

Supplemental Tables

McCoy et al. State of the Science for Kidney Disorders in Phelan-McDermid Syndrome 2022

Supplemental Table S1. Genes known to be associated with renal abnormalities used as a training set with ToppGene [1-5]

Gene	Kidney dysplasia	Renal hypoplasia	Renal cysts or Cystic disease	Renal agenesis	Vesicoureteral reflux	Renal hypodysplasia
<i>BMP4</i>						✓
<i>CHDIL</i>					✓	✓
<i>DICER1</i>			✓			
<i>DSTYK</i>						✓
<i>EYA1</i>		✓				
<i>FRAS1</i>				✓		
<i>FREM2</i>				✓		
<i>GATA3</i>	✓					
<i>GDNF</i>		✓		✓		
<i>GREB1L</i>		✓		✓		
<i>HNF1B</i>	✓	✓	✓			
<i>ITGA8</i>		✓		✓		
<i>KAL1/ANOS1</i>				✓		
<i>MUC1</i>			✓			
<i>PAX2</i>	✓	✓				
<i>PKD1, PKD2</i>			✓			
<i>RET</i>		✓		✓		
<i>ROBO2</i>					✓	
<i>SALL1</i>						✓
<i>SEMA3A</i>	✓					
<i>SIX1, SIX2, SIX5</i>					✓	✓
<i>SOX17</i>					✓	
<i>TNXB</i>					✓	
<i>TRAP1</i>				✓	✓	
<i>UMOD</i>			✓			
<i>UPK3A</i>		✓		✓		
<i>VUR1</i>					✓	
<i>WNT4</i>						✓

Supplemental Table S2. Source of patient data for the pooled analysis (N=152)

<i>Study</i>	<i>Patients included in the study</i>
Kirkpatrick et al, 2011 [6]	1
Soorya et al, 2013 [7]	30
Bonaglia et al, 2011 [8]	4
Disciglio et al, 2014 [9]	9
Palumbo et al, 2017 [10]	15
Fontes et al, 2015 [11]	1
Ishikawa et al, 2015 [12]	1
Tabet et al, 2017 [13]	46
Lei et al, 2016 [14]	1
Toruner et al, 2009 [15]	1
Ha et al, 2017 [16]	1
Samogy-Costa et al, 2019 [17]	27
Xu et al, 2020 [18]	15

Supplemental Table S3. Genes within the CNV segments associated with kidney disorders at $P < 0.05$ with gene location, prioritization rank out of 38 genes, HI < 10% estimate [19], and pLI score if > 0.9 from gnomAD [20].

<i>Gene</i>	<i>Empirical P-value</i>	<i>Gene location</i>	<i>Prioritization rank</i>	<i>pLI > 0.9 or HI < 10%</i>
<i>PRR5</i>	0.034997	chr22:45,064,427-45,133,561	15	
<i>PRR5-ARHGAP8</i>	0.020698	chr22:45,098,113-45,258,664	5	
<i>ARHGAP8</i>	0.020698	chr22:45,098,355-45,258,665	12	
<i>PHF21B</i>	0.013199	chr22:45,277,042-45,405,880	21	pLI 0.99
<i>NUP50</i>	0.018398	chr22:45,559,722-45,583,896	13	
<i>KIAA0930</i>	0.007899	chr22:45,586,219-45,636,650	23	
<i>MIR1249</i>	0.007899	chr22:45,596,835-45,596,900	26	
<i>UPK3A*</i>	0.005399	chr22:45,680,830-45,691,755	4	
<i>FAM118A</i>	0.0028	chr22:45,704,849-45,737,836	24	
<i>SMC1B</i>	0.0028	chr22:45,739,944-45,809,500	11	
<i>RIBC2</i>	0.0017	chr22:45,809,572-45,828,376	27	
<i>FBLN1</i>	0.0009	chr22:45,898,118-45,997,015	2	
<i>ATXN10</i>	0.0005	chr22:46,067,678-46,241,187	8	
<i>MIR4762</i>	0.002	chr22:46,156,404-46,156,478	33	
<i>WNT7B</i>	0.0003	chr22:46,316,242-46,373,009	1	
<i>LOC730668</i>	0.0003	chr22:46,402,496-46,406,657	37	
<i>LINC00899</i>	0.0003	chr22:46,435,787-46,440,748	36	
<i>PRR34</i>	0.0003	chr22:46,445,358-46,450,024	20	
<i>MIRLET7BHG</i>	0.0003	chr22:46,449,749-46,509,808	22	
<i>MIR3619</i>	0.0003	chr22:46,486,924-46,487,006	30	

<i>MIRLET7A3</i>	0.0003	chr22:46,508,629-46,508,702	18	
<i>MIR4763</i>	0.0003	chr22:46,509,446-46,509,537	38	
<i>MIRLET7B</i>	0.0003	chr22:46,509,566-46,509,648	7	
<i>PPARA</i>	0.0012	chr22:46,546,424-46,639,653	3	HI 7.3%
<i>CDPF1</i>	0.0012	chr22:46,639,908-46,646,576	25	
<i>PKDREJ</i>	0.0012	chr22:46,651,560-46,659,219	9	
<i>TTC38</i>	0.0012	chr22:46,663,858-46,689,905	28	
<i>GTSE1</i>	0.0012	chr22:46,692,638-46,726,707	10	
<i>TRMU</i>	0.0012	chr22:46,726,772-46,753,237	16	
<i>CELSR1</i>	0.004	chr22:46,756,731-46,933,067	6	pLI 1.00
<i>GRAMD4</i>	0.004	chr22:46,971,909-47,075,688	19	pLI 1.00
<i>CERK</i>	0.004	chr22:47,080,307-47,134,158	17	
<i>TBC1D22A</i>	0.0005	chr22:47,158,518-47,571,342	14	
<i>LINC00898</i>	0.0002	chr22:48,016,792-48,027,318	36	
<i>MIR3201</i>	0.005199	chr22:48,670,176-48,670,227	31	
<i>LOC284933</i>	0.006699	chr22:48,934,712-48,943,199	32	
<i>MIR4535</i>	0.0042	chr22:49,176,107-49,176,165	35	
<i>LINC01310</i>	0.0042	chr22:49,262,582-49,294,198	34	

**UPK3A* is already a known kidney-related gene.

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