A Suspicion of Potter Syndrome in G4P2A1 at 33 Weeks Gestation with Oligohydramnios and Severe Preeclampsia: A Case Report

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Abstract
Introduction: Potter syndrome is a rare case caused by oligohydramnios due to kidney failure in a fetus. Potter syndrome is characterized by pulmonary hypoplasia, skeletal malformation, and kidney abnormalities.
Case presentation: A 45-year-old G4P2A1 at 33 weeks gestation with oligohydramnios and severe preeclampsia came to the maternal and perinatal Emergency Unit at the Cibabat Regional General Hospital, Cimahi, complaining of labor contractions and delivered a baby. A male baby of 33 weeks gestation with a birth weight of 1295 grams was delivered. The baby had severe respiratory distress at birth, requiring resuscitation. Multiple congenital anomalies, such as widely separated eyes with epicanthic folds, low-set ears, receding chin, broad nasal bridge, and polydactyly, were noted. After 6 hours of life, the baby died due to respiratory failure.
Discussion: In this case, clinical manifestations of Potter's facies and a history of oligohydramnios support the suspicion of Potter syndrome. An autopsy or further examination was not carried out, so the etiology in this case has not been obtained.
Conclusion: There was a suspicion of Potter syndrome obtained from clinical manifestations and a history of oligohydramnios.

Keywords: Oligohydramnios, Potter Syndrome

Dugaan Sindrom Potter pada G4P2A1 Parturien 33 Minggu dengan Oligohidramnion dan Preeklamsia Berat: Sebuah Laporan Kasus

Abstrak
Pendahuluan: Sindrom Potter merupakan kasus langka yang disebabkan oleh kurangnya cairan ketuban akibat gangguan pada ginjal janin. Pada kasus potter syndrome dapat ditemukan hipoplasia pulmonal, malformasi tulang, dan kelainan ginjal.
Presentasi kasus: Ibu 45 tahun dengan G4P2A1 usia kehamilan 33 minggu dengan oligohidramnion dan preeklamsia berat datang ke IGD Maternal dan Perinatal RSUD Cibabat Cimahi dengan keluhan kontraksi persalinan dan melahirkan spontan bracht pervaginam. Bayi laki-laki usia 33 minggu, berat badan 1295gram, lahir dengan mengalami *distress* pernafasan berat dan dilakukan resusitasi. Kelainan kongenital multiple didapatkan pada bayi, yaitu jarak mata lebih lebar, adanya lipatan kulit yang menutupi sudut mata, posisi telinga lebih rendah, dagu lebih kecil, pangkal hidung yang lebih lebar, dan polidaktili. Dilakukan terapi suportif, setelah enam jam bayi meninggal karena *distress* pernafasan.
Diskusi: Pada kasus ini ditemukan manifestasi klinis *potter facies* dan riwayat oligohidramnion yang mendukung kecurigaan terhadap Sindrom Potter. Pada kasus tidak dilakukan pemeriksaan otopsi maupun pemeriksaan lanjutan sehingga etiologi pada kasus ini belum didapatkan.
Kesimpulan: Pada kasus ini didapatkan kecurigaan Sindrom Potter yang didapat dari manifestasi klinis dan riwayat oligohidramnion.

Kata kunci: Oligohidramnion, Sindrom Potter

Introduction

Potter Syndrome is a rare condition caused by a deficiency of amniotic fluid due to a renal disorder in the fetus, also known as oligohydramnios. This syndrome was first described by Edith Potter in 1946. Its manifestations include low-set and underdeveloped ears, long epicanthic folds, a flat nasal bridge, a small mandible, and other facial features. In addition, there are associated spinal malformations, such as clubbed hands and feet and joint contractures. Cases of Potter Syndrome often exhibit pulmonary hypoplasia, bone malformations, and renal abnormalities.[1,2]

Oligohydramnios can occur due to reduced urine production or excretion or due to amniotic fluid leakage, such as in premature rupture of membranes. Moreover, various causes of oligohydramnios include premature rupture of membranes, placental insufficiency, fetal anomalies, maternal drug use, complications in multiple pregnancies, chromosomal abnormalities, and idiopathic factors.[3] The incidence of Potter Syndrome is estimated to be 1 in 2,000 to 5,000 births, and it occurs more frequently in male infants.[4,5] Meanwhile, the incidence of Potter Syndrome in Indonesia is unknown. Potter Syndrome is generally fatal within the first few days of life, most often due to respiratory failure. Bilateral renal agenesis is incompatible with life outside the womb, and 33% of fetuses die in utero. A 70% survival rate has been reported among 23 infants with antenatal oligohydramnios and pulmonary hypoplasia.[6]

The severity of Potter Syndrome depends on the degree and duration of oligohydramnios.[7] Therefore, early detection through ultrasound during prenatal and early pregnancy is crucial to facilitate rapid evaluation and timely decisions regarding further examination and, if necessary, pregnancy termination. This case report highlights the importance of routine antenatal screening and assessment to identify suspicious cases requiring further investigation, definitive diagnosis, and timely management decisions.

Case Illustration

A male infant, born at 33 weeks gestation through spontaneous breech delivery, weighing 1295 grams, with scant amniotic fluid and a normal placenta. The baby did not cry immediately at birth, exhibited weak muscle tone, and had a flexed posture. After tactile stimulation, positive pressure ventilation was administered due to the absence of spontaneous breathing efforts. The baby's APGAR scores were 1/10 and 3/10 at 1 and 5 minutes after birth, respectively. Multiple congenital anomalies were observed, including low-set ears (Figure 1), widely spaced eyes, skin folds covering the corners of the eyes, a small chin, a broad nasal bridge (Figure 2), and polydactyly (Figure 3). Despite supportive therapy for six hours, the baby succumbed to respiratory distress.

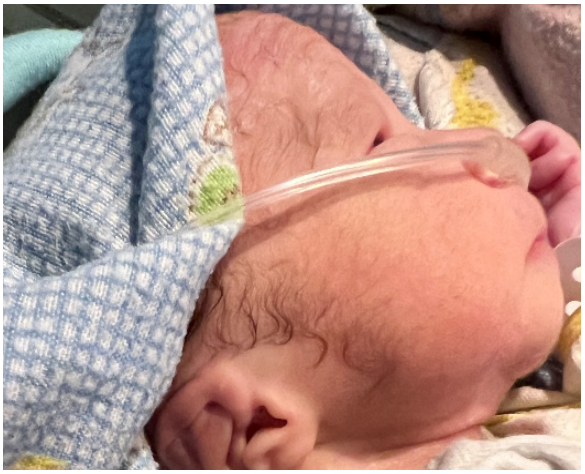

Figure 1 Low Set Ears

Figure 2 Widely spaced eyes, skin folds covering the corners of the eyes, small chin, and broad nasal bridge

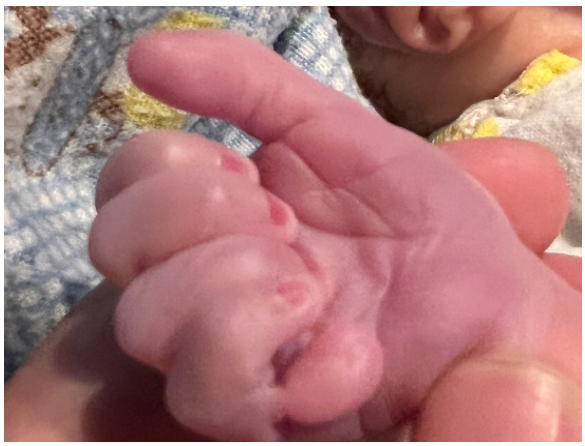

Figure 3 Polydactyly

The baby was born to a 45-year-old mother, G4P2A1, at 33–34 weeks gestation, although the ultrasound performed on the same day indicated a gestational age of 29 weeks, with oligohydramnios and fetal growth restriction accompanied by severe preeclampsia. The mother had a previously normal obstetric history, including two healthy children born through spontaneous vaginal delivery and a history of a miscarriage at six weeks gestation. She had a history of hypertension since four months of pregnancy and was not consistently taking antihypertensive medication. Prior antenatal examinations had raised suspicions of congenital anomalies and oligohydramnios. The mother was referred to a fetomaternal specialist but later presented to the Obstetrics and Gynecology Emergency Department at Cibabat Regional General Hospital, Cimahi, with labor contractions and subsequently delivered spontaneously in breech presentation.

Discussion

Potter Syndrome presents with clinical manifestations caused by intrauterine pressure due to a deficiency of amniotic fluid or oligohydramnios.[8] Oligohydramnios can be influenced by maternal, fetal, or placental factors and lead to poor neonatal outcomes. It is associated with obstetric conditions that cause uteroplacental insufficiency, such as chronic hypertension, vascular disease, thrombophilia, and preeclampsia. Additionally, oligohydramnios is correlated with the use of angiotensin-converting enzyme (ACE) inhibitors, non-steroidal anti-inflammatory drugs (NSAIDs), cocaine use, and maternal diabetes.[3] Moreover, decreased placental perfusion in preeclampsia can lead to oligohydramnios.[9] In this case, the mother of the infant with Potter Syndrome experienced severe preeclampsia, which probably played a role in the development of oligohydramnios.

Renal agenesis or hypoplasia is the most common cause of oligohydramnios due to reduced urine production, which determines the volume of amniotic fluid. Oligohydramnios is a condition of decreased amniotic fluid during pregnancy. In the intrauterine life before 16 weeks gestation, amniotic fluid production depends on transmembrane flow; afterward, it is primarily produced by fetal urine. A lack or absence of urine excretion from the fetal kidneys results in a reduced volume of amniotic fluid. Oligohydramnios

causes fetal compression within the uterus, limiting fetal movement and leading to physical deformities.[4,10] The genetic causes of this condition are still not fully understood, and further research is ongoing. However, evidence has been found regarding the role of Lim1 and Pax2 transcription factors during nephrogenesis, where a deficiency in these factors can lead to renal agenesis in mice.[11]co-expressed in the Wolffian duct and ureteric bud epithelia, play essential roles during the early steps of mouse kidney development. In humans, heterozygous mutations in these genes display a number of common kidney phenotypes, including hypoplasia and multicystic hypoplastic kidneys. Moreover, a high prevalence of mutations either in HNF1B or PAX2 has been observed in children with renal hypodysplasia. To gain a better understanding of Hnf1b and Pax2 interactions in vivo, we generated compound heterozygous mice for Hnf1b and Pax2 null alleles. We show here that compound heterozygous mutants display phenotypes similar to severe congenital anomalies of the kidney and the urinary tract (CAKUT Other mechanisms of Potter Syndrome include congenital polycystic kidney disease, urinary obstruction, and premature rupture of membranes, which are also generally associated with oligohydramnios.[4]

Most of Potter Syndrome cases are associated with pulmonary hypoplasia. This leads to insufficient lung oxygenation and respiratory distress syndrome. The infant may become cyanotic and develop respiratory acidosis due to carbon dioxide accumulation and ventilation-perfusion mismatch within hours after birth. Adequate chest expansion is crucial for fetal lung development. Compression of the fetus by the uterus and the pressure of intra-abdominal organs on the diaphragm result in failed thoracic expansion, causing the alveoli and airways to remain collapsed, leading to hypoplastic lungs.[4,12,13] In this case, a history of oligohydramnios during pregnancy and fetal renal anomalies detected antepartum support the suspicion of Potter Syndrome. At birth, the infant also experienced severe respiratory distress due to pulmonary hypoplasia, ultimately leading to the infant's death.

Potter Syndrome is generally characterized by distinct physical features caused by intrauterine pressure due to oligohydramnios. These include Potter facies, which consist of a flattened nose, epicanthic folds, a recessed chin, and low-set abnormal ears. Additionally, Potter Syndrome may present with pulmonary hypoplasia, Eagle-Barrett syndrome, bone malformations, ocular malformations, and cardiac defects.[2,12,13] In this case, the suspicion of Potter Syndrome arose from the clinical manifestations observed in the infant, including widely spaced eyes, epicanthic folds, low-set ears, a recessed chin, a flattened nasal bridge, and polydactyly. The history of antepartum oligohydramnios further supports the diagnosis of Potter Syndrome in this case. This is similar to the Potter Syndrome case reported by Latika et al., where a 40-year-old mother with oligohydramnios gave birth to a stillborn infant with similar facial characteristics and congenital anomalies, including ambiguous genitalia, anal atresia, and limb defects.[7]

Potter Syndrome can be categorized into several groups based on its underlying etiology: classic Potter Syndrome, caused by bilateral renal agenesis; Type I, caused by autosomal recessive polycystic kidney disease (ARPKD); Type II, caused by renal cystic dysplasia; Type III, caused by autosomal dominant polycystic kidney disease (ADPKD); and Type IV, caused by a prolonged obstruction in the kidneys or ureters. Classic Potter Syndrome is always fatal.[14] Therefore, it is crucial to detect this condition through ultrasound during prenatal care and early pregnancy to make timely decisions regarding pregnancy termination.

The evaluation of patients with Potter Syndrome includes investigations for non-renal abnormalities, autopsy, chromosomal analysis, urological examinations, or renal ultrasound in the parents. Routine screening and antenatal examinations are essential to diagnose oligohydramnios, thereby preventing complications that may arise from it. Although not all cases of oligohydramnios will lead to Potter Syndrome, regular fetal anomaly scans and amniotic fluid index assessments should be conducted to identify this syndrome.[15]

Potter Syndrome is an autosomal congenital disorder with a poor prognosis and is associated with various complications. The psychological impact on the family is often significant. Therefore, counseling family members, especially the mother, about the consequences of this condition is crucial for managing postnatal stress. Genetic counseling should also be provided to at-risk families. Continuous prenatal ultrasound monitoring is essential in subsequent pregnancies within affected families due to the risk of recurrence.[7] In this case, there was already a suspicion of oligohydramnios and congenital anomalies during antenatal care. However, before further action could be taken, the baby was born and identified as having Potter Syndrome.

The prognosis for Potter Syndrome is extremely poor. Nearly all infants born with this condition are either stillborn or die shortly after birth. Infants with hypoplastic lungs are at very high risk, with respiratory distress syndrome being the leading cause of death within hours or days after birth. According to an experimental study conducted at Johns Hopkins Hospital, injecting normal saline into the mother's womb can enhance lung development, but individuals with this condition typically require lifelong dialysis. Prognostic factors include the gestational age at diagnosis and the type and location of associated structural anomalies. The survival rate for Potter Syndrome is higher when the condition is caused by factors other than bilateral renal agenesis (classic Potter Syndrome). The infant mortality rate can reach 100% if no interventions are made during pregnancy.[16]

Conclusion

Potter Syndrome occurs when there is a deficiency of amniotic fluid during pregnancy (oligohydramnios). This lack of amniotic fluid can lead to several physical abnormalities in the fetus, primarily due to intrauterine compression and deformation. In this case, the suspicion of Potter Syndrome was based on clinical manifestations and a history of antepartum oligohydramnios. Therefore, routine antenatal examinations are crucial for detecting pregnancy-related conditions like oligohydramnios and Potter Syndrome, allowing for timely and targeted interventions.

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