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Autosomal Recessive Dominant Polycystic Kidney Disease: A Case Report and Review of The Literature

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Introduction:-

Autosomal recessive polycystic kidney disease is a serious, usually early-onset condition.
It mainly affects the kidneys and biliary tract.
It is associated with high neonatal morbidity and mortality.

Observation:

Our 31-year-old patient, with no notable pathological antecedents, in particular no family history of renal anomalies, G2P1 (a child with good psychomotor development), consulted for a pregnancy follow-up. The course of the pregnancy was unremarkable, and the gestational age was estimated at 27 SA.
Obstetrical ultrasound revealed the following on sagittal section: large homogeneous hyperechoic kidneys (+4 D.S), with loss of cortico-medullary differentiation, small cysts in the medullary region, bladder not visualized, with oligohydramnios.
Morphological ultrasound showed no associated fetal malformation, so the diagnosis of autosomal recessive polycystic kidney disease was suspected. Ultrasound monitoring was performed every 3 weeks, showing decrease in the quantity of amniotic fluid.

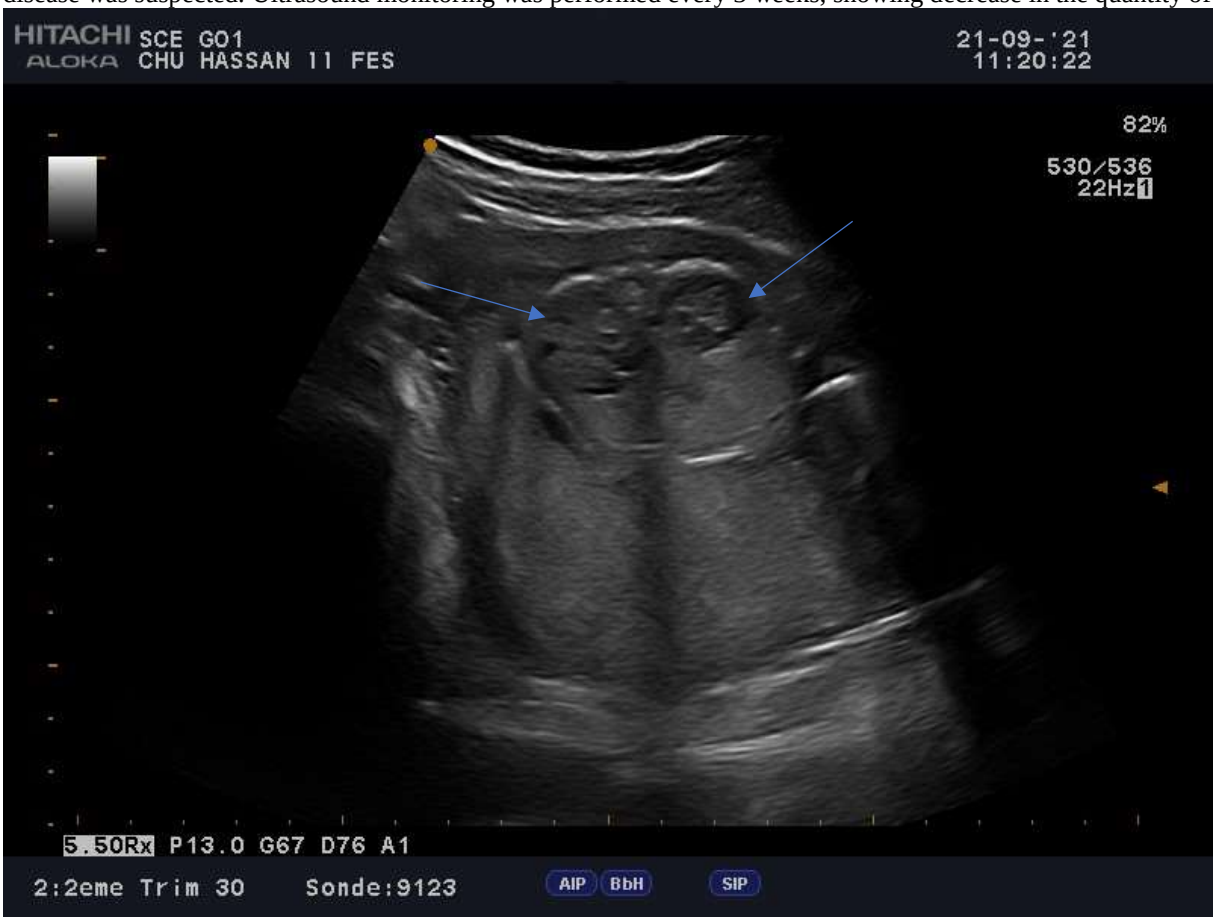

Figure 1: Axial section: the kidneys are enlarged and hyperechoic due to microcysts.

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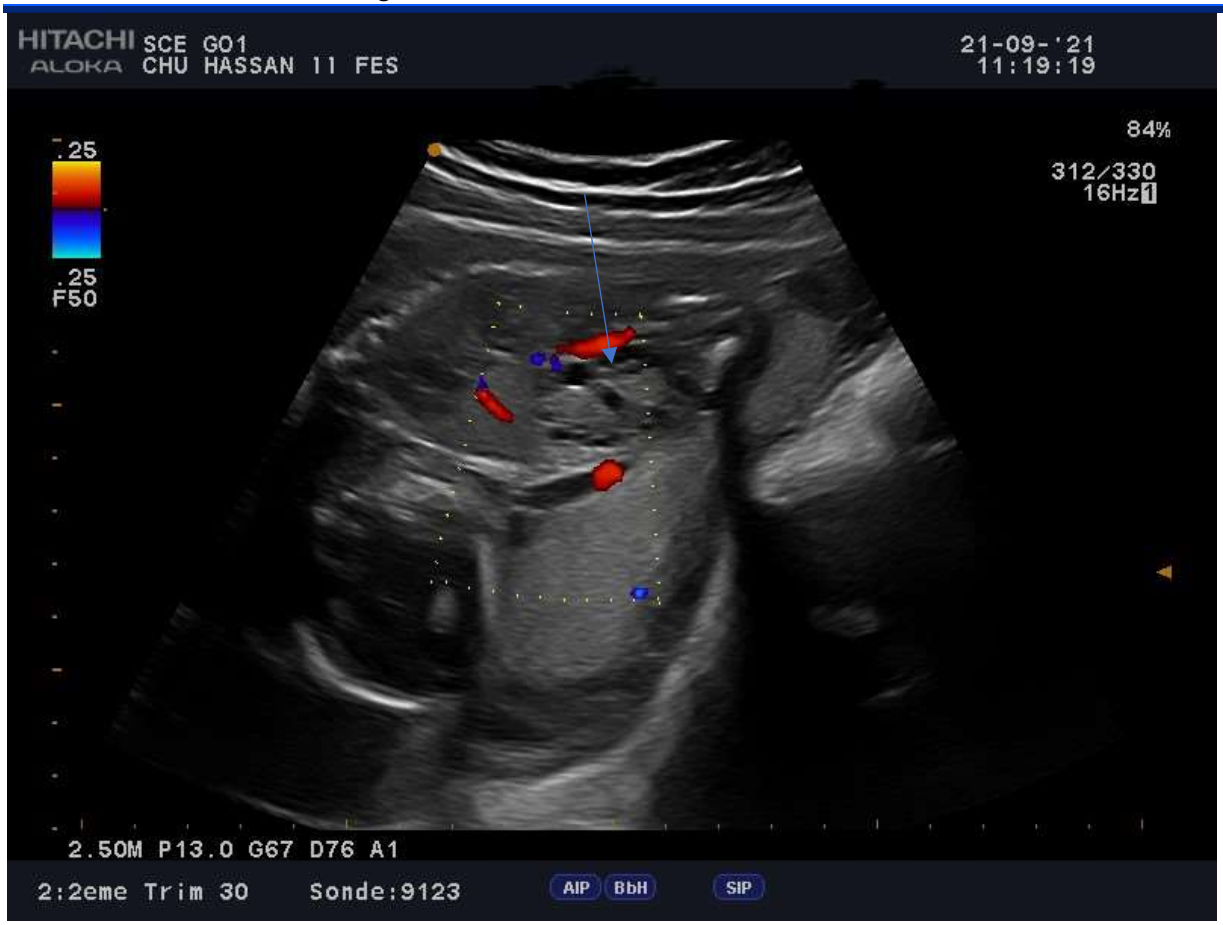

Figure 2:sagittal section; large hyperechoic kidney with loss of cortco-medullary differentiation.

Discussion:-

Autosomal recessive polycystic kidney disease (PKD) is a genetic disorder, with mutation usually identified in the PKHD1 gene (86 exons, located on chromosome 6). [1]
Its incidence is 1/ 20,000 live births. [1]
Anatomopathology. It is defined by the association of two very enlarged kidneys, with cystic dilatation of the collecting tubes in a radiated corticomedullary arrangement; there are no glomerular cysts, but hepatic involvement with periportal fibrosis and ectasia of the intrahepatic bile ducts[1].
Diagnosis is based on the existence of a family history compatible with autosomal recessive transmission, confirmed by the absence of renal cysts in both parents. Parental consanguinity is a further argument in favor. Transmission is autosomal recessive. Parents who do not have the disease can pass it on to their offspring, provided both carry the abnormal gene and both pass it on to their child.
The diagnosis can be made as early as 14 weeks' amenorrhea (SA), when the kidneys are bilaterally enlarged, hyperechoic, and the kidneys are in the centre of the uterus.
That the amount of amniotic fluid may still be normal and the bladder visible [2].
Diagnosis is usually made in the second trimester, with large antenatal kidney size (+5 to +15 DS) and medullary or diffuse hyperechogenicity. Cortico-medullary differentiation is absent, with medullary cysts frequently observed (30% of cases). The bladder is never visible [3].
A rarer but more characteristic ultrasonographic appearance is that of hyperechoic but only slightly enlarged kidneys, with reversed corticomedullary differentiation due to involvement limited to the medullary region. Liver damage has not been demonstrated in utero. Oligohydramnios is moderate to severe, and increases during pregnancy.
justified, with a risk of early renal failure [1].
Amniocentesis should be considered to study the fetal karyotype and polymerase chain reaction to identify CMV infection, which can manifest itself antenatally as large hyperechogenic kidneys. [4].

However, in the case of early prenatal diagnosis using molecular genetics, it is impossible to predict the evolution of kidney size and the impact of the disease on the quantity of amniotic fluid.
some forms remain stable, others evolve unfavorably, sometimes early, sometimes late [5].
Once the

Conclusion:
Ultrasound diagnosis of recessive polycystic kidney disease is late and unreliable.
Molecular biology enables earlier diagnosis.
Multidisciplinary prenatal care, involving maternal-fetal medicine, neonatology, pediatrics and nephrology, is essential to manage the disease.

Conflicts of interest:
The authors declare no conflicts of interest.

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