Condition: Polycystic kidney disease

Inheritance:

Autosomal dominant or autosomal recessive.

Genetic etiology:

The autosomal dominant form is due to mutation in PKD1 (85%) or *PKD2,* which encode polycystin 1 and polycystin 2, respectively. The autosomal recessive form is due to mutation in the *PKHD1* gene.

Frequency:

Autosomal dominant: 1/400 – 1/1,000; Autosomal recessive: 1/20,000 – 1/40,000.

Clinical features:

Individuals with autosomal dominant polycystic kidney disease develop renal cysts, a renal concentrating defect, and hypertension. End-stage renal disease is common by late adulthood. Other manifestations include liver and pancreatic cysts, arterial aneurysms, including cerebral artery aneurysms, and mitral valve prolapse. Autosomal recessive polycystic kidney disease is characterized by congenital polycystic kidneys and hepatic fibrosis. There is early onset of renal failure and hypertension, and progressive liver disease. Most children have pulmonary hypoplasia as a result of oligohydramnios.

Management:

Surveillance and symptomatic treatment.

Genetic counseling:

Based on mode of inheritance; genetic testing is available and renal imaging can identify individuals at risk with autosomal dominant form.

Human Genetics and Genomics, Fourth Edition. Bruce R. Korf and Mira B. Irons.