita is the first child born to a couple who are second cousins. Her mother had regular antenatal care and was supplemented with folic acid 5 mg tablet from eighth week and with ferrous sulphate 100 mg from the 20th week

Fig. 1



Dysmorphic facial features showing low-set ears, folded ear lobes, beaked nose, micrognathia (a); claw-like and varus deformity of the left foot having two toes cleft between two toes, till metatarsal head (b); and right foot having five toes with varus deformity (c).

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DOI: 10.1097/MCD.0000000000000050

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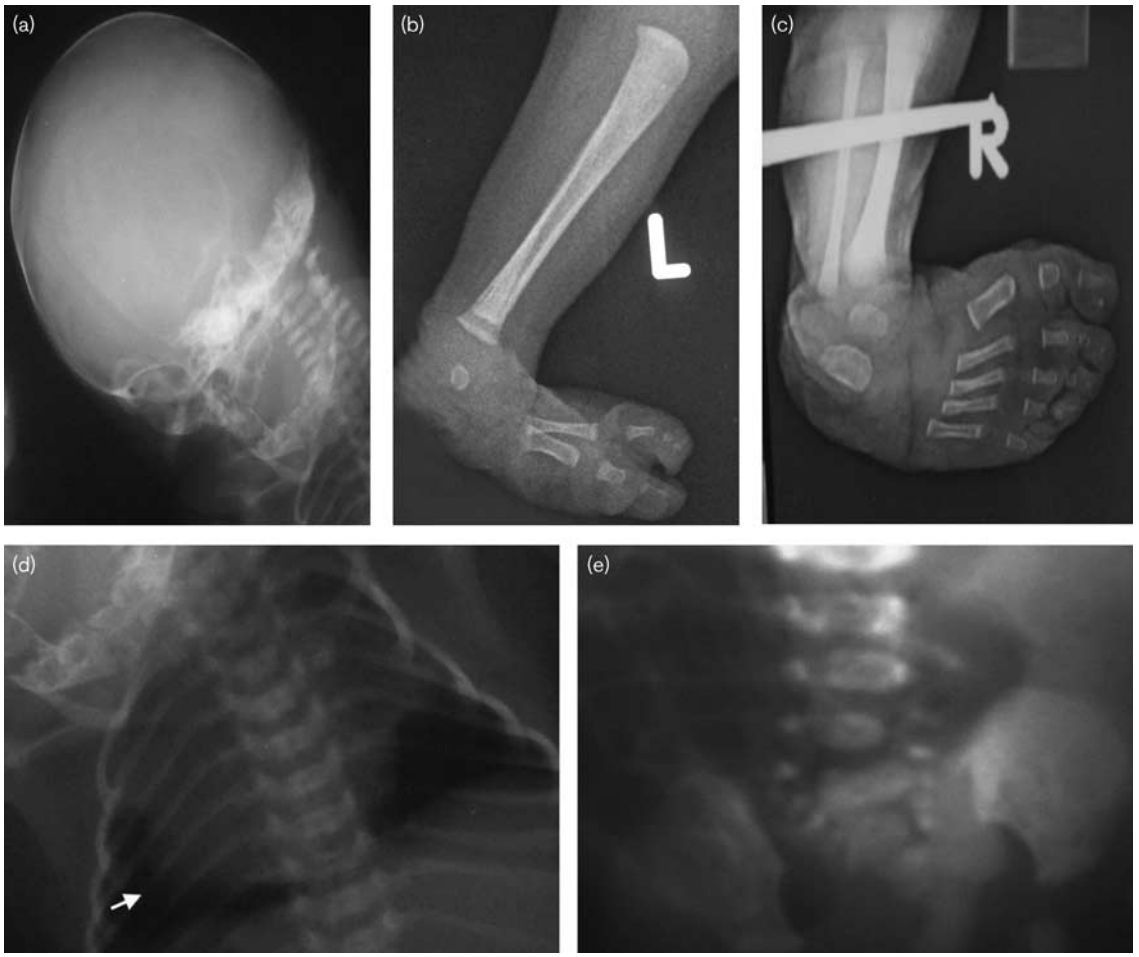
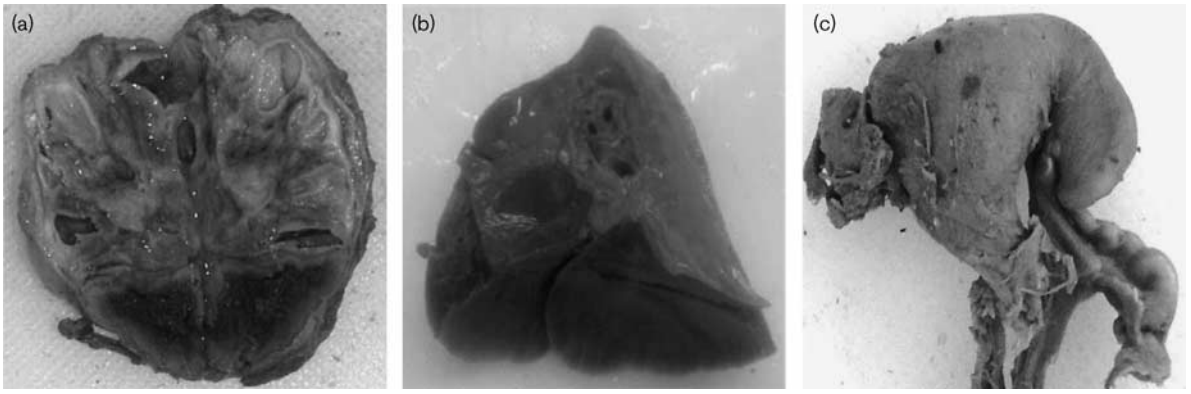
Fig. 2



Radiological features: mandibular hypoplasia, absent mandibular ramus (a); left foot ectrodactyly two metatarsal, two toes, talus (b); right foot varus deformity, five metatarsal, five toes, calcaneum, talus (c); cyst in the thorax (white arrow) (d); and spinal dysraphism, broadened sacral vertebra (e).

Fig. 3



Gross pathological images: right kidney uniformly enlarged, multiple cysts (a); right lung with cyst in lower lobe (b); and uterus unicornis, right fallopian tube distended with obliterated fimbria (ovary removed) (c).

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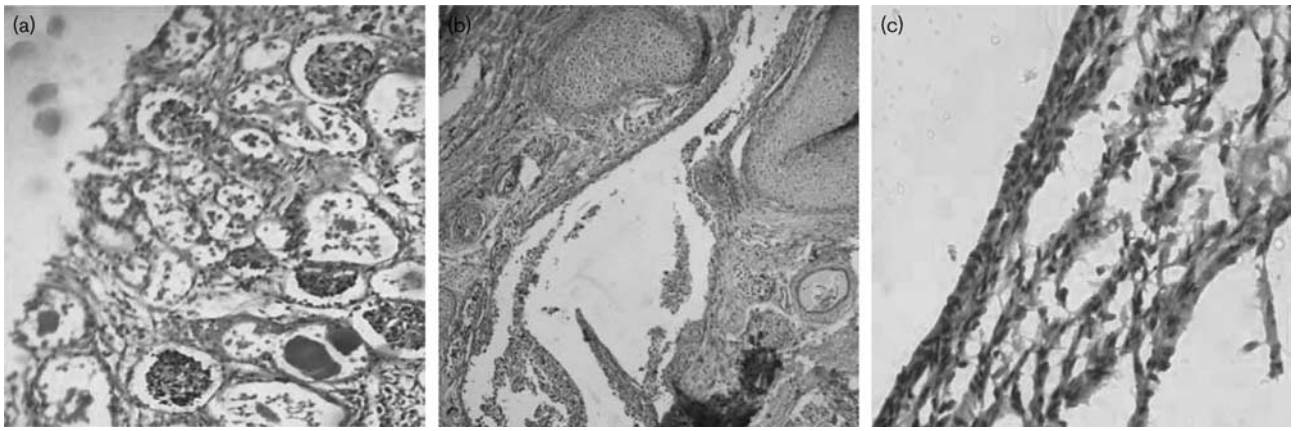
Fig. 4



Microscopic features of hematoxylin and eosin-stained smears: right kidney, glomerulocystic disease, foci of cyst wall lined by glomerular tuft, distended Bowman's spaces of glomeruli (a); bronchogenic cyst, cyst wall lined by ciliated columnar epithelium, surrounded by fibrous wall with islands of cartilage (b); and hydrosalpinx of right fallopian tube mucosal layer lined by low cuboidal epithelium, no plica (c).

of gestation daily. No history of teratogenic drug exposure was present. Ultrasound examination at the 31st week revealed severe oligohydramnios and multiple cystic lesions in the right kidney; foetal weight was 1787 g. The female neonate with multiple abnormalities was delivered at full-term by lower segment Caesarean section. Subseptate uterus in the mother was recorded. Birth weight was 1.5 kg with prenatal onset of growth retardation. Crown to heel length was 45 cm and head circumference was 30 cm; both were less than third percentile. Death occurred 35 min after birth, despite cardiopulmonary resuscitation. External anomalies were low-set ears, beaked nose, micrognathia (Fig. 1a), bilateral varus deformity, left claw foot having two toes, cleft between two toes, till metatarsal head (Fig. 1b) and right foot having five toes (Fig. 1c). There was no dimple, and long thick hair along the spine was present. Radiology showed mandibular hypoplasia (Fig. 2a), left foot ectrodactyly (Fig. 2b), right foot varus deformity (Fig. 2c), normal upper limb and long bones, cyst in the right thorax (Fig. 2d) and spinal dysraphism and broadened sacral vertebra (Fig. 2e). Spina Bifida Neurological Score was grade-V. Because of the limits of verbal descriptions, radiology and microscopy features are explained with illustrations. Gross pathology revealed enlarged right kidney, and multiple cysts were seen (Fig. 3a). In multicystic renal dysplasia, the kidney is enlarged, misshapen and irregularly cystic (Gilbert-Barness, 2007). The left kidney was normal. A cyst in the right lung lower lobe measured 1×2 cm (Fig. 3b). Uterus unicornis and right fallopian tube distended with obliterated fimbria (Fig. 3c). Right tube and genital organs were normal. Microscopy revealed glomerulocystic disease of the right kidney (Fig. 4a). Renal dysplastic features such as metaplastic cartilage and primitive ducts were not seen. Bronchogenic cyst (Fig. 4b) and right fallopian tube hydrosalpinx were seen.

Discussion

In 1980, Halal and colleagues described two cases of acrorenal mandibular syndrome with birth defect number 2778 and OMIM 200980 (Buyse, 1990) comprising of split hand–foot malformation, renal agenesis, uterine anomalies and severe mandibular hypoplasia. We considered but discounted a number of differential diagnoses. There are case reports showing a strong association between limb and renal anomalies. This association is seen in a very heterogenous group of acrorenal syndrome with OMIM 102520 and 201310 (Kroes *et al.*, 2004). Discussion of acrorenal syndrome is beyond the scope of this case report. As compared with ectrodactyly and polycystic kidney syndrome (Cameron, 1961), our case presented with mild mandibular hypoplasia, left split foot, bronchogenic cyst and unilateral right glomerular cystic disease, a prominent feature in early onset autosomal dominant polycystic kidney disease (Gilbert-Barness *et al.*, 2005). Subseptate uterus in the mother and uterus unicornis and unilateral hydrosalpinx in the neonate depict probable mode of inheritance as autosomal dominant. Karyotyping of our case could not be performed as we received the neonate fixed in formalin.

Bronchogenic cyst is a congenital bronchopulmonary foregut malformation. Other malformation occurring in conjunction with these type of lesions brings to mind the well-known VACTERL association, but our case lacks cardiovascular and gastrointestinal manifestation, imperforate anus and tracheoesophageal fistula (Newman, 2006). Spina Bifida Neurological Score was grade-V (Oi and Matsumoto, 1992).

Genital defects are frequently observed in severe renal anomalies (Halal *et al.*, 1980), similar to our case who had uterus unicornis left-side hydrosalpinx with right-side glomerulocystic kidney disease. We focused our attention on

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Table 1 Review and comparison of our case with cases of acrorenal mandibular syndrome reported

	Halal *et al.* (1980)		Evans *et al.* (2000)							
Features	Case 1	Case 2	Case 1	Case 2	Case 3	Tobias *et al.* (2001)	Phadke and Manisha (2006)	John (2007)	Girisha *et al.* (2012)	This case
Sex	Female	Female	Male	Female	Female	Male	Male	Male	Female	Female
Consanguinity	Present	Present	Present	Present	Present	–	–	–	–	Present
Downslanting palpebral fissures	–	–	Present	–	–	Present	–	–	–	–
Low-set, posteriorly rotated ears	–	Present	–	–	–	Present	Present	Present	Present	Present
Depressed nasal bridge	–	–	–	–	–	–	Present	Present	Present	–
Mandibular hypoplasia	Present	Present	Present	Present	–	Present	Present	Present	Present	Present
Narrow high-arched palate	–	–	Present	Present	Present	–	–	–	–	–
Cleft palate	–	–	–	–	–	–	Present	Present	Present	–
Philtrum	Normal	Normal	Short	Normal	Normal	Long and flat	Normal	Normal	Long	–
Lungs hypoplasia	Normal	–	Hypoplastic	Hypoplastic	Normal	–	Hypoplastic	Normal	Normal	Normal
Renal artery	Present	Present	Present	Present	Absent	Present	Present	Present	Present	Present
Hydronephrosis	–	–	–	–	–	–	–	–	Present	–
Bladder	Normal	Normal	Hypoplastic	Normal	Normal	Normal	–	Normal	Normal	Normal
Uterus	Didelphys	Unicornis	NA	Bicornuate	Cleft in the dome of uterus	NA	NA	NA	Hydrometrocolpos	Unicornis
Oligohydramnios	–	Present	–	Present	–	–	Present	–	–	–
Spine	Normal	Scoliosis	Kyphoscoliosis	Kyphoscoliosis	Normal	Normal	Normal	Normal	Normal	Spina bifida/spinal dysraphism
Joint dislocation	No	No	Hip	Hip	Hip	No	No	No	Knee and hip	No
Split foot (bilateral)	Bilateral	Bilateral	–	–	–	Unilateral	Bilateral	Bilateral	Bilateral	–
Syndactyly (feet)	–	–	–	Present	Present	Present	Present	–	Present	–
Others	–	–	Epicanthal folds, athelia	Microglossia, Potter's phenomenon	–	–	Deep-set eyes, hirsute body	–	Pinched nose, telecanthus	–
Sacral dimple	–	–	–	Present	–	–	–	–	–	–
Olfactory nerve	–	–	–	–	–	–	–	–	–	–
Severe limb deficiency	–	–	Present	Present	Present	–	–	–	–	–
Split hand (unilateral)	–	Present	–	–	–	–	–	–	–	–
Split foot (unilateral)	–	–	–	–	–	Present	–	–	–	Present
Varus deformity of the feet	–	Present	–	–	–	Present	–	–	–	Present
Bilateral renal agenesis	–	Present	Present	Present	Present	Present	Present	Present	–	–
Ureters	Present	–	Present	Present	–	–	–	Present	Present	Present
Polycystic kidneyes	Present	–	–	–	–	–	–	–	–	Present
Large cervical spine	Present	–	–	–	–	–	–	–	–	–
Sternal deformity	Present	–	–	Present	–	–	–	–	–	–
Rib hypoplasia (irregular and thin)	–	Present	–	–	–	–	–	–	–	–
Missing ribs	–	–	–	–	–	Present	–	–	–	–
Bronchogenic cyst	–	–	–	–	–	–	–	–	–	Present
Diaphragmatic hernia	Present	–	–	–	–	–	–	–	–	–
Pulmonary cysts or alveolar duct dilatations	–	Present	–	–	–	Present	–	–	–	–
Hemivertebra	–	Present	–	–	–	Present	–	–	–	–
Butterfly vertebrae	–	–	–	–	–	Present	–	–	–	–
Hydrosalpinx	–	–	–	–	–	–	–	–	–	Present
Choanal stenosis	–	–	–	–	–	Present	–	–	–	–
Scaphocephaly	–	–	–	–	–	Present	–	–	–	–
Flexion contractures (upper limb)	–	–	–	–	–	Present	–	–	–	–
Tibial shortening (unilateral)	–	Present	–	–	–	–	–	–	–	–
Broadened sacral vertebra	–	–	–	–	–	–	–	–	–	Present

–, features are absent/not present.

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features like ectrodactyly, renal anomaly, uterine malformation and mandibular hypoplasia, which characterize acrorenal mandibular syndrome. We reviewed and compared reports of cases having such anomalies in Table 1, and concluded that our case had features that should be considered under the description of acrorenal mandibular syndrome. Reporting and studying acrorenal mandibular syndrome cases help counselling parents regarding further issues.

Acknowledgements

Conflicts of interest

There are no conflicts of interest.

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