Prenatal presentations of autosomal dominant polycystic kidney disease (ADPKD): a case report

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Objective – background
Autosomal dominant polycystic kidney disease (ADPKD) is the most common inherited renal cystic disease. The typical age of clinical onset is in the third to fifth decade of life, rarely (<1 percent of cases) ADPKD can present in utero or in the neonatal period with ultrasound changes similar to those seen in autosomal recessive polycystic kidney disease (ARPKD).

Methods
Evaluation of prenatal diagnostic methods (ultrasound, MRI, fetal karyotype) and fetal outcome in a rare prenatal form of ADPKD.

Results
• Prenatal: Massive cystic enlargement of the kidneys was diagnosed during the ultrasound examination of the fetus in the 18th gestational week (pict.1).

The ultrasound image was atypical, therefore various forms of renal cystic disease were considered. MRI examination was performed and normal fetal karyotype was obtained by amniocentesis. ADPKD was given as the most probable diagnosis thanks to a family history (father and paternal grandmother both manifested ADPKD in adult age) by cooperation of geneticists, gynecologists and nephrologists. Despite the uncertain prognosis, mother decided to continue with the pregnancy. The ultrasound assessment was performed in 2-3 week intervals (pict.2-5).

Enlargement progressed with kidneys measuring 87x53mm and 92x57mm respectively by the 35th gestational week, both being hyperechogenic and showing cystic changes.

Childbirth: In 38th gestational week, girl with a birth weight of 3200g and Apgar score of 9-9-9 was delivered through a cesarean section due to patient’s history of two prior cesarean sections.

Postnatal: In the presence of elevated diaphragm, progressive dyspnea with oxygen saturation at 80% developed after birth and required oxygen therapy. Postnatal examination – tab. 1

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<td>R kidney</td>
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Nephrologist was consulted because of gradual onset of hypertension and antihypertensive medication (Nifedipine) was given. ADPKD with very early prenatal manifestation was established as definitive diagnosis.

Follow-up: last examination was performed in 2 months of age (weight 4270g). Ultrasound and MRI images didn’t show significant progression, hypertension was persisting with blood pressure levels between 117/72 mm Hg and TK 135/90 mm Hg (both percentile 110/70 mm Hg).

Serum creatinine was 0.31 mg/dL, with mild proteinuria. Medication was raised to Nifedipine 2 mg b.i.d.

Results of ongoing genetic testing point towards PKD1 gene mutation with possibility of combined PKD1 and PKHD1 gene mutations.

Conclusion
Rare prenatal form of ADPKD is associated with more rapid disease progression after birth and prognosis of the disease is uncertain. In prena tally diagnosed cases, termination of pregnancy might be discussed with parents. When decision to continue with the pregnancy is made, both prenatal multidisciplinary approach and watchful postnatal follow-up are essential.