Voice of the Patient report Externally-Led Patient-Focused Drug Development (EL-PFDD) Meeting for ARPKD Patients and their Families

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Clinical Summary

Autosomal Recessive Polycystic Kidney Disease (ARPKD) is a rare genetic disease. It occurs in about one in 20,000 births, with an estimated 1,500 children and young adults living with ARPKD in the United States. ARPKD can be caused by many different gene variants. The majority of patients with ARPKD have variations in the PKHD1 gene, which encodes the fibrocystin protein, but variants in other genes can also be involved as well. The type of gene variant influences the severity of kidney and liver involvement; null gene variations, which are those that lead to the total loss of gene/protein function, are associated with worse kidney survival and earlier liver-associated complications. ARPKD is a progressive disease that affects the kidneys as well as the liver. The severity of both liver and kidney symptoms are highly variable and there is not a clear relationship between the severity of kidney and liver disease. ARPKD is a highly variable disease that can present any time from before birth to adulthood. Two-thirds of patients are diagnosed prenatally or in infancy, and the remaining one-third are diagnosed in childhood or adolescence.

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