

Table S1: CAKUT panel genes

	Gene	OMIM Gene No.	Inheritance	Key Associated Phenotype(s)
1	<i>ACE</i>	106180	Autosomal recessive	Renal tubular dysgenesis
2	<i>ACTG2</i>	102545	Autosomal dominant	Visceral myopathy; megacystis–microcolon–intestinal hypoperistalsis syndrome
3	<i>AGT</i>	106150	Autosomal recessive	Renal tubular dysgenesis
4	<i>AGTR1</i>	106165	Autosomal recessive	Renal tubular dysgenesis, Hypertension
5	<i>ANOS1</i>	300836	X-linked recessive	Kallmann syndrome; renal agenesis
6	<i>ARHGEF6</i>	300267	X-linked	Neurodevelopmental disorder
7	<i>BICC1</i>	614295	Autosomal dominant	Renal adysplasia
8	<i>BMP4</i>	112262	Autosomal dominant	microphthalmia, orofacial cleft 11
9	<i>BMP7</i>	112267	//	Candidate gene
10	<i>BNC2</i>	608669	Autosomal dominant	Lower urinary tract obstruction, congenital
11	<i>CCNQ</i>	616415	X-linked dominant	STAR syndrome
12	<i>CDC5L</i>	602868	//	Candidate gene
13	<i>CENPF</i>	600236	Autosomal recessive	Stromme syndrome
14	<i>CHD1L</i>	613039	//	Candidate gene
15	<i>CHRM3</i>	118494	Autosomal recessive	Prune-belly syndrome
16	<i>CHRNA3</i>	118503	Autosomal recessive	lung cancer, CAKUT
17	<i>COL4A1</i>	120130	Autosomal dominant	tortuosity of Retinal arteries; susceptibility intracerebral hemorrhage; hereditary angiopathy with nephropathy,
18	<i>CRKL</i>	602007	//	CAKUT, renal hypodysplasia
19	<i>DHCR7</i>	602858	Autosomal recessive	Smith–Lemli–Opitz syndrome
20	<i>DSTYK</i>	612666	Autosomal recessive / dominant	Congenital anomalies of kidney and urinary tract ; Spastic paraplegia
21	<i>ETV4</i>	600418	//	Candidate gene
22	<i>EYA1</i>	601653	Autosomal dominant	Branchio-oto-renal syndrome
23	<i>FANCB</i>	300515	X-linked recessive	Fanconi anemia
24	<i>FAT4</i>	612411	Autosomal recessive	Hennekam syndrome; renal lymphangiectasia
25	<i>FGF20</i>	605558	Autosomal recessive	Renal hypodysplasia
26	<i>FOXC1</i>	601090	Autosomal dominant	Axenfeld–Rieger syndrome;
27	<i>FOXC2</i>	602402	Autosomal dominant	Lymphedema–distichiasis
28	<i>FOXP1</i>	605515	Autosomal dominant	Intellectual developmental disorder
29	<i>FRAS1</i>	607830	Autosomal recessive	Fraser syndrome
30	<i>FREM1</i>	608944	Autosomal recessive / dominant	Manitoba oculotrichoanal syndrome, trigonocephaly
31	<i>FREM2</i>	608945	Autosomal recessive	Fraser syndrome
32	<i>GATA3</i>	131320	Autosomal dominant	HDR syndrome (hypoparathyroidism, deafness, renal dsplasia)
33	<i>GFRA1</i>	601496	Autosomal recessive	Renal hypodysplasia
34	<i>GLI3</i>	165240	Autosomal dominant	Pallister–Hall, renal anomalies
35	<i>GREB1L</i>	617782	Autosomal dominant	deafness, renal hypodysplasia
36	<i>GRIPI</i>	604597	Autosomal recessive	Fraser syndrome
37	<i>HNF1B</i>	189907	Autosomal dominant	Renal cysts and type 2 diabetes mellitus

38	HOXA11	142958	Autosomal dominant	Radioulnar synostosis
39	HOXA13	142959	Autosomal dominant	Hand-foot-genital syndrome;Guttmacher syndrome
40	HPSE2	613469	Autosomal recessive	Urofacial syndrome
41	ITGA8	604063	Autosomal recessive	Renal hypodysplasia
42	KYNU	605197	Autosomal recessive	Hydroxykynureninuria
43	LIFR	151443	Autosomal recessive	Stüve–Wiedemann syndrome
44	LMOD1	616104	Autosomal recessive	Megacystis-microcolon-intestinal hypoperistalsis syndrome
45	LRIG2	608869	Autosomal recessive	Urofacial syndrome
46	LRP4	604270	Autosomal dominant / recessive	Cenani–Lenz syndrome;Sclerosteosis
47	MYH11	160745	Autosomal dominant / recessive	Visceral myopathy; megacystis
48	MYL9	609905	Autosomal recessive	Megacystis-microcolon-intestinal hypoperistalsis syndrome
49	MYLK	600922	Autosomal dominant / recessive	Aortic aneurysm; Megacystis-microcolon-intestinal hypoperistalsis syndrome
50	MYOCD	613055	Autosomal dominant	congenital Megabladder
51	NIPBL	608667	Autosomal dominant	Cornelia de Lange syndrome 1
52	NOTCH2	600275	//	Candidate gene
53	NPNT	607803	//	Candidate gene
54	NRIP1	602490	Autosomal dominant	CAKUT
55	PAX2	167409	Autosomal dominant	Glomerulosclerosis, focal segmental; Papillorenal syndrome
56	PBX1	176310	Autosomal dominant	Congenital anomalies of kidney and urinary tract syndrome with or without hearing loss, abnormal ears, or developmental delay
57	REN	179820	Autosomal dominant / recessive	Tubulointerstitial kidney disease
58	RET	164761	Autosomal dominant	Hirschsprung disease;Medullary thyroid carcinoma;Pheochromocytoma
59	ROBO1	602430	Autosomal dominant / recessive	Nystagmus, congenital; Neurooculorenal syndrome; Pituitary hormone deficiency
60	ROBO2	602431	Autosomal dominant	Vesicoureteral reflux
61	SALL1	602218	Autosomal dominant	Townes–Brocks syndrome
62	SALL4	607343	Autosomal dominant	Duane-radial ray syndrome; IVIC syndrome
63	SIX1	601205	Autosomal dominant	Branchio-oto-renal syndrome
64	SIX2	604994	//	Candidate gene
65	SIX5	600963	//	Branchiootorenal syndrome 2
66	SLIT2	603746	//	Candidate gene
67	SOX11	600963	Autosomal dominant	Intellectual developmental disorder with microcephaly and with or without ocular malformations or hypogonadotropic hypogonadism
68	SOX17	610928	Autosomal dominant	Pulmonary hypertension, primary; Vesicoureteral reflux
69	SRGAP1	606696	Autosomal dominant / somatic mutation	Thyroid cancer, nonmedullary
70	TBC1D1	612465	//	Candidate gene
71	TBX18	604613	Autosomal dominant	CAKUT
72	TRAP1	606219	//	Candidate gene
73	UMOD	191845	Autosomal dominant	AD tubulointerstitial kidney disease

74	<i>UPK3A</i>	611559	//	Candidate gene
75	<i>WNT4</i>	603490	Autosomal dominant / recessive	Mullerian aplasia and hyperandrogenism; SERKAL syndrome
76	<i>WT1</i>	607102	Autosomal dominant / somatic mutation	Denys–Drash, Frasier syndrome
77	<i>ZMYM2</i>	603209	Autosomal dominant	Neurodevelopmental disorder