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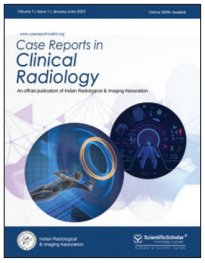

Case Report

A rare case report: Renal coloboma syndrome

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ABSTRACT

Renal coloboma syndrome (RCS), also called papillorenal syndrome, is a rare syndrome characterized by renal abnormalities and optic nerve dysplasia. We present a case of a neonate with renal and eye abnormalities. Along with the known diagnosis of multicystic dysplastic kidney, the neonate was diagnosed with coloboma of the right eye, making a probable diagnosis of RCS. Retinal detachment and decreased visual acuity are implications of the ocular malformations. Hypertension, proteinuria, and renal insufficiency, which frequently lead to end-stage kidney disease, are serious consequences of renal dysplasia.

Keywords: Renal coloboma syndrome, Papillorenal syndrome, Rare renal syndromes, Rare eye syndromes, Multicystic dysplastic kidney

INTRODUCTION

Renal coloboma syndrome is a rare congenital syndrome consisting of ocular and renal anomalies. Findings like hearing loss, central nervous system anomalies, laxity of ligaments, and genital anomalies are less common and well-documented. Ocular abnormalities may be inconspicuous and difficult to diagnose in utero without advanced screening methods. We report a case of a child with antenatal findings of renal anomaly which on post-natal investigations showed ocular findings with a possible diagnosis of renal coloboma syndrome.

CASE REPORT

A neonate born out of non-consanguineous marriage, weighing 2.5 kg at birth, and delivered through full-term normal vaginal delivery, presented with respiratory distress since birth and non-opening of the right eye. Neonate's mother at the 26th week of pregnancy for antenatal ultrasound screening revealed decreased amniotic fluid volume and a single live intrauterine fetus with a right enlarged kidney showing altered echotexture and multiple variable sized cysts. Left kidney was seen in the midline just above the urinary bladder. In the antenatal fetal magnetic resonance imaging study, on T2-weighted image, multiple variable-sized non-communicating cysts were seen, separated by hypointense septa. The left kidney is seen in the pelvic cavity more on the left side [Figure 1].

At birth, baby presented with acute respiratory distress and non-opening of the right eye. Further, computed tomography (CT) scan revealed an enlarged right kidney with renal parenchymal replacement by multiple variable-sized cysts, the largest of size (24 × 16) mm which, further, confirmed the antenatal diagnosis of the multicystic dysplastic kidney (MCDK) [Figure 2a].

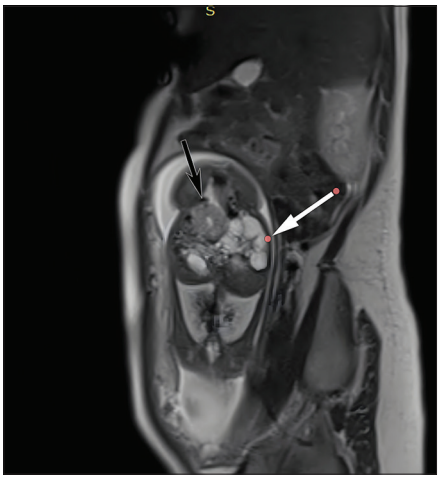

Figure 1: On T2 coronal plane fetal magnetic resonance imaging, the right kidney (White arrow) seems enlarged with multiple variable sized non-communicating cysts separated by hypointense septa, suggestive of multicystic dysplastic kidney, and left kidney (Black arrow) seen in the pelvic cavity more on the left side.

Furthermore, the left kidney was not seen in the left renal fossa but seen in the umbilical region in the midline below the level of origin of the inferior mesenteric artery with hilum facing anteriorly, measuring (23.5 × 30 × 39) mm (AP × TRA × CC) at the level of the upper border of L3 to lower border of L5 vertebra, which was suggestive of a left ectopic kidney [Figure 2b]. Both kidneys showed parenchymal enhancement and contrast filling in the bilateral ureters on the delayed scan, suggestive of normally excreting bilateral kidneys [Figure 2c and d]. CT scan also revealed a defect from the lateral globe wall on the temporal side through which outpouching of ocular contents suggestive of coloboma [Figure 2e]. On visible chest scan, areas of air space opacification in basal segments of bilateral (Left>Right) lower lobes with mild adjacent parenchymal atelectasis was noted and was conveyed to the clinicians [Figure 2f]. On clinical examination at birth, the right eye of the baby was entirely hazy white with non visualization of the structures behind the cornea [Figure 3]. Genetic studies for mutations in genes like PAX-2 or PAX-6 were not done due to resource constraint.

Along with the known diagnosis of MCDK, the neonate was diagnosed with coloboma of the right eye, making a probable

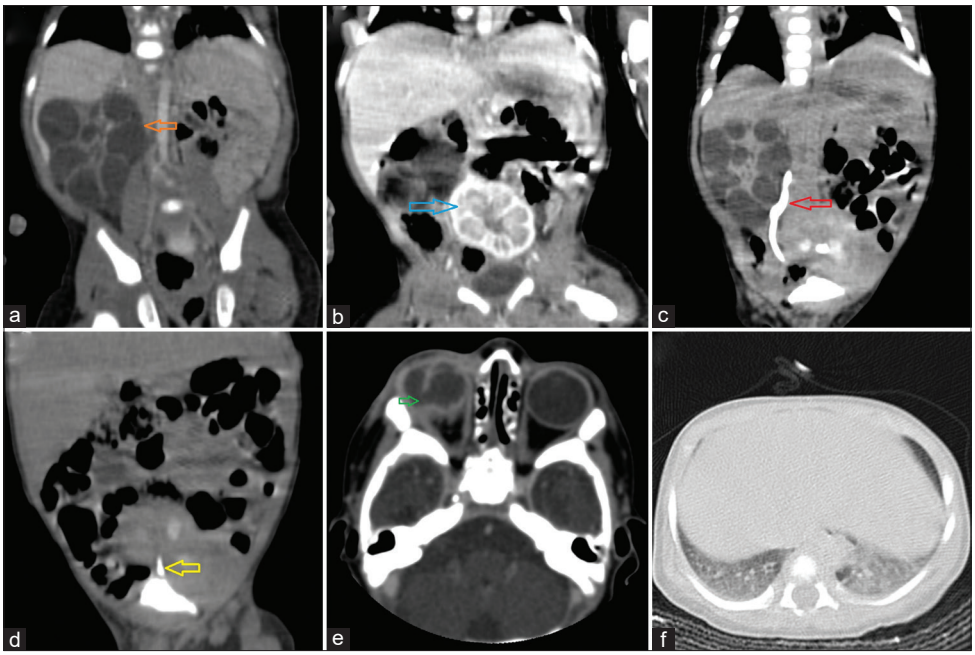

Figure 2: (a) computed tomography (CT) plain coronal view shows right enlarged kidney (average HU 30) with cysts of variable size with no communication between them (Orange arrow). (b) CT coronal plane view shows the left kidney at the level of the upper border of L3 to the lower border of the L5 vertebra, suggestive of the left ectopic kidney (Blue arrow). (c) Right kidney shows mild parenchymal enhancement (average HU 100) with contrast filling in the ureter on the delayed scan (Red arrow). (d) CT coronal image shows contrast filling in the left ureter in the delayed phase. (Yellow arrow). (e) CT axial section of the orbits discovered defect in the lateral globe wall on the temporal side (Green arrow), which allowed ocular contents to protrude. (f) Visible chest CT axial image shows areas of air space opacification in basal segments of bilateral (Left>Right) lower lobes with mild adjacent parenchymal atelectasis.

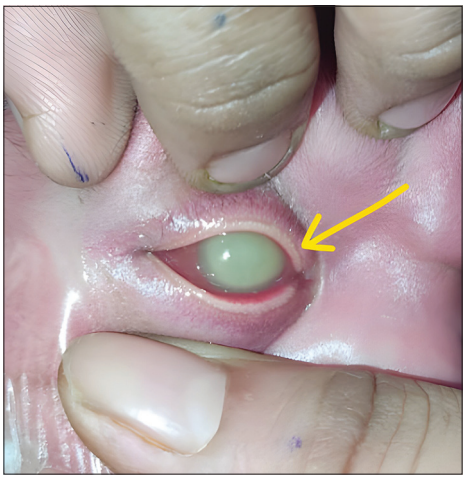

Figure 3: Image shows an entirely white eye with no visualization of any orbital structures.

diagnosis of renal coloboma syndrome (RCS) likely.

DISCUSSION

RCS, also called papillorenal syndrome, is an autosomal dominant condition characterized by optic nerve dysplasia and renal hypodysplasia.[1]

It results from autosomal dominant mutations in the PAX2 gene. Renal-coloboma syndrome predominantly affects the eyes and kidneys, some patients also experience high frequency hearing loss, central nervous system anomalies, and/or genital anomalies. This is consistent with the expression of PAX2 in these tissues during development.[2]

Although variability is found with differing mutations, there is also phenotypic variability with the same mutation and within the same family. MCDK is hypothesized to result from congenital ureteral obstruction during early nephrogenesis due to the frequently present finding of ureteric atresia.[3] A few cases of unilateral renal agenesis have also been documented.[3] Some forms of unilateral renal agenesis could also be due to the involution of early MCDK. Anatomic and structural studies have suggested that the anomalies observed in MCDK arise from malformation of the ureteric bud branches and ampullae. Studies on transgenic mice have also shown small numbers of kidney retinal defects, renal agenesis, unilateral kidney cysts, dilated ureters, and cystic abnormalities within the renal medulla.[3]

Renal findings in individuals with mutations in PAX2 include renal hypoplasia, renal dysplasia, MCDK, oligomeganephronia, and horseshoe kidney.

MCDK has been identified in 10% of reported cases.[1] Renal malformations or insufficiency are identified in nearly all patients with RCS and are frequently identified before the eye malformations.[1]

Developmental abnormalities of the optic fissure during the optic cup and stalk development result in a group of defects including orbital cysts, microphthalmia, optic disk dysplasia, and colobomas of the optic nerve and retina. All of the patients identified to have mutations in PAX2 have been observed to have colobomatous defects at the posterior pole of the globe.[2] Poor visual acuity has been described in about 75% of those with Papillorenal syndrome.[4]

There is insufficient data to suggest lung abnormalities as a part of RCS. The areas of air space opacification and adjacent parenchymal atelectasis found in this case, may explain the breathlessness with which the neonate presented, but this might be an incidental finding. Further studies might be needed to prove its association with the RCS.

CONCLUSION

RCS, or papillorenal syndrome, is a rare clinical, radiologic, and ophthalmological disorder. When both renal and ocular defects are present, the possibility of papillorenal syndrome should be taken into account. The patient with papillorenal syndrome should be monitored closely for high-frequency sensorineural hearing loss as well as ocular and renal anomalies.

Differential diagnosis

- CHARGE syndrome (coloboma, heart malformations, atresia choanae, retardation of growth and development, genital anomalies, and ear and hearing abnormalities)
- Branchio-oto-renal syndrome
- COACH or Joubert syndrome

TEACHING POINTS

1 RCS is an uncommon diagnosis, however, if present, monitoring for blindness, end-stage renal disease, and hearing loss should be done as they develop.
2 Family history pattern of autosomal dominance is due to PAX 2 mutation. Absence of significant family history is not an exclusion criterion like in our case.

MCQs

1. Possible diagnosis of a newborn with small dysplastic kidneys and optic nerve dysplasia is?
   a. Cat-eye syndrome
   b. Renal coloboma syndrome
   c. 22q11. 2 deletion syndrome
   d. CHARGE syndrome

Answer Key: b

2. Clinical manifestations in patients having coloboma renal syndrome include…
   a. Reduced visual acuity

b. Renal hypertension and end-stage renal disease
c. Associated sensory neural hearing loss
d. All of the above

Answer Key: d

3. Which of the following does not characterize coloboma renal syndrome?
   a. Antenatal ultrasound may reveal oligohydramnios.
   b. Malformation of the optic nerve is a common finding.
   c. CHARGE Syndrome can be a differential diagnosis
   d. Conductive hearing loss in half of the patients

Answer Key: d

Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent.

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Nil.

Conflicts of interest

There are no conflicts of interest.

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