

Evidence Summary

SEC63 – isolated polycystic liver disease – Autosomal dominant inheritance

Classification owner: Kidney Cystic and Ciliopathy Disorders
Calculated classification: Definitive
Modified classification: No Modification
Reason for modified classification: None

SOP: [Gene Clinical Validity Standard Operating Procedures \(SOP\), Version 7](#)

Classification status: APPROVED PUBLISHED
Date classification saved: 2020 Oct 06, 8:15 am
Date classification published: 2020 Oct 06, 8:16 am
Replication Over Time: Yes
Contradictory Evidence? Proband: **No**, Experimental: **No**
Disease: [isolated polycystic liver disease](#)

Evidence Summary

SEC63 was FIRST reported in relation to autosomal dominant polycystic liver disease (ADPLD) in 2004 (Davila et al., PMID 15133510). ADPLD due to SEC63 mutations (ADPLD-SEC63) is diagnosed in adults with a family history of liver cysts, under age 40 with any number of liver cysts and individuals over the age of 40 with four or more liver cysts. Women are associated with more severe course of disease, and kidney cysts are present in approximately 28-35% of cases, but in these cases the kidney disease is mild with no association with progression to end stage renal disease (PMID 20408995). At least twenty unique variants (e.g. nonsense, frameshift, splice site, missense) have been reported in humans. Evidence supporting this gene-disease relationship includes case-level data, segregation data, population data and experimental data. Summary of Case Level Data: 12 POINTS. Variants in this gene have been reported in at least 19 probands and in at least four publications (PMID 15133510, PMID 16835903, PMID 20095989, PMID 28375157). Variants in this gene segregated with disease in at least 24 additional family members. More evidence is available in the literature, but the maximum score for genetic evidence (12 pts.) has been reached. The mechanism for disease is heterozygous loss of function (PMID 15133510, PMID 20095989), with experimental evidence suggesting endoplasmic reticulum stress with induction of the unfolded protein response (UPR) and association with aberrant polycystin 1 (PC1) post-translational modifications leading to cystogenesis (PMID 22864019, PMID 25844898). This gene-disease association is supported by expression studies in cell models, zebrafish models, and mouse models (PMID 21685914, PMID 22864019, PMID 25844898). In summary, SEC63 is definitively associated with AUTOSOMAL DOMINANT POLYCYSTIC LIVER DISEASE. This has been repeatedly demonstrated in both the research and clinical diagnostic settings, and has been upheld over time. This classification was approved by the ClinGen Cystic and Ciliopathy Disorders GCEP on 09/23/2020 (SOP Version 7).

Calculated Classification Matrix

Evidence Type				Count	Total Points	Points Counted	
Genetic Evidence	Case-Level	Variant	Autosomal Dominant OR X-linked Disorder	Proband with other variant type with some evidence of gene impact	7	1.35	1.35
				Proband with predicted or proven null variant	12	9.5	9.5
				Variant is <i>de novo</i>	0	0	0
		Autosomal Recessive Disorder	Two variants (not predicted/proven null) with some evidence of gene impact in <i>trans</i>	0	0	0	
			Two variants in <i>trans</i> and at least one <i>de novo</i> or a predicted/proven null variant	0	0		
			Segregation			Summed LOD	Family Count
	Candidate gene sequencing			6.32	4		
	Exome/genome or all genes sequenced in linkage region			0	0		
	Total Summed LOD Score			6.32			
	Case-Control				0	0	0
Genetic Evidence Total						12	
Experimental Evidence	Functional	Biochemical Functions			0	0	0
		Protein Interactions			0	0	
		Expression			0	0	
	Functional Alteration	Patient cells			0	0	0
		Non-patient cells			0	0	
	Models	Non-human model organism			2	3	4.5
		Cell culture model			0	0	
	Rescue	Rescue in human			0	0	
		Rescue in non-human model organism			1	1	
		Rescue in cell culture model			1	0.5	
Rescue in patient cells			0	0			
Experimental Evidence Total						4	
Total Points						16	

Genetic Evidence: Case Level (variants, segregation)

Label	Variant Type ^	Variant	Reference (PMID) ↕	Proband Sex	Proband Age	Proband Ethnicity	Proband Phenotypes	Segregations				Proband Previous Testing	Proband Methods of Detection	Score Status	Proband Points (default points)	Reason for Changed Score
								# Aff	# Unaff	LOD Score	Counted					

Segregations

Label	Variant Type ^	Variant	Reference (PMID) ⇅	Proband Sex	Proband Age	Proband Ethnicity	Proband Phenotypes	Segregations					Proband Previous Testing	Proband Methods of Detection	Score Status	Proband Points (default points)	Reason for Changed Score
								# Aff	# Unaff	LOD Score	Counted	Sequencing					
F-226	Proband with other variant type with some evidence of gene impact		Davila S, et al., 2004 , PMID: 15133510	Unknown			HPO term(s): <ul style="list-style-type: none"> Autosomal dominant inheritance Polycystic liver disease Free text: Polycystic liver disease	-	-	-	-			Score	0 (0.5)	Variant has conflicting reports in ClinVar (Pathogenic via literature, Uncertain Significance via Mendelics, Benign via Illumina). Found in additional 4 individuals (PMID 20095989). Has 0.3% (1063/282096 alleles in gnomAD) occurrence in general population which is higher than expected for a pathogenic rare disease variant.	
c.502G>C	Proband with other variant type with some evidence of gene impact		Waanders E, et al., 2006 , PMID: 16835903	Unknown			HPO term(s): <ul style="list-style-type: none"> Autosomal dominant inheritance Polycystic liver disease Free text: Patients with 2 or more liver cysts on abdominal imaging or were on/had a liver transplant due to cysts.	-	-	-	-			Score	0.25 (0.5)	No further family data or functional data given, not found in ClinVar with clinical significance	
SEC63 c.359T>C, PRKCSH c.1362-2A>G	Proband with other variant type with some evidence of gene impact		Waanders E, et al., 2010 , PMID: 20095989	Unknown			HPO term(s): <ul style="list-style-type: none"> Polycystic liver disease Autosomal dominant inheritance Free text: A proband was defined as a patient with at least one affected family member. PCLD was diagnosed in probands or isolated individuals when >20 liver cysts were present. In case of a positive family history, patients aged 40 years or younger with any liver cysts or patients above the age of 40 with four or more liver cysts were diagnosed with PCLD.	-	-	-	-			Score	0.1 (0.5)	In silico indicates SEC63 p.Ile120Thr is pathogenic. Also carries PRKCSH c.1362-2A>G well documented in ADPLD in Ductch kindreds PMID (12577059). Unsure of variant impact. No further clinical information is given regarding severity of phenotype.	

								Segregations									
Label	Variant Type ^	Variant	Reference (PMID) ⇅	Proband Sex	Proband Age	Proband Ethnicity	Proband Phenotypes	# Aff	# Unaff	LOD Score	Counted	Sequencing	Proband Previous Testing	Proband Methods of Detection	Score Status	Proband Points (default points)	Reason for Changed Score
c.649C>T	Proband with other variant type with some evidence of gene impact		Waanders E, et al., 2010, PMID: 20095989	Unknown			HPO term(s): <ul style="list-style-type: none"> Polycystic liver disease Autosomal dominant inheritance Free text: A proband was defined as a patient with at least one affected family member. PCLD was diagnosed in probands or isolated individuals when >20 liver cysts were present. In case of a positive family history, patients aged 40 years or younger with any liver cysts or patients above the age of 40 with four or more liver cysts were diagnosed with PCLD.	-	-	-	-			Method 1: Sanger sequencing	Score	0.25 (0.5)	In silico indicates pathogenicity. No family information or additional functional evidence
c.801A>C	Proband with other variant type with some evidence of gene impact		Waanders E, et al., 2010, PMID: 20095989	Unknown			HPO term(s): <ul style="list-style-type: none"> Polycystic liver disease Autosomal dominant inheritance Free text: A proband was defined as a patient with at least one affected family member. PCLD was diagnosed in probands or isolated individuals when >20 liver cysts were present. In case of a positive family history, patients aged 40 years or younger with any liver cysts or patients above the age of 40 with four or more liver cysts were diagnosed with PCLD.	-	-	-	-			Method 1: Sanger sequencing	Score	0.25 (0.5)	In silico indicates pathogenicity. No family information or additional functional evidence.

								Segregations									
Label	Variant Type ^	Variant	Reference (PMID) ⇅	Proband Sex	Proband Age	Proband Ethnicity	Proband Phenotypes	# Aff	# Unaff	LOD Score	Counted	Sequencing	Proband Previous Testing	Proband Methods of Detection	Score Status	Proband Points (default points)	Reason for Changed Score
c.1124A>C	Proband with other variant type with some evidence of gene impact		Waanders E, et al., 2010 , PMID: 20095989	Unknown			HPO term(s): <ul style="list-style-type: none"> Polycystic liver disease Autosomal dominant inheritance Free text: A proband was defined as a patient with at least one affected family member. PCLD was diagnosed in probands or isolated individuals when >20 liver cysts were present. In case of a positive family history, patients aged 40 years or younger with any liver cysts or patients above the age of 40 with four or more liver cysts were diagnosed with PCLD.	-	-	-	-			Method 1: Sanger sequencing	Score	0.25 (0.5)	In silico indicates pathogenicity. No family information or additional functional evidence.
c.1951T>G	Proband with other variant type with some evidence of gene impact		Waanders E, et al., 2010 , PMID: 20095989	Unknown			HPO term(s): <ul style="list-style-type: none"> Polycystic liver disease Autosomal dominant inheritance Free text: A proband was defined as a patient with at least one affected family member. PCLD was diagnosed in probands or isolated individuals when >20 liver cysts were present. In case of a positive family history, patients aged 40 years or younger with any liver cysts or patients above the age of 40 with four or more liver cysts were diagnosed with PCLD.	-	-	-	-			Method 1: Sanger sequencing	Score	0.25 (0.5)	In silico indicates pathogenicity. No family information or additional functional evidence.
A-6 ll:1	Proband with predicted or proven null variant		Davila S, et al., 2004 , PMID: 15133510	Male				7	-	Calculated: 1.81	Yes	Candidate gene sequencing			Score	1 (1.5)	Familial segregation present but no further evidence of gene-disease validation

Segregations

Label	Variant Type ^	Variant	Reference (PMID) ↕	Proband Sex	Proband Age	Proband Ethnicity	Proband Phenotypes	# Aff	# Unaff	LOD Score	Counted	Sequencing	Proband Previous Testing	Proband Methods of Detection	Score Status	Proband Points (default points)	Reason for Changed Score
F-1 II:2	Proband with predicted or proven null variant		Davila S, et al., 2004 , PMID: 15133510	Female			HPO term(s): <ul style="list-style-type: none"> Autosomal dominant inheritance Polycystic liver disease Free text: Polycystic liver disease	5	-	Calculated: 1.2	Yes	Candidate gene sequencing		Method 1: Linkage analysis; Method 2: Genotyping Description of genotyping method: We screened the 21-exon coding sequence and flanking splice sequences of SEC63 by direct sequencing of amplified genomic PCR products (primer sequences on request).	Score	1 (1.5)	Family segregation present and clinical interpretation present for variant
F-4 II:3	Proband with predicted or proven null variant		Davila S, et al., 2004 , PMID: 15133510	Male			HPO term(s): <ul style="list-style-type: none"> Autosomal dominant inheritance Polycystic liver disease Free text: Polycystic liver disease	8	-	Calculated: 2.11	Yes	Candidate gene sequencing		Method 1: Linkage analysis; Method 2: Genotyping Description of genotyping method: We screened the 21-exon coding sequence and flanking splice sequences of SEC63 by direct sequencing of amplified genomic PCR products (primer sequences on request).	Score	1 (1.5)	Family segregation and clinical interpretation available
B-7 II:1	Proband with predicted or proven null variant		Davila S, et al., 2004 , PMID: 15133510	Male	Age of Diagnosis: 50 Years			5	-	Calculated: 1.2	Yes	Candidate gene sequencing		Method 1: Linkage analysis; Method 2: Genotyping Description of genotyping method: We screened the 21-exon coding sequence and flanking splice sequences of SEC63 by direct sequencing of amplified genomic PCR products (primer sequences on request).	Score	1.5 (1.5)	No clinical significance is given with variant information Segregates in family B-7 and 2 other families: PMID 16835903, PMID 20095989 has ADPLD with the same variant

								Segregations									
Label	Variant Type ^	Variant	Reference (PMID) ↕	Proband Sex	Proband Age	Proband Ethnicity	Proband Phenotypes	# Aff	# Unaff	LOD Score	Counted	Sequencing	Proband Previous Testing	Proband Methods of Detection	Score Status	Proband Points (default points)	Reason for Changed Score
27 II:2	Proband with predicted or proven null variant		Davila S, et al., 2004 , PMID: 15133510	Male			HPO term(s): <ul style="list-style-type: none"> Autosomal dominant inheritance Polycystic liver disease Free text: Polycystic liver disease	4	-	Calculated: 0.9	No			Method 1: Linkage analysis; Method 2: Genotyping Description of genotyping method: We screened the 21-exon coding sequence and flanking splice sequences of SEC63 by direct sequencing of amplified genomic PCR products (primer sequences on request).	Score	1 (1.5)	There is familial segregation, but no further data supporting gene-disease relationship
51	Proband with predicted or proven null variant		Davila S, et al., 2004 , PMID: 15133510	Unknown			HPO term(s): <ul style="list-style-type: none"> Autosomal dominant inheritance Polycystic liver disease Free text: Polycystic liver disease	-	-	-	-			Method 1: Linkage analysis; Method 2: Genotyping Description of genotyping method: We screened the 21-exon coding sequence and flanking splice sequences of SEC63 by direct sequencing of amplified genomic PCR products (primer sequences on request).	Score	1 (1.5)	0.5 for proband 51 because of no further information 0.5 for proband 52 because of no further information
52	Proband with predicted or proven null variant		Davila S, et al., 2004 , PMID: 15133510	Unknown			HPO term(s): <ul style="list-style-type: none"> Autosomal dominant inheritance Polycystic liver disease Free text: Polycystic liver disease	-	-	-	-			Method 1: Linkage analysis; Method 2: Genotyping Description of genotyping method: We screened the 21-exon coding sequence and flanking splice sequences of SEC63 by direct sequencing of amplified genomic PCR products (primer sequences on request).	Review	1.5 (1.5)	Variant is already scored with Proband 51
c.1577C>A	Proband with predicted or proven null variant		Waanders E, et al., 2006 , PMID: 16835903	Unknown			HPO term(s): <ul style="list-style-type: none"> Autosomal dominant inheritance Polycystic liver disease Free text: Patients with 2 or more liver cysts on abdominal imaging or were on/had a liver transplant due to cysts.	-	-	-	-			Method 1: Exome sequencing; Method 2: Restriction digest Description of genotyping method: Missense variants were verified using restriction fragment length polymorphism technique.	Score	0.5 (1.5)	No family information or additional functional evidence

								Segregations									
Label	Variant Type ^	Variant	Reference (PMID) ⇅	Proband Sex	Proband Age	Proband Ethnicity	Proband Phenotypes	# Aff	# Unaff	LOD Score	Counted	Sequencing	Proband Previous Testing	Proband Methods of Detection	Score Status	Proband Points (default points)	Reason for Changed Score
3	Proband with predicted or proven null variant		Waanders E, et al., 2006, PMID: 16835903	Unknown			HPO term(s): <ul style="list-style-type: none"> Autosomal dominant inheritance Polycystic liver disease Free text: Patients with 2 or more liver cysts on abdominal imaging or were on/had a liver transplant due to cysts.	-	-	-	-			Method 1: Exome sequencing; Method 2: Restriction digest Description of genotyping method: Missense variants were verified using restriction fragment length polymorphism technique.	Review	1.5 (1.5)	Variant already scored with PMID15133510
c.422delT	Proband with predicted or proven null variant		Waanders E, et al., 2010, PMID: 20095989	Unknown			HPO term(s): <ul style="list-style-type: none"> Polycystic liver disease Autosomal dominant inheritance Free text: A proband was defined as a patient with at least one affected family member. PCLD was diagnosed in probands or isolated individuals when >20 liver cysts were present. In case of a positive family history, patients aged 40 years or younger with any liver cysts or patients above the age of 40 with four or more liver cysts were diagnosed with PCLD.	-	-	-	-			Method 1: Sanger sequencing	Score	0.5 (1.5)	In silico indicates pathogenicity. No family information or additional function evidence.
c.1118_1126del9bp	Proband with predicted or proven null variant		Waanders E, et al., 2010, PMID: 20095989	Unknown			HPO term(s): <ul style="list-style-type: none"> Polycystic liver disease Autosomal dominant inheritance Free text: A proband was defined as a patient with at least one affected family member. PCLD was diagnosed in probands or isolated individuals when >20 liver cysts were present. In case of a positive family history, patients aged 40 years or younger with any liver cysts or patients above the age of 40 with four or more liver cysts were diagnosed with PCLD.	-	-	-	-			Method 1: Sanger sequencing	Score	0.5 (1.5)	In silico indicates pathogenicity. No family information or additional functional evidence.

								Segregations									
Label	Variant Type ^	Variant	Reference (PMID) ⇅	Proband Sex	Proband Age	Proband Ethnicity	Proband Phenotypes	# Aff	# Unaff	LOD Score	Counted	Sequencing	Proband Previous Testing	Proband Methods of Detection	Score Status	Proband Points (default points)	Reason for Changed Score
c.1813_1817delCAAAA	Proband with predicted or proven null variant		Waanders E, et al., 2010 , PMID: 20095989	Unknown			HPO term(s): <ul style="list-style-type: none"> Polycystic liver disease Autosomal dominant inheritance Free text: A proband was defined as a patient with at least one affected family member. PCLD was diagnosed in probands or isolated individuals when >20 liver cysts were present. In case of a positive family history, patients aged 40 years or younger with any liver cysts or patients above the age of 40 with four or more liver cysts were diagnosed with PCLD.	-	-	-	-			Method 1: Sanger sequencing	Score	0.5 (1.5)	In silico indicates pathogenicity. No family information or additional functional evidence.
c.292C>T	Proband with predicted or proven null variant		Waanders E, et al., 2010 , PMID: 20095989	Unknown			HPO term(s): <ul style="list-style-type: none"> Polycystic liver disease Autosomal dominant inheritance Free text: A proband was defined as a patient with at least one affected family member. PCLD was diagnosed in probands or isolated individuals when >20 liver cysts were present. In case of a positive family history, patients aged 40 years or younger with any liver cysts or patients above the age of 40 with four or more liver cysts were diagnosed with PCLD.	-	-	-	-			Method 1: Sanger sequencing	Score	0.5 (1.5)	In silico indicates pathogenicity. No family information or additional functional evidence.

								Segregations									
Label	Variant Type ^	Variant	Reference (PMID) ⇅	Proband Sex	Proband Age	Proband Ethnicity	Proband Phenotypes	# Aff	# Unaff	LOD Score	Counted	Sequencing	Proband Previous Testing	Proband Methods of Detection	Score Status	Proband Points (default points)	Reason for Changed Score
c.715C>T	Proband with predicted or proven null variant		Waanders E, et al., 2010, PMID: 20095989 ↗	Unknown			HPO term(s): <ul style="list-style-type: none"> Polycystic liver disease Autosomal dominant inheritance Free text: A proband was defined as a patient with at least one affected family member. PCLD was diagnosed in probands or isolated individuals when >20 liver cysts were present. In case of a positive family history, patients aged 40 years or younger with any liver cysts or patients above the age of 40 with four or more liver cysts were diagnosed with PCLD.	-	-	-	-			Method 1: Sanger sequencing	Score	0.5 (1.5)	In silico indicates pathogenicity. No family information or additional functional evidence.
Total points: 10.85																	

Genetic Evidence: Case Level (family segregation information without proband data or scored proband data)

No segregation evidence for a Family without a proband was found.

Genetic Evidence: Case-Control

No scored Case-Control evidence was found.

Experimental Evidence

Label	Experimental Category ⇅	Reference ⇅	Explanation	Score Status ⇅	Points (default points)	Reason for Changed Score
Cystic Kidney Rescue in SEC63KO and DKO mouse models	Rescue Non-human model organism	Fedeles SV, et al., 2015, PMID: 25844898 ↗	Reduction of cystic kidneys	Score	1 (2)	The rescue experiments better support the SEC63 pathology described by the model in this paper - that include cells derived from the mouse models and further mouse models to characterize the relationship with SEC63, XBP1, and PC1.
Cell Culture Rescue for PC1 cleavage	Rescue Cell culture model	Fedeles SV, et al., 2015, PMID: 25844898 ↗	Retroviral transduction to rescue PC1 cleavage	Score	0.5 (1)	One experiment to show SEC63 and XBP1 directly affect PC1 cleavage.
SEC63 KO Model	Model Systems Non-human model organism	Fedeles SV, et al., 2015, PMID: 25844898 ↗	SEC63KO and DKO mouse models develop kidney cysts similar to the phenotype seen in humans: inactivation of SEC63 results activation of the IRE1α-XBP1 branch of unfolded protein response (UPR), SEC63 exists in a protein interaction complex with PC1, and cleavage of PC1 is dependent on SEC63 and XBP1	Score	1 (2)	Development of mouse model with cystic kidneys.
Zebrafish sec63-st67	Model Systems Non-human model organism	Monk KR, et al., 2013, PMID: 22864019 ↗	Increase in ER stress. Abnormal liver development - fragmentation and swelling of the ER, smaller mitochondria, regions of empty cytoplasm, large lysosomes filled with debris. Development of liver steatosis. Translates to missense mutation in region conserved in humans.	Score	2 (2)	
Mouse Tissue Northern Blot	Expression A	Davila S, et al., 2004, PMID: 15133510 ↗	Mouse tissues (heart, brain, spleen, lung, liver, skeletal muscle, kidney, testis) were probed with a human RT-PCR based probe. SEC63 is expressed 2x higher in liver compared to other tissues.	Review	0.5 (0.5)	No scoring because it only shows SEC63 is expressed higher in liver compared to other tissues that also express SEC63.

Total points: 4.50

Biochemical Function: The gene product performs a biochemical function shared with other known genes in the disease of interest (A), OR the gene product is consistent with the observed phenotype(s) (B)

Protein Interactions: The gene product interacts with proteins previously implicated (genetically or biochemically) in the disease of interest

Expression: The gene is expressed in tissues relevant to the disease of interest (A), OR the gene is altered in expression in patients who have the disease (B)

Functional Alteration of gene/gene product: The gene and/or gene product function is demonstrably altered in cultured patient or non-patient cells carrying candidate variant(s)

Model Systems: Non-human model organism OR cell culture model with a similarly disrupted copy of the affected gene shows a phenotype consistent with human disease state

Rescue: The phenotype in humans, non-human model organisms, cell culture models, or patient cells can be rescued by exogenous wild-type gene or gene product

Non-scorable Evidence

PMID: 16835903

Waanders E, te Morsche RH, de Man RA, Jansen JB, Drenth JP. Extensive mutational analysis of PRKCSH and SEC63 broadens the spectrum of polycystic liver disease. *Human mutation*. **2006** Aug;27(8):830.

Explanation: Most mutations in ADPLD (PRKCSH and SEC63) are found in individuals with 8 - 20 liver cysts.

PMID: 20408955

Van Keimpema L, De Koning DB, Van Hoek B, Van Den Berg AP, Van Oijen MG, De Man RA, Nevens F, Drenth JP. Patients with isolated polycystic liver disease referred to liver centres: clinical characterization of 137 cases. *Liver international : official journal of the International Association for the Study of the Liver*. **2011** Jan;31(1):92-8.

Explanation: Clinical Study of 137 patients, of these genetic testing was performed on 91 patients. Association of worsening disease in females. More severe disease with earlier age of onset seen in those with a mutation in PRKCSH or SEC63. 34% of patients had renal cysts.

PMID: 32405593

Wilson EM, Choi J, Torres VE, Somlo S, Besse W. Large Deletions in GANAB and SEC63 Explain 2 Cases of Polycystic Kidney and Liver Disease. *Kidney international reports*. **2020** May;5(5):727-731.

Explanation: Large deletion found in SEC63 spanning intron 1, exon 2 and intron 2 causing a loss of the first cytoplasmic loop of SEC63. They were unable to amplify a product due to PCR-related challenges and define the exact deletion at this time.

PMID: 28375157

Besse W, Dong K, Choi J, Punia S, Fedeles SV, Choi M, Gallagher AR, Huang EB, Gulati A, Knight J, Mane S, Tahvanainen E, Tahvanainen P, Sanna-Cherchi S, Lifton RP, Watnick T, Pei YP, Torres VE, Somlo S. Isolated polycystic liver disease genes define effectors of polycystin-1 function. *The Journal of clinical investigation*. **2017** May 1;127(5):1772-1785.

Explanation: Supporting evidence of aberrant genes causing autosomal dominant polycystic liver disease affect proper post-translational modifications of polycystin-1 leading to cytsogenesis.

 For best printing, choose "Landscape" for layout, 50% for Scale, "Minimum" for Margins, and select "Background graphics".

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