

Disease

Polycystic kidney disease 1^{1, 2, 3}

Gene

PKD1

Inheritance	AR and AD
Current Treatments	Tolvaptan
Evidence for Treatment	guideline ⁴
Non-Molecular Testing Availability	yes
Non-Molecular Test	renal ultrasound ⁵
ClinGen	https://actionability.clinicalgenome.org/ac/Adult/ui/stg2SummaryRpt?doc=AC037
Disease Frequency (per 100,000)	63.5 ^{2, 6}
Genes Included in Disease Frequency	PKD1, PKD2
Experimental / Investigational Therapies	https://clinicaltrials.gov/ct2/results?cond=Polycystic+Kidney+Diseases&term=&cntry=&state=&city=&dist=

References

- ¹ <https://www.ncbi.nlm.nih.gov/pubmed/31692482>
- ² <https://www.ncbi.nlm.nih.gov/books/NBK1246/#pkd-ad.Summary>
- ³ <https://pubmed.ncbi.nlm.nih.gov/23624871/>
- ⁴ <https://pubmed.ncbi.nlm.nih.gov/31118499/>
- ⁵ <https://www.omim.org/entry/173900>
- ⁶ <https://www.ncbi.nlm.nih.gov/pubmed/29338003>