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Phenotypic Variability in Siblings with Autosomal Recessive Polycystic Kidney Disease

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DESCRIPTION

Polycystic renal disorder (PKD) is an inherited disease that causes clusters of cysts, primarily within the kidneys, that over time cause enlargement and loss of function. Cysts vary in size and are often very large. Polycystic renal disorder can also cause cysts in the liver and other parts of the body. The disease can cause serious complications such as high blood pressure and kidney failure. A mutation causes polycystic kidney disease. In most cases, parents pass their disability on to their children. In other words, it inherits PKD. But sometimes genes mutate or change randomly. People of all ages, races, and ethnicities can develop PKD. It occurs equally in women and men. Polycystic renal disorder (PKD) is hereditary and causes cysts to form in the kidneys. Cysts can enlarge the kidneys and disrupt kidney function. Most PKD patients are adults. However, in rare cases, babies have a dangerous sort of PKD. Treatment relieves symptoms and improves kidney function. Most PKD patients require dialysis or kidney transplantation. There are two sorts of PKD: Autosomal dominant PKD and autosomal recessive PKD. Autosomal dominant PKD causes cysts only within the kidney. People with this sort of PKD are often referred to as "adult PKD" because they may not notice symptoms until they are in their 30's to 50's. In autosomal recessive PKD, cysts grow in both the kidney and liver. Autosomal recessive PKD is usually referred to as infantile PKD. This is often because infants may show signs of the disease during the first few months of life or before birth. PKD can affect organs aside from the kidneys. PKD patients may have cysts within the liver, pancreas, spleen, ovaries, and colon. Cysts in these organs don't usually cause serious problems, but they will occur in some people. PKD also can affect the brain and heart. When PKD affects the brain, it can cause an aneurysm. An aneurysm may be a bulging blood vessel that can burst, resulting in stroke or even death. When PKD affects the guts, it can loosen valves and cause heart murmurs in some patients. Ultrasound is that the most reliable, inexpensive, and non-invasive method of diagnosing PKD. If someone in danger for PKD is over 40 and has a normal kidney ultrasound, he probably doesn't have PKD. Computerized tomography (CT) scans and magnetic resonance imaging (MRI) may detect small cysts that cannot be seen with ultrasound. In some circumstances, genetic testing can also be done. This includes blood tests that search for abnormal genes that cause disease. There is no cure for this disease. Treatment focuses on preventing complications and relieving symptoms. Drugs are used to control high blood pressure and treat urinary tract infections. Growth hormone can be used to promote growth. As kidney failure progresses, dialysis and a transplant are required.

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CONFLICT OF INTEREST

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