NephU™ Understanding Polycystic Kidney Disease (PKD)

PKD includes a group of hereditary disorders that originates from a single gene. Symptoms include renal cysts, enlarged kidneys, and kidney failure. [1]

ARPKD

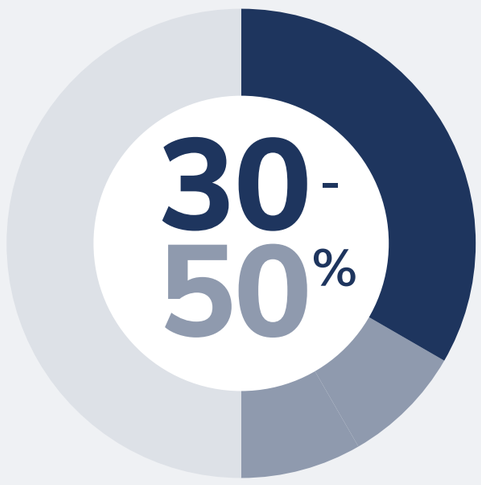
Autosomal recessive polycystic kidney disease

1 in **20,000** live births[1-3]

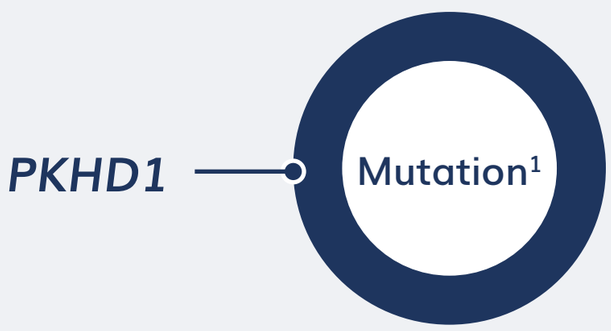
ARPKD can be a cause of kidney failure in children.

children

ARPKD is typically detected before birth.[1,2]



of affected newborns die shortly after birth[3]



Syndromic Cystic Disorders Are Considered Rare Diseases

Prevalence[1]: **Rare**

Syndromic cystic disorders are rare inherited diseases that have renal cysts as part of their observable traits[1]

Includes[1]:

Nephronophthisis:

Symptoms include inflammation and scarring that impairs kidney function leading to increased urine production, excessive thirst, weakness, and fatigue.

Joubert syndrome and related disorders
Meckel syndrome
Bardet-Biedl syndrome
Oro-facial-digital syndrome

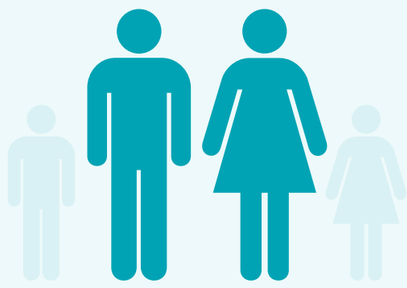
Associated genes

ADPKD

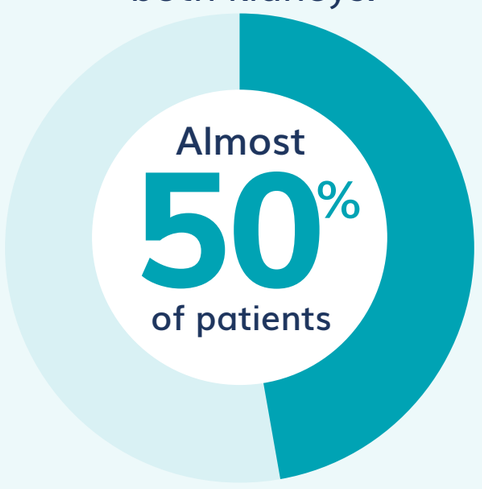
Autosomal dominant polycystic kidney disease

1 in **400–1000** people[1]

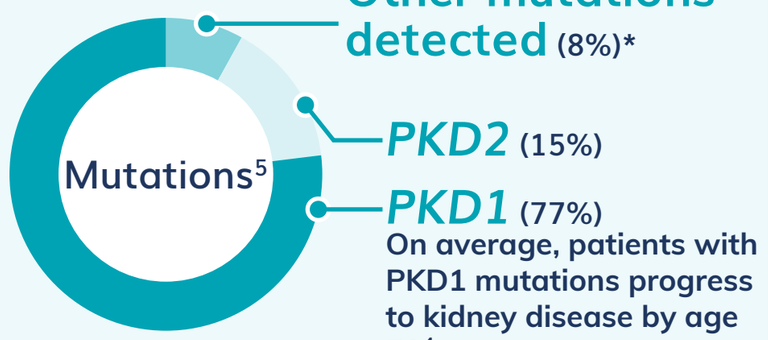
ADPKD is typically diagnosed in adults



ADPKD is characterized by development and progressive enlargement of fluid-filled cysts in both kidneys.



who have ADPKD develop Kidney Failure by age 60[4]



On average, patients with PKD1 mutations progress to kidney disease by age 58[4]

*Based on data from the HALT Progression of PKD Study and The Consortium of Radiologic Imaging of PKD Study (N=1119 patients, 945 families).

References

1. Harris PC, Torres VE. *Annu Rev Med*. 2009;60:321-337. 2. Hartung EA, Guay-Woodford LM. *Pediatrics*. 2014;134(3):e833-e845. 3. Bergmann C. *Nephron*. 2019;141:50-60. 4. Chebib FT, Torres VE. *Am J Kidney Dis*. 2016;67(5):792-810. 5. Heyer CM et al. *JASN*. 2016;27:2872-2884.

Abbreviations

kidney disease = end-stage renal disease; *PKD1*, polycystic kidney disease 1 gene; *PKD2*, polycystic kidney disease 2 gene; *PKHD1*, polycystic kidney and hepatic disease 1 gene.

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