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CASE REPORT

Neonatal Polycystic Kidney Disease in a One-Day-Old Baby: A Case Report

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Background: Polycystic kidney disease in neonates is a rare genetic disease which can be either autosomal dominant or autosomal recessive with each presenting at a certain period in life. They can both be diagnosed before or after birth using fetal ultrasound. This is a case of a five-hour-old baby with suspected polycystic kidney disease in a tertiary hospital in northern Tanzania.

Case Presentation: We present a case of a five-hour-old female baby referred to us with a complaint of non-progressive abdominal distension since birth. The birth weight was 2.4 kilograms with a good APGAR score. Clinically, the baby had palpable kidneys bilaterally, widened anterior fontanelle communicating with the posterior as well as rocker bottom feet. Her abdominal ultrasound showed bilaterally enlarged echogenic kidneys with loss of cortico-medullary differentiation and multiple tiny cystic spaces. An echocardiogram showed patent ductus arteriosus and moderate tricuspid regurgitation with mild pulmonary regurgitation. The patient was started on medication but unfortunately on day two post admission the baby succumbed.

Conclusion: Neonatal polycystic kidney disease is associated with high morbidity and mortality rates. It may not be as rare as previously reported. Minimal to no awareness exists on the condition or its effects in our setup due to underdiagnosis and neither availability of neonatal screening nor availability of genetic analysis. It is likely underdiagnosed due to a lack of skills in fetal ultrasounds and no neonatal ICU to care for these babies. Increased awareness will increase the index of suspicion. This is the first case report in our setup highlighting this condition.

Keywords: newborn screening, polycystic kidney disease, ultrasound

Background

Cystic kidneys are a frequent cause of end-stage renal disease in both children and adults. The two main forms of cystic kidney disease are autosomal dominant polycystic kidney disease (ADPKD) and autosomal recessive polycystic kidney disease (ARPKD).[1] ARPKD is rare and commonly manifests in children whereas ADPDK is common and usually presents in adults.[1] Genetic analysis has been shown to be able to improve the management of the condition but the pathogenesis of polycystic kidney disease remains unclear.[1] Cystic kidney diseases are defined by the presence of one or multiple cysts, which are benign lesions contained in a serous fluid-filled sac.[2] These are as a result of certain gene mutations that lead to dysfunctions in the primary cilia of the tubular epithelium.[3,4] The two most important and well-known hereditary cystic diseases are autosomal dominant polycystic disease (ADPKD) and autosomal recessive polycystic disease (ARPKD).[5]

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The main differentiation between these two disorders is related to the time of diagnosis. ADPKD is usually diagnosed during young adulthood, whereas ARPKD is noticed immediately after birth or even before birth.[6]

ARPKD is a severe rare genetic condition, with high mortality rates with an autosomal recessive pattern of transmission similar to most early onset cystic kidney diseases.[7] The mortality rates can reach up to 30% during the neonatal period.[8] The incidence of ARPKD is approximately 1:20,000 live births, but it represents the most important cause of end-stage renal disease that requires renal replacement therapy during early childhood.[9]

We report the case of a five-hour-old female baby who was diagnosed with polycystic kidney disease to underline the difficulties in early diagnosis and management of the condition.

Case Presentation

A five-hour-old female baby was referred to our centre due to non-progressive abdominal distension since birth. This baby was born by caesarean section at term. The reason for C/section is unknown as the mother was unavailable at the time of admission. Birth weight at delivery was 2.4 kilograms and the APGAR score was 8 and 9 in the 1st and 5th minutes respectively. Antenatal history was uneventful and ultrasound done during pregnancy was normal. This is the first-born child to both parents. There are no reported cases of renal or malformations in the family.

Examination on admission showed the baby was sick looking, in respiratory distress and pink on oxygen therapy saturating at 97%. Her temperature was 36.9 °C, respiratory rate was 58 breaths/minute and Silverman Anderson score was 7. Her random blood glucose on admission was 5.8 mmol/L. In her respiratory system examination she had marked lower chest indrawing, nasal flaring and grunting with coarse crepitations throughout. Her abdomen was distended, soft with palpable kidneys bilaterally and the digital rectal exam was normal. On her central nervous system examination, she had a wide anterior fontanelle communicating with the posterior fontanelle with present reflexes. She had rocker bottom feet on musculo-skeletal examination.

Her abdominal pelvic ultrasound showed bilaterally enlarged echogenic kidneys with loss of corticomedullary differentiation and multiple tiny cystic spaces. The right kidney was 8 x 3.3 cm and the left kidney was 7.4 x 3 cm. The rest of the findings were unremarkable. An echocardiogram showed patent ductus arteriosus and moderate tricuspid regurgitation with mild pulmonary regurgitation. Her blood workup showed a normal serum sodium level of 136.8 mmol/L, potassium of 5.6 mmol/L, urea of 3.74 mmol/L, serum creatinine of 133 µmol/L, AST of 28.3 U/L and ALT of 154.0 U/L, total protein of 58.0 g/L and albumin of 33.93 g/L. Serology for HIV was negative. We reached a diagnosis of polycystic kidney disease, early onset neonatal sepsis and acyanotic congenital heart disease. We started the baby on IV fluids of 10% dextrose 140 mL in 24 hours (we gave 75% of the maintenance fluid due to the underlying cardiac lesion), IV ampicillin 240 mg twice daily and kept the baby on CPAP.

The next day after admission during morning rounds this baby was still very sick looking, with coarse crepitations all over the chest and had passed only 10 mL of urine in the catheter. We gave intravenous furosemide 2 mg once daily. Unfortunately, seven hours later the baby changed condition and stopped breathing. She was resuscitated but it was unsuccessful and death was certified.

Discussion

ARPKD belongs to a group of congenital hepatorenal fibrocystic syndromes and causes renal and liver-related comorbidities in children. The majority of these patients present with enlarged echogenic kidneys and more than 50% progress to end-stage renal disease (ESRD) within the first decade of life, hence requiring kidney transplant.[10] ADPKD is usually late onset, and hence manifests in adulthood. It is characterised by bilateral renal cysts, liver cysts and intracranial aneurysms.[11] It can also be associated with pancreatic cysts, seminal vesicles, abdominal wall hernias and other cardiovascular abnormalities. About half of these individuals present ESRD by the age of 60 years.[11] Our baby likely had ARPKD as opposed to ADPKD based on the time of presentation (ARPKD is early in life versus ADPKD which occurs later on).

Prenatal ultrasound has a major role in the early diagnosis of different malformations, such as polycystic kidney disease. Nevertheless, the accuracy of fetal ultrasound is limited.[12,13] ARPKD can be detected by ultrasonography which has high-resolution probes that will show hyperechoic, increased kidney volume with multiple microcysts within the renal cortex and medulla, with the lack of corticomedullary differentiation.[14] In our case, the mother had only one routine ultrasound during pregnancy which was found to be normal. Ultrasonography remains the most appropriate diagnostic tool for polycystic kidney disease during both pre- and post-natal periods, but alternative choices might be MRI and computed tomography scans.[15] Unfortunately,

our patient was unstable to undergo any of these investigations as they are not portable in our setup and she passed away before we could take them.

Management of these patients usually is dependent on what the baby has. Angiotensin converting enzyme inhibitors, calcium channel blockers, diuretics and beta blockers are the mainstay of treatment when hypertension is detected.[15] Antibiotics are also indicated as these babies have high risk of urinary tract infections.[15] Our patient was given IV ampicillin as well as furosemide. Some of these children do end up developing end stage renal disease requiring renal transplantation[10,16], which in our setup is still an ongoing novelty under design. There is a lack of awareness on the occurrence of this condition, hence it is usually mis- and underdiagnosed.

Conclusion

Neonatal polycystic kidney disease is associated with high morbidity and mortality rates. It may not be as rare as previously reported. It is likely underdiagnosed due to lack of skills in fetal ultrasounds. Availability of good antenatal care with early ultrasound detection may improve case detection with early and timely referral of neonates. The prognosis of this condition is unpredictable due to its range of complications. Newborn screening is lacking in our setting, but this could increase case detection resulting in early intervention and monitoring. Minimal to no awareness exists on the condition or its effects in our setup due to underdiagnosis and unavailability of neonatal screening nor the availability of genetic analysis to classify if its ARPKD or ADPKD. Management of such critically ill neonates is also a challenge as we do not have a neonatal ICU in our centre.

Abbreviations

ARPKD, autosomal recessive polycystic kidney disease; ADPKD, autosomal dominant polycystic kidney disease; ICU, intensive care unit.

Data Sharing Statement

All data and materials pertaining to this case report can be made available on request.

Ethics Approval and Consent to Participate

Written informed consent was obtained from the patient's parents for writing of this case report; additionally, accompanying images have been censored to ensure that the patient cannot be identified. A copy of the consent is available on record. Approval to publish was obtained from relevant authorities.

Consent for Publication

Written informed consent was obtained from the patient's parents for publication for this case report.

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Author Contributions

All authors made a significant contribution to the work reported, whether that is in the conception, study design, execution, acquisition of data and interpretation, or in all these areas; took part in drafting, revising or critically reviewing the article; gave final approval of the version to be published; have agreed on the journal to which the article has been submitted; and agree to be accountable for all aspects of the work.

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Disclosure

The authors declare they have no competing interests. All authors of the manuscript have read and agreed to its contents.

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