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**AUTOSOMAL DOMINANT POLYCYSTIC KIDNEY DISEASE (ADPKD)**

ADPKD can be diagnosed prenatally or at a very young age. Any diagnostic tests done during pregnancy should be done along with medical counseling so the results of the test can be completely understood. The number of cysts a child has affects his/her signs and symptoms. Just as in adults, children with many cysts are more likely to have back, side or stomach pain and are also more likely to have high blood pressure than children with only a few cysts. Almost all children who are diagnosed after the first year of life have perfectly normal kidney function that seems to stay normal throughout childhood. Most children will maintain normal kidney function at least until they are into their mid-20s.

**Genetics**

When a parent has ADPKD, every conception carries a 50 percent chance of the child inheriting the mutated gene and having ADPKD. In ADPKD, only one mutated PKD gene of the pair is required to pass on the disease.

**Diagnosis and Screening**

Diagnosis is most often made through radiology, such as an ultrasound, CT scan or MRI.

**Characteristics of children diagnosed in first year of life:**
- Most of these children have brothers and/or sisters who are also diagnosed in the first year of life.
- Most are diagnosed before birth with large kidneys, but often they do not have actual cysts.
- Most develop high blood pressure in childhood.

**Characteristics of children diagnosed after one year of age:**
- Just as likely to have an affected father as an affected mother.
- Likely to have cysts, even though their kidneys are not necessarily enlarged.
- Affected with only one cyst (in half the children), or just a few cysts. Whereas, in an adult one cyst alone is not enough to diagnose ADPKD, in children who are part of an ADPKD family, even one cyst means they are more likely to have the disease.

**Symptoms of ADPKD**

- High blood pressure
- Frequent urinary tract infections
- Blood in urine
- Mitral valve prolapse
- Hernia
- Back/Flank pain
- Kidney stones
- Proteinuria (excess protein in urine)
- Enlarged kidneys found during abdominal exam
- Depression and anxiety (due to stress and emotional impact)

**Health Management in ADPKD**

- Live a healthy life-style, watching diet and including moderate exercise
- Monitor blood pressure, controlling it with medication if it is elevated
- Avoid too much salt, it can increase blood pressure
- Limit caffeine intake

**AUTOSOMAL RECESSIVE POLYCYSTIC KIDNEY DISEASE (ARPKD)**

ARPKD is a relatively rare form of PKD, affecting approximately 1 in 20,000 children. ARPKD often causes death in the first month of life. For the 70% of ARPKD children who survive the newborn period, approximately one-third will need dialysis or transplantation by age 10. Previously thought to be a fatal condition, the prognosis for children with ARPKD has improved dramatically. Twenty years ago, only half of the children born with the disease survived to their 10th birthday, but now that percentage has increased to 85 percent.

**Genetics**

Each conception carries a 25 percent chance of the child inheriting both mutated genes and having ARPKD. In recessive disorders such as ARPKD, the child must inherit a copy of the disease gene from both parents in order to be affected. Since the parents each have only one copy of the disease gene, they do not have the disease and are referred to as “carriers.” With each conception, there is a 50 percent chance the child will be a carrier and a 25 percent chance there will be no mutated genes inherited.

**Diagnosis**

Typically in ARPKD, the kidneys appear to be larger than normal. In some babies, prenatal ultrasound can detect the enlarged kidneys as early as 18 weeks after conception. After birth, the kidneys may be large enough to examine by touch or through an ultrasound. Prenatal genetic testing is possible using samples from either chorionic villus sampling or amniocentesis. These genetic tests can either involve a direct search of the gene for mutations or an indirect association using linkage analysis. For linkage analysis, DNA samples are required from the fetus, a previously affected child, and the parents.

**Symptoms of ARPKD**

- Diminished amniotic fluid levels during pregnancy
- Enlarged kidneys on fetal ultrasound
- Lung immaturity and functioning issues

**Immediately after birth**

- Enlarged kidneys due to cysts
- Breathing problems due to lack of space because of enlarged kidneys and decreased urine production. Ventilation is frequently required to sustain life.
- Excessive urine production
- Hypertension
- Growth problems
- Congenital hepatic fibrosis
- Enlarged spleen with low red blood cell, white blood cell and platelet counts

**Health Management in ARPKD**

Families should work with specialists in several areas, including liver, kidney and lung specialists, to monitor the child’s health.
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**Screening**
- Ultrasound
- CT scan
- MRI
- MR Cholangiography (x-ray examination of the bile ducts)

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PKD Foundation Support
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