Resource Summary Report

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Autosomal Recessive Polycystic Kidney Disease Mutation Database

RRID:SCR_002290
Type: Tool

Proper Citation

Autosomal Recessive Polycystic Kidney Disease Mutation Database (RRID:SCR_002290)

Resource Information

URL: http://www.humgen.rwth-aachen.de/

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Description: Catalog of all changes detected in PKHD1 (Polycystic Kidney and Hepatic Disease 1) in a locus specific database. Investigators are invited to submit their novel data to this database. These data should be meaningful for clinical practice as well as of relevance for the reader interested in molecular aspects of polycystic kidney disease (PKD). There are also some links and information for ARPKD patients and their parents. Autosomal recessive polycystic kidney disease (ARPKD/PKHD1) is an important cause of renal-related and liver-related morbidity and mortality in childhood. This study reports mutation screening in 90 ARPKD patients and identifies mutations in 110 alleles making up a detection rate of 61%. Thirty-four of the detected mutations have not been reported previously. Two underlying mutations in 40 patients and one mutation in 30 cases are disclosed, and no mutation was detected on the remaining chromosomes. Mutations were found to be scattered throughout the gene without evidence of clustering at specific sites. PKHD1 mutation analysis is a powerful tool to establish the molecular cause of ARPKD in a given family. Direct identification of mutations allows an unequivocal diagnosis and accurate genetic counseling even in families displaying diagnostic challenges.

Synonyms: Mutation Database Autosomal Recessive Polycystic Kidney Disease (ARPKD/PKHD1)

Resource Type: data or information resource, service resource, database, storage service resource, data repository

Defining Citation: PMID:16199545, PMID:11919560

Keywords: clinical, gene, genetic, mutation, protein, recessive, renal

Related Condition: Autosomal recessive polycystic kidney disease, Polycystic kidney disease

Funding:

Availability: Permission required, Terms of use

Resource Name: Autosomal Recessive Polycystic Kidney Disease Mutation Database

Resource ID: SCR_002290

Alternate IDs: nif-0000-21038

Old URLs: http://www.humgen.rwth-aachen.de/index.asp?subform=database.html&nav=database_nav.html

Record Creation Time: 20220129T080212+0000

Record Last Update: 20250521T060833+0000

Ratings and Alerts

No rating or validation information has been found for Autosomal Recessive Polycystic Kidney Disease Mutation Database.

No alerts have been found for Autosomal Recessive Polycystic Kidney Disease Mutation Database.

Data and Source Information

Source: SciCrunch Registry

Usage and Citation Metrics

We found 14 mentions in open access literature.

Listed below are recent publications. The full list is available at NIF.

Yang C, et al. (2023) Pkhd1cyli/cyli mice have altered renal Pkhd1 mRNA processing and hormonally sensitive liver disease. Journal of molecular medicine (Berlin, Germany), 101(9), 1141.

Coovadia A, et al. (2023) A taxonomic schema of potential pitfalls in clinical variant analysis based on real-world evidence. PloS one, 18(11), e0295010.

Giacobbe C, et al. (2022) Rare variants in PKHD1 associated with Caroli syndrome: Two case reports. Molecular genetics & genomic medicine, 10(8), e1998.

Jung J, et al. (2020) Fatal outcome of autosomal recessive polycystic kidney disease in neonates with recessive PKHD1 mutations. Medicine, 99(19), e20113.

Obeidova L, et al. (2020) Results of targeted next-generation sequencing in children with cystic kidney diseases often change the clinical diagnosis. PloS one, 15(6), e0235071.

Wang J, et al. (2019) Novel compound heterozygous PKHD1 mutations cause autosomal recessive polycystic kidney disease in a Han Chinese family. Molecular medicine reports, 20(6), 5059.

Al Alawi I, et al. (2019) Molecular Genetic Diagnosis of Omani Patients With Inherited Cystic Kidney Disease. Kidney international reports, 4(12), 1751.

Obeidova L, et al. (2015) Molecular genetic analysis of PKHD1 by next-generation sequencing in Czech families with autosomal recessive polycystic kidney disease. BMC medical genetics, 16, 116.

Eisenberger T, et al. (2015) An efficient and comprehensive strategy for genetic diagnostics of polycystic kidney disease. PloS one, 10(2), e0116680.

Tavira B, et al. (2015) A labor and cost effective next generation sequencing of PKHD1 in autosomal recessive polycystic kidney disease patients. Gene, 561(1), 165.

Xu Y, et al. (2014) A novel mutation identified in PKHD1 by targeted exome sequencing: guiding prenatal diagnosis for an ARPKD family. Gene, 551(1), 33.

Gunay-Aygun M, et al. (2010) PKHD1 sequence variations in 78 children and adults with autosomal recessive polycystic kidney disease and congenital hepatic fibrosis. Molecular genetics and metabolism, 99(2), 160.

Denamur E, et al. (2010) Genotype-phenotype correlations in fetuses and neonates with autosomal recessive polycystic kidney disease. Kidney international, 77(4), 350.

Bergmann C, et al. (2005) Clinical consequences of PKHD1 mutations in 164 patients with autosomal-recessive polycystic kidney disease (ARPKD). Kidney international, 67(3), 829.