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Case Report

Potter Syndrome and Congenital Heart Disease

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ABSTRACT

Potter syndrome is a lethal congenital anomaly resulting from oligohydramnios due to renal agenesis and dysfunction. Because neonates with Potter syndrome have pulmonary hypoplasia, it is incompatible with life and the neonates will expire with respiratory distress during the first hours of life. Potter syndrome is rarely accompanied by congenital heart disease. We report a case of severe Potter syndrome with pulmonary valve atresia that expired a few hours after birth.

Keywords: Potter syndrome, Renal agenesis, Oligohydramnios, Pulmonary valve atresia

Introduction

The prevalence rate of Potter syndrome is 1 per 2000-5000 births and is a rare disorder [1] It is more common in men than in women [2, 3] Most mothers are primigravida between 20 and 30 years of age.

A multifactorial inheritance pattern leads to Potter syndrome. Autosomal dominant and recessive forms are reported, and some sporadic cases are seen as well. The most severe form is autosomal dominant with incomplete penetrance and variable expressibility.

Subtype I is associated with a polycystic kidney that is autosomal recessive, subtype II is due to renal dysplasia, subtype III is due to an autosomal dominant polycystic kidney, and subtype IV is accompanied by obstruction of the ureter or pelvis leading to hydronephrosis [4].

The amniotic fluid volume is determined by urine production and renal agenesis or hypoplasia leads to oligohydramnios. During the second and third trimesters of pregnancy, most of the amniotic fluid volume is contributed by urine production. Oligohydramnios limits fetal movements and leads to fetal compression, therefore different deformities occur in the fetal structures [5].

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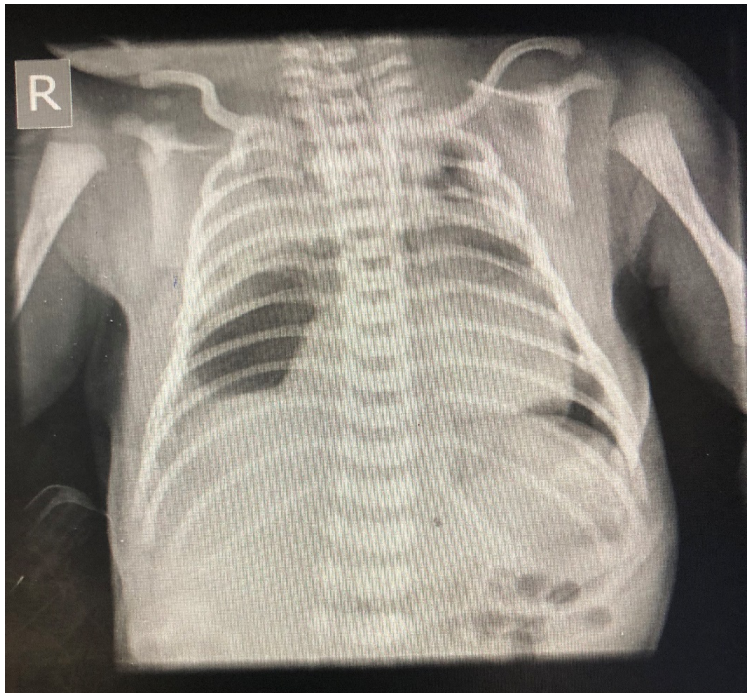

Figure 1. Chest x ray of the patient

Physical examination reveals infants with characteristic facies, defects in lower limbs, and genital organs. Potter facies is characterized by prominent epicanthic folds, low set ears, flattened nose due to uterine pressure, and recessed chin.

The congenital heart anomalies associated with Potter syndrome are ventricular septal defects, tetralogy of Fallot, and patent ductus arteriosus [5].

Potter syndrome has a poor prognosis in most cases. A large number of neonates die in utero or during the first few hours after birth. The most high-risk neonates are those suffering from pulmonary hypoplasia and the leading factor in death is respiratory distress [6].

Case Presentation

A 34-week-old baby boy was born by vaginal delivery of a primipar 18-year-old mother with a history of oligohydramnios with an Apgar score of 7-8/10.

Fetal sonography reported oligohydramnios and bilateral polycystic kidney at 20 weeks of gestation and an amniotic fluid index of zero was reported an hour before birth. The newborn was admitted to the neonatal intensive care unit after birth.

At the physical examination, the skin was wrinkled and bilateral club foot existed. The baby was hypotonic, and cyanosis and respiratory distress were prominent.

All the limbs had a saturation of 76%. After the hyperoxia and hyperventilation test, the baby was still cyanotic; therefore he was intubated and was set under a respirator.

Chest radiograph revealed severe pulmonary hypoplasia and due to prematurity, surfactant was administered intratracheally (Figure 1).

The condition of the newborn did not improve after surfactant administration and blood gas showed severe respiratory acidosis. The ventilator setup was changed to overcome the acidosis (Table1).

A 2.6 systolic murmur was audible at the upper left sternal border, without any sign of congestive heart failure.

The heart rate was 132 beats per minute and blood pressure in the right arm was 65.37 mmHg.

Bedside transthoracic echocardiography was performed and showed pulmonary valve hypoplasia, intact ventricular septum, relatively normal-sized right ventricle, left aortic arch, and functioning ductus arteriosus with continuous flow. The left ventricular ejection fraction was 60% and normal.

Table 1. Laboratory tests of the patient

CBC	Result
WBC (×1000mm^3)	19.5
RBC (Mill/mm)	4.4
Hb (gm/dL)	16
Hct (%)	48.4
MCV (fL)	109
MCH (Pgm)	36
MCHC (%)	33
Platelet (×1000/mm^3)	244
RDW (%)	16.6
Blood group	O
RH	Positive
Blood sugar (mg/dL)	99
BUN (mg/dL)	10
Creat (mg/dL)	0.8
Serum Na (mEq/L)	131
Serum K (mEq/L)	4.5
Serum Ca (mg/dL)	8.5
PH	7.015
PCO_2 (mmHg)	63.5
BE (mmol/L)	-15.6
BEecf (mmol/L)	-13.4
BB (mmol/L)	33.3
HCO_3 (mmol/L)	15.6
PO_2 (mmHg)	52.3
O_2 sat (%)	69.5

Prostaglandin E1 infusion was started for the newborn to maintain the ductal shunt to supply blood to the lungs.

The baby did not urinate at all and the trial to insert a urinary catheter failed.

Abdominal sonography was performed and showed normal liver size and parenchyma with normal gallbladder and biliary tract. Spleen was observed with a diameter of 29 mm and homogenous parenchyma.

At the anatomic position of the kidneys, no kidney was observed bilaterally and no ectopic kidney was observed in other parts of the abdomen, the bladder was empty.

Brain sonography showed normal ventricles without manifestations of intraventricular hemorrhage. No hydrocephaly was observed, without extra-axial effusion and the posterior fossa was normal.

The condition of the neonate deteriorated and the baby succumbed after 8 hours due to cardiorespiratory arrest.

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Discussion

Pulmonary hypoplasia is the most common and grave association seen in Potter syndrome. It leads to neonatal respiratory distress syndrome and pulmonary insufficiency. Many neonates present with cyanosis in the first hours of birth and suffer from respiratory acidosis due to CO_2 retention and ventilation-perfusion mismatch.

The prominent characteristics associated with Potter syndrome are structural deformities. The deficient amniotic fluid that is necessary for suitable fetal movement and body parts development, as well as uterine compression of the fetus, leads to deformities of the face and extremities. Genital abnormalities are seen in up to 70% of cases. Other abnormalities include congenital heart defects, pancreatic cysts, esophageal atresia, duodenal abnormalities, colonic agenesis, and Meckel's diverticulum. Hepatic fibrosis and biliary tree abnormalities are observed in subtype II. Intrauterine growth retardation, preterm labor, and preeclampsia are observed in multiple pregnancies.

Cardiac lesions have rarely been reported with Potter syndrome. Artune et al reported a total of 54 pregnancies with a mean gestational week of 19.8±4.6 at the time of the diagnosis. The mean maternal age was 27.28±6.03. Thirty-seven pregnancies were anhydramniotic, 13 fetuses had different associated anomalies, five cases suffered from multicyctic dysplastic kidney, five had bilateral renal agenesis, hypoplastic right heart syndrome was observed in one patient, one had clubfoot, and one had a ventricular septal defect and cleft palate. Karyotyping was performed and the result was normal in the fetuses with structural anomalies [7].

A large-scale mouse mutagenesis screen has been evaluated by San Agustin et al and showed that 29% of mutations causing congenital heart disease (CHD) also cause renal anomalies. The reported renal anomalies included duplex, multiplex kidneys, renal agenesis, hydronephrosis, and cystic kidney disease.

San Agustin et al assessed the clinical relevance of these findings. By examining the patients with CHD, it was revealed that a 30% co-occurrence of renal anomalies of a similar spectrum existed in the patients. The authors also demonstrated a common shared genetic etiology for CHD and renal anomalies, demonstrating that CHD patients are at increased risk for complications from renal anomalies. To elucidate the developmental link between renal anomalies and CHD, a collection of mutant mouse models provides a valuable resource [8].

Ethical Considerations

Compliance with ethical guidelines

No ethical considerations are declare in this report.

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Authors' contributions

All authors contributed equally to the preparation of this article.

Conflict of interest

The authors declare no conflict of interest.

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