LACHT syndrome with renal agenesis: A case report.

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Abstract
LACHT syndrome is characterized by pulmonary agenesis, congenital cardiac defects, and thumb anomalies. It was first described by Mardini and Nyhan in 1985 in four distinct families. There have been 11 cases in the literature with varying abnormalities. It has an autosomal recessive inheritance pattern. We present a case with pulmonary agenesis, pulmonary artery agenesis, subarterial ventricular septal defect (VSD), and ipsilateral upper limb and thumb anomaly on the left; and distinct from every single case in the literature; renal agenesis on the right.

Keywords: LACHT syndrome, Pulmonary agenesis, Congenital heart disease.

Introduction
LACHT syndrome which was first defined by Mardini et al. [1] in 1985 in four families has 3 main components: pulmonary agenesis, congenital cardiac defects, and thumb anomalies. It is a very rare autosomal recessive disorder whose underlying pathophysiology is unclear. It could occur with or without other abnormalities. 11 cases have been reported in the literature so far with various abnormalities. We present an unprecedented case with accompanying contralateral renal agenesis.

Case
The female neonate was delivered at 37th gestational week with a reported birth weight of 2100g by a 24 year-old healthy mother as her second child. Immediately after birth baby was admitted to neonatal intensive care unit due to respiratory distress. Her physical exam revealed left low-set ear and left upper limb deformity with thumb anomaly (Figure 1). We could not confidently identify the left lung parenchyma on chest X-ray. Therefore, we requested chest CT scan, and demonstrated left pulmonary agenesis (Figure 2). In addition, echocardiography showed left pulmonary agenesis and large subarterial ventricular septal defect (VSD); and abdominal US showed right renal agenesis. The parents did not have a consanguineous relationship, and the patient’s sister was healthy. Chromosome analysis was normal (46, XX). Taken together, we diagnosed the patient with LACHT syndrome. The patient had normal renal function, and the respiratory distress subsided with XXX. The patient was scheduled for VSD closure by cardiovascular surgery.

Discussion
All of the first 4 cases reported by Mardini et al. [1] in 1985 which were in consanguineous marriages had unilateral pulmonary agenesis, cardiac defects, and thumb anomaly. In 2009, Hasting et al. [2] reported three more cases without parental consanguinity, and all three patients had postnatal respiratory distress and cardiac surgery-requiring...
defects. In these cases, other comorbidities include rib abnormalities, hydrocephaly, epilepsy, spina bifida, and hypoplastic ovary [3-6].

Atik et al. [3] reported a 4.5-month old female case who was diagnosed before the elective inguinal hernia surgery with LACHT syndrome due to right pulmonary agenesis, pulmonary artery agenesis, dextrocardia, thumb anomaly, and ovary anomaly. Gomez et al. [4] presented a case with substantial upper respiratory hypoplasia. Jaiman et al. [5] reported a case with right pulmonary agenesis, ipsilateral pulmonary artery agenesis, and right thumb anomaly which resulted in fetal demise at 36th gestational week.

**Conclusion**

Out of 11 cases in the literature with LACHT syndrome, no urinary tract abnormalities had been reported. We hereby define the first case with LACHT syndrome with renal agenesis.

**References**


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