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Review article

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The genetics and pathogenesis of CAKUT

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Table S1: 54 genes representing monogenic causes of isolated or syndromic human CAKUT, if mutated.

Gene	OMIM Gene	MOI	Protein	OMIM phenotype	MIM number
Autosomal recessive					
ACE	106180	AR	Angiotensin I-converting enzyme	Renal tubular dysgenesis	267430
AGT	106150	AR	Angiotensinogen	Renal tubular dysgenesis	267430
AGTR1	106165	AR	Angiotensin II receptor, type 1	Renal tubular dysgenesis	267430
CHRM3*	118494	AR	Cholinergic receptor, muscarinic 3	Prune belly syndrome	100100
CHRNA3	118503	AR	Cholinergic receptor, neuronal nicotinic, alpha polypeptide 3	Bladder dysfunction, autonomic, with impaired pupillary reflex and secondary CAKUT	191800
ETV4	60711	AR	ETS variant transcription factor 4	-	-
FAT4*	612411	AR	FAT atypical cadherin 4	Van Maldergem syndrome 2	615546
FGF20	605558	AR	Fibroblast growth factor 20	?Renal hypodysplasia/aplasia 2	615721
FRAS1*	607830	AR	Extracellular matrix protein FRAS1	Fraser syndrome 1	219000
FREM1*	608944	AR	FRAS1 related extracellular matrix protein 1	Bifid nose with or without anorectal and renal anomalies; Manitoba oculotrichoanal syndrome	608980; 248450
FREM2*	608945	AR	FRAS1 related extracellular matrix protein 2	Fraser syndrome 2	617666
GFRA1	601496	AR	GDNF family receptor alpha-1	Renal hypodysplasia/aplasia 4	619887
GRIP1*	604597	AR	Glutamate receptor interacting protein 1	Fraser syndrome 3	617667
HOXA11	142958	AR	Homeobox A11	Radioulnar synostosis with amegakaryocytic thrombocytopenia 1 (AD)	605432
HPSE2*	613469	AR	Heparanase 2 (Inactive)	Urofacial syndrome 1	236730
ITGA8	604063	AR	Integrin α8	Renal hypodysplasia/aplasia 1	191830
LRIG2*	608869	AR	Leucine-rich repeats-and immunoglobulin-like domains-containing protein 2	Urofacial syndrome 2	615112
NPNT	610306	AR	Nephronectin	-	-
REN	179820	AR	Renin	Renal tubular dysgenesis	267430
TRAP1*	606219	AR	Heat-shock protein 75 (also known as TNF receptor-associated protein 1)	-	-
VWA2*	618281	AR	Von Willebrand factor A domain-containing protein 2	-	-
Autosomal dominant					
BNC2	608669	AD	Basonuclin 2	Lower urinary tract obstruction, congenital	618612
COL4A1	120130	AD	Collagen, type 4, alpha-1	HANAC syndrome, brain small vessel disease 1 with or without ocular anomalies	611773; 175780
CRKL	602007	AD	CRK Like Proto-Oncogene, adaptor protein	-	-
DSTYK	612666	AD	Dual serine/threonine and tyrosine protein kinase	Congenital anomalies of kidney and urinary tract 1	610805
EYA1*	601653	AD	Eyes absent homolog 1	Branchiootorenal syndrome 1, with or without cataracts	113650
FOXC1*	601090	AD	Forkhead box C1	Axenfeld–Rieger syndrome; anterior segment dysgenesis	602482; 601631
FOXC2*	602402	AD	Forkhead box C2	Lymphedema-distichiasis syndrome with renal disease and diabetes mellitus	153400
FOXP1*	605515	AD	Forkhead box P1	Intellectual developmental disorder with language impairment with or without autistic features	613670
GATA3*	131320	AD	GATA binding protein 3	Hypoparathyroidism, sensorineural deafness, and renal dysplasia	146255
GREB1L	617782	AD	Growth regulation by estrogen in breast cancer 1-like	Renal hypodysplasia/aplasia 3	617805
HNF1B*	189907	AD	HNF homeobox B	Renal cysts and diabetes syndrome	137920
MUC1	158340	AD	Mucin 1	Tubulointerstitial kidney disease, autosomal dominant, 2	174000
MYOCD	606127	AD	Myocardin	Megabladder, congenital	618719

NRIP1	602490	AD	Nuclear receptor interacting protein 1	?Congenital anomalies of kidney and urinary tract 3	618270
RET	164761	AD	RET protooncogene	Medullary thyroid carcinoma; Multiple endocrine neoplasia IIA; Multiple endocrine neoplasia IIB; Pheochromocytoma; Hirschsprung disease, protection against; Hirschsprung disease, susceptibility to, 1	155240; 171400; 162300; 171300; 142623
PAX2*	167409	AD	Paired box 2	Papillorenal syndrome	120330
PBX1*	176310	AD	PBX homeobox 1	Congenital anomalies of kidney and urinary tract syndrome with or without hearing loss, abnormal ears, or developmental delay	617641
ROBO1	602430	AD	Roundabout, axon guidance receptor, homolog 1 (*Drosophila*)	-	-
ROBO2*	602431	AD	Roundabout, axon guidance receptor, homolog 2 (*Drosophila*)	Vesicoureteral reflux 2	610878
SALL1*	602218	AD	Sal-like protein 1 (also known as spalt-like transcription factor 1)	Townes-Brocks branchiootorenal-like syndrome; Townes-Brocks syndrome 1	107480
SIX1*	601205	AD	SIX homeobox 1	Branchiootic syndrome 3	608389
SIX2	604994	AD	SIX homeobox 2	-	-
SIX5*	600963	AD	SIX homeobox 5	Branchiootorenal syndrome 2	610896
SLIT2	603746	AD	Slit homolog 2	-	-
SOX17	610928	AD	Transcription factor SIX-17	Vesicoureteral reflux 3	613674
SRGAP1	606523	AD	SLIT-ROBO Rho GTPase activating protein 1	{Thyroid cancer, nonmedullary, 2}	188470
TBX18	604613	AD	T-Box transcription factor	Congenital anomalies of kidney and urinary tract 2	143400
TNXB	600985	AD	Tenascin XB	Vesicoureteral reflux 8	615963
UPK3A	611559	AD	Uroplakin 3A	-	-
WNT4*	603490	AD	Protein Wnt-4	Mullerian aplasia and hyperandrogenism	158330
ZMYM2*	602221	AD	Zink finger, MYM-type 2	Neurodevelopmental-craniofacial syndrome with variable renal and cardiac abnormalities	619522
X-linked					
ARHGEF6	300267	XL	Rac/Cdc42 Guanine Nucleotide Exchange Factor 6	-	-
FAM58A*	300708	XL	Family with sequence similarity 58 member A	STAR syndrome	300707

AD, autosomal dominant; **AR**, autosomal recessive; **MOI**, Mode of inheritance; **OMIM**, Online Mendelian Inheritance in Men; **XL**; X-linked.

Asterisk **(*)** is added to the gene symbol if mainly a syndromic phenotype has been described in the literature. A CAKUT phenotype that is accompanied by extrarenal features can be found in ≥50% of reported cases.

Table S2. 131 genes for syndromes with facultative CAKUT features in humans.

Genes in which variants typically result in a multi-organ syndrome with facultative CAKUT features in less than 50% of reported cases. Genes are listed by mode of inheritance and alphabetically by gene symbol. The prevalence of CAKUT may be underestimated as a comprehensive analysis of the kidney and urinary tract is not always reflected in the literature.

Gene	Encoded protein	Reference	MOI	OMIM Gene number
B3GALTL	Beta 3-glucosyltransferase	Lesnik Oberstein *Am J Hum Genet* 79:562, 2006[1]	AR	610308
BMPER	Bone morphogenetic protein-binding endothelial regulator protein	Greenbaum *Eur J Med Genet* 62:167, 2019[2]	AR	608699
BSCL2	BSCL2, Seipin lipid droplet biogenesis associated	Haghighi *Clin Genet* 89: 434, 2016[3]	AR	606158
CCBE1	Collagen and calcium-binding EGF domain-containing protein 1	Van Balkom *Am J Med Genet* 112:412, 2002[4]	AR	612753
CD151	CD151 molecule (Raph blood group)	Karamatic *Blood* 104:2217, 2004[5]	AR	602243
CENPF	Centromeric protein F	Filges *Hum Mutat* 37:359, 2016[6]	AR	600236
CEP55	Centrosomal protein, 55-kD	Frosk *J Med Genet* 54:490, 2017[7]	AR	610000
CHRNG	Cholinergic receptor nicotinic gamma subunit	Vogt *J Med Genet* 49:21, 2012[8]	AR	100730
CISD2	CDGSH iron sulfur domain 2	Amr *Am J Hum Genet* 81:673, 2007[9]	AR	611507
COL18A1	Collagen, type XVIII, alpha-1	Caglayan *Pediatr Neurol* 51:806, 2014[10]	AR	120328
CTU2	Cytosolic thiouridylase, subunit 2	Shaheen *Am J Med Genet* 170:3222, 2016[11]	AR	617057
DHCR7	7-Dehydrocholesterol reductase	Löffler *Am J Med Genet* 13:174, 2000[12]	AR	602858
DIS3L2	DIS3 like 3'-5' exoribonuclease 2	Astuti *Nat Genet* 44:277, 2012 [13]	AR	614184
DYNC2H1	Dynein cytoplasmic 2 heavy chain 1	Baujat *J Med Genet* 50:91, 2013[14]	AR	603297
EMG1	EMG1, N1-specific pseudouridine methyltransferase	Armistead *Am J Hum Genet* 84:728, 2009[15]	AR	611531
ERCC8	Excision repair cross-complementing, group 8	Bertola *J Hum Genet* 51:701, 2006[16]	AR	609412
ESCO2	Establishment of sister chromatid cohesion N-acetyltransferase 2	Vega *J Med Genet* 47:30, 2010[17]	AR	609353
ETFA	Electron transfer flavoprotein alpha subunit	Lehnert *Eur J Pediatr* 139:56, 1982[18]	AR	608053
ETFB	Electron transfer flavoprotein beta subunit	Lehnert *Eur J Pediatr* 139:56, 1982[18]	AR	130410
ETFDH	Electron transfer flavoprotein dehydrogenase	Lehnert *Eur J Pediatr* 139:56, 1982[18]	AR	231675
FANCA	Fanconi anemia complementation group A	Joenje *Nat Rev Genet* 2:466, 2001[19]	AR	607139
FANCD2	Fanconi anemia complementation group D2	Kalb *Am J Hum Genet* 80:895, 2007[20]	AR	613984
FANCE	Fanconi anemia complementation group E	Wegner *Clin Genet* 50:479, 1996[21]	AR	613976
FANCI	Fanconi anemia complementation group I	Savage *Am J Med Genet A* 170A:386, 2015[22]	AR	611360
FANCL	Fanconi anemia complementation group L	Vetro *Hum Mutat* 36:562, 2015[23]	AR	608111
FIBP	Fibroblast growth factor, acidic, intracellular binding protein	Thauvin-Robinet *Clin Genet* 89:e1-4, 2016[24]	AR	608296
HAAO	3-Hydroxyanthranilate 3,4-dioxygenase	Shi *N Engl J Med* 377:544, 2017[25]	AR	604521
HYLS1	HYLS1, centriolar and ciliogenesis associated	Paetau *J Neuropathol Exp Neurol* 67:750, 2008[26]	AR	610693
ICK	Intestinal cell kinase	Lahiry *Am J Hum Genet* 84:822, 2009[27]	AR	612325
ITGA3	Integrin subunit alpha 3	Yalcin *Hum Mol Genet* 24:3679, 2015[28]	AR	605025

Gene	Encoded protein	Reference	MOI	OMIM Gene number
JAM3	Junctional adhesion molecule 3	Mochida *Am J Hum Genet* 87:882, 2010[29]	AR	606871
KYNU	Kynureninase	Shi *N Engl J Med* 377:544, 2017[25]	AR	605197
LMNA	Lamin A/C	Klupa *Endocrine* 36:518, 2009[30]	AR	150330
LRP2	LDL receptor related protein 2	Kantarci *Nat Genet* 39:957, 2007[31]	AR	600073
LRP4	LDL receptor related protein 4	Li *Am J Hum Genet* 86:696, 2010[32]	AR	604270
NADSYN1	NAD synthetase 1	Szot *Am J Hum Genet* 106:129, 2020[33]	AR	608285
NCAPG2	Non-SMC condensin II complex subunit G2	Khan *Am J Hum Genet* 104:94, 2019[34]	AR	608532
PEX1	Peroxisomal biogenesis factor 1	Crane *Hum Mutat* 26:167, 2005[35]	AR	602136
PEX5	Peroxisomal biogenesis factor 5	Sundaram *Nat Clin Pract Gastroenterol Hepatol* 5:456, 2008[36]	AR	600414
PIGL	Phosphatidylinositol glycan anchor biosynthesis class L	Schnur *Am J Med Genet* 72:24, 1997[37]	AR	605947
PIGN	Phosphatidylinositol glycan anchor biosynthesis class N	Ohba *Neurogenetics* 15:85, 2014[38]	AR	606097
PIGO	Phosphatidylinositol glycan anchor biosynthesis class O	Krawitz *Am J Hum Genet* 91:146, 2012[39]	AR	614730
PIGT	Phosphatidylinositol glycan anchor biosynthesis class T	Nakashima *Neurogenetics* 15:193, 2014[40]	AR	610272
PIGV	Phosphatidylinositol glycan anchor biosynthesis class V	Horn *Eur J Hum Genet* 22:762, 2014[41]	AR	610274
PIGY	Phosphatidylinositol glycan anchor biosynthesis class Y	Ilkovski *Hum Mol Genet* 24:6146, 2015[42]	AR	610662
PMM2	Phosphomannomutase 2	Horslen *Arch Dis Child* 66:1027, 1991[43]	AR	601785
POC1A	POC1 centriolar protein	Shaheen *Am J Hum Genet* 91:330, 2012[44]	AR	614783
RAB23	RAS-associated protein RAB23	Alessandri *Am J Med Genet A* 152A:982, 2010[45]	AR	606144
RBM8A	RNA-binding motif protein 8A	Skorka *Genet Couns* 16:377, 2005[46]	AR	605313
RECQL4	RecQ Like Helicase 4	Siitonen *Eur J Hum Genet* 17:151, 2009[47]	AR	603780
ROR2	Receptor tyrosine kinase like orphan receptor 2	Wiens *Clin Genet* 37:481, 1990[48]	AR	602337
STRA6	Stimulated by retinoic acid 6	Golzio *Am J Hum Genet* 80:1179, 2007[49]	AR	610745
TMCO1	Transmembrane and coiled-coil domains 1	Xin *Proc Natl Acad Sci U S A* 107:258, 2010[50]	AR	614123
UBR1	Ubiquitin protein ligase E3 component N-recognin 1	Vanlieferinghen *Genet Couns* 14:105, 2003[51]	AR	605981
WFS1	Wolframin ER transmembrane glycoprotein	Salih *Acta Paediatr Scand* 80:567, 1991[52]	AR	606201
WNT3	Wnt family member 3	Niemann *Am J Hum Genet* 74:558, 2004[53]	AR	165330
ZMPSTE24	Zinc metallopeptidase STE24	Chen *Am J Med Genet A* 149A:1550, 2009[54]	AR	606480
ACTB	Actin beta	Rivière *Nat Genet* 44:440, 2012[55]	AD	102630
ACTG1	Actin gamma 1	Rivière *Nat Genet* 44:440, 2012[55]	AD	102560
ACTG2	Actin, gamma-2, smooth muscle, enteric	Thorson *Hum Genet* 133:737, 2014[56]	AD	102545
ARID1B	AT-Rich interaction domain 1B	Levy *J Med Genet* 28, 1991[57]	AD	614556
BMP4	Bone morphogenic protein 4	Weber *J Am Soc Nephrol* 19:891, 2008[58]	AD	112262
CD96	CD96 molecule	Kaname *Am J Hum Genet* 81:835, 2007[59]	AD	606037
CDKN1C	Cyclin dependent kinase inhibitor 1C	Mussa *Pediatr Nephrol* 27:397, 2012[60]	AD	600856
CHD7	Chromodomain helicase DNA binding protein 7	Janssen *Hum Mutat* 33:1149, 2012[61]	AD	608892
CREBBP	CREB binding protein	Kanjilal *J Med Genet* 29:669, 1992[62]	AD	600140

Gene	Encoded protein	Reference	MOI	OMIM Gene number
DACT1	Dishevelled binding antagonist of beta catenin 1	Webb *Hum Mutat* 38:373, 2017[63]	AD	607861
EP300	E1A binding protein P300	Roelfsema *Am J Hum Genet* 76:572, 2005[64]	AD	602700
FBN1	Fibrillin 1	Tokhmafshan *Pediatr Nephrol* 32:565, 2017[65]	AD	134797
FGF10	Fibroblast growth factor 10	Milunsky *Clin Genet* 69:349[66], 2006; Bamforth *Am J Med Genet* 43:932, 1992[67]	AD	602115
FGF8	Fibroblast growth factor 8	Falardeau *J Clin Invest* 118:2822, 2008[68]	AD	600483
FGFR1	Fibroblast growth factor receptor 1	Farrow *Am J Hum Genet A* 140A:537, 2006[69]	AD	136350
FGFR2	Fibroblast growth factor receptor 2	LeHeup *Eur J Pediatr* 154:130, 1995[70]	AD	176943
FGFR3	Fibroblast growth factor receptor 3	Rohmann *Nat Genet* 38:495, 2006[71]	AD	134934
FOXF1	Forkhead box F1	Hilger *Hum Mutat* 36:1150, 2015[72]	AD	601089
GDF6	Growth differentiation factor 6	Tassabehji *Hum Mutat* 29:1017, 2008[73]	AD	601147
GLI2	GLI family zinc finger 2	Carmichael *J Urol* 190:1884, 2013[74]	AD	165230
GLI3	GLI family zinc finger 3	Cain *PLoS One* 4:e7313, 2009[75]	AD	165240
HOXA13	Homeobox A13	Halal *Am J Med Genet* 30:793, 1998[76]	AD	142959
JAG1	Jagged 1	Kamath *Nat Rev Nephrol* 9:409, 2013[77]	AD	601920
KAT6B	Lysine acetyltransferase 6B	Campeau *Am J Med Genet* 90:282, 2012[78]	AD	605880
KCNQ1OT1	KCNQ1 opposite strand/antisense transcript 1	Chiesa *Hum Mol Genet* 21:10, 2012[79]	AD	604115
KCTD1	Potassium channel tetramerization domain containing 1	Marneros *Am J Hum Genet* 92:621, 2013[80]	AD	613420
KRAS	KRAS proto-oncogene, GTPase	Schubbert *Nat Genet* 38:331, 2006[81]	AD	190070
MLL2 / KMT2D	Myeloid/lymphoid or mixed-lineage leukemia protein 2	Banka *Eur J Hum Genet* 20:381, 2012[82]	AD	602113
MNX1	Motor neuron and pancreas homeobox 1	Hagan *Am J Hum Genet* 66:1504, 2000[83]	AD	142994
MTOR	Mechanistic target of rapamycin	Moosa *Am J Med Genet A* 173:264, 2017[84]	AD	601231
MYCN	MYCN proto-oncogene	Marcelis *Hum Mutat* 29:1125, 2006[85]	AD	164840
NFIX	Nuclear factor IX	Malan *Am J Hum Genet* 87:189, 2010[86]	AD	164005
NIPBL	NIPBL, cohesin loading factor	Rohatgi *Am J Med Genet A* 152A:1641, 2010[87]	AD	608667
NOTCH2	Notch 2	Kamath *Nat Rev Nephrol* 9:409, 2013[77]	AD	600275
PAX8	Paired box 8	Meeus *J Clin Endocrinol Metab* 89:4285, 2004[88]	AD	167415
PROK2	Prokineticin 2	Madan *Mol Genet Metab Rep* 12:57, 2017[89]	AD	607002
PROKR2	Prokineticin receptor 2	Sarfati *Front Horm Res* 39:121, 2010[90]	AD	607123
PTEN	Phosphatase and tensin homolog	Reardon *J Med Genet* 38:820, 2001[91]	AD	601728
PTPN11	Protein tyrosine phosphatase, non-receptor type 11	Bertola *Am J Med Genet A* 130A:378, 2004[16]	AD	176876
RAF1	Raf-1 proto-oncogene, serine/threonine kinase	Razzaque *Nat Genet* 39:1013, 2007[92]	AD	164760
RAI1	Retinoic acid induced 1	Vilboux *PLoS One* 6:e22861, 2011[93]	AD	607642
SALL4	Spalt like transcription factor 4	Kohlhase *GeneReviews®Book Section,* 1993[94]	AD	607343
SEMA3E	Semaphorin 3E	Lalani *J Med Genet* 41:e94, 2004[95]	AD	608166
SETBP1	SET binding protein 1	Schinzel *Am J Med Genet* 1:361, 1978[96]	AD	611060
SF3B4	Splicing factor 3b subunit 4	Bernier *Am J Hum Genet* 90:925, 2012[97]	AD	605593
SHH	Sonic hedgehog	Lurie Am *J Med Genet* 35:286, 1990[98]	AD	600725

Gene	Encoded protein	Reference	MOI	OMIM Gene number
SNRPB	Small nuclear ribonucleoprotein polypeptides B and B1	Tooley *Am J Med Genet A* 170A:1115, 2016[99]	AD	182282
SOS1	SOS Ras/Rac guanine nucleotide exchange factor 1	Ferrero *Eur J Med Genet* 51:566, 2008[100]	AD	182530
SOX9	SRY-box 9	Airik *Hum Mol Genet* 19:4918, 2010[101]	AD	608160
SRCAP	Snf2 related CREBBP activator protein	Hood *Am J Hum Genet* 90:308, 2012[102]	AD	611421
TBX1	T-box 1	Kujat *Am J Med Genet A* 140:1601, 2006[103]	AD	602054
TBX3	T-box 3	Meneghini *Eur J Med Genet* 49:151, 2006[104]	AD	601621
TFAP2A	Transcription factor AP-2 alpha	Milunsky *Am J Hum Genet* 82:1171, 2008[105]	AD	107580
TP63	Tumor protein P63	Celli *Cell* 99:143, 1999[106]	AD	603273
TSC1	Tuberous sclerosis 1	Curatolo *Lancet* 372:657, 2008[107]	AD	605284
TSC2	Tuberous sclerosis 2	Kumar *Hum Mol Genet* 4:1471, 1995[108]	AD	191092
TWIST2	Twist family BHLH transcription factor 2	Stevens *Am J Med Genet* 107:30, 2002[109]	AD	607556
VANGL1	VANGL planar cell polarity protein 1	Bartsch *Mol Syndromol* 3:76, 2012[110]	AD	610132
WNT5A	Wnt family member 5A	Roifman *Clin Genet* 87:34, 2015; Person *Dev Dyn* 239:327, 2010[111]	AD	164975
AMER1	APC membrane recruitment protein 1	Pellegrino *Am J Med Genet* 16:159, 1997[112]	XL	300647
BCOR	BCL6 corepressor	Ng *Nat Genet* 36:411, 2004[113]	XL	300485
FANCB	Fanconi anemia complementation group B	McCauley *Am J Med Genet A* 155A:2370, 2011[114]	XL	300515
FLNA	Filamin A	Robertson *Am J Med Genet A* 140:1726, 2006[115]	XL	300017
GPC3	Glypican 3	Cottereau *Am J Med Genet C Semin Med Genet* 163:92, 2013[116]	XL	300037
KAL1	Anosmin 1	Hardelin *Proc Natl Acad Sci U S A* 89:8190, 1992[117]	XL	300836
KDM5C	Lysine-specific demethylase 5C	Jensen *Am J Hum Genet* 76:227, 2005[118]	XL	314690
KDM6A	Lysine-specific demethylase 6A	Rosenberg *Am J Med Genet* 182:85, 2020[119]	XL	300128
MID1	Midline 1	Preiksaitiene *Clin Dysmorphol* 24:7, 2015[120]	XL	300552
NSDHL	NAD(P) dependent steroid dehydrogenase-like	König *J Am Acad Dermatol* 46:594, 2002[121]	XL	300275
PIGA	Phosphatidylinositol glycan anchor biosynthesis class A	Johnston *Am J Hum Genet* 90:295, 2012[122]	XL	311770
PORCN	Porcupine O-acyltransferase	Suskan *Pediatr Dermatol* 7:283, 1990[123]	XL	300651
SMC1A	Structural maintenance of chromosomes 1A	Deardorff *GeneReviews® Book Section Seattle(WA),* 1993[124]	XL	300040
UPF3B	UPF3B, regulator of nonsense mediated MRNA decay	Lynch *Eur J Med Genet* 55:476, 2012[125]	XL	300298
ZIC3	Zic family member 3	Chung *Am J Med Genet* 155:1123, 2011[126]	XL	300265

AD, autosomal dominant; **AR**, autosomal recessive; **MOI**, mode of inheritance; **OMIM**, Online Mendelian Inheritance in Men; **XL**, X-linked

Supplementary Table 3. 243 Monogenic causes of murine CAKUT.

243 genes that if mutated cause murine CAKUT phenotypes were retrieved from the MGI database (http://www.informatics.jax.org/) by searching for the following terms: “abnormal mesonephric mesenchyme”, “abnormal mesonephric mesenchyme morphology”, “abnormal metanephric mesenchyme”, “abnormal metanephric mesenchyme morphology”, “abnormal metanephric ureteric bud development”, “abnormal ureter development”, “abnormal ureter morphology”, “abnormal ureteric bud elongation”, “abnormal ureteric bud invasion”, “abnormal ureterovesical junction”, “abnormal urinary system development”, “absent kidney”, “absent metanephric mesenchyme”, “absent metanephros”, “absent ureter”, “dilated ureter”, “double kidney pelvis”, “double kidney pelvis”, “double ureter”, “duplex kidney”, “ectopic ureter”, “ectopic ureteric bud”, “hydroureter”, “impaired branching involved in ureteric bud morphogenesis”, “pelvic kidney”, “renal hypoplasia”, “short ureter”, “single kidney”, “small metanephros”, “ureter hypoplasia”, “abnormal nephrogenic mesenchyme morphogenesis”, “ureteropelvic junction obstruction”. Mouse CAKUT genes are listed together with the corresponding human phenotype that was extracted from the OMIM database (www.omim.org). Human phenotypes representing CAKUT are underlined (source: clinical synopsis table, OMIM, www.omim.org). These murine genes represent candidate genes for human CAKUT.

Gene	Protein	PubMed ID/ MGI Ref. ID	Human Phenotype (contains CAKUT)	Human phenotype OMIM #
Ace	Angiotensin I converting enzyme	8642790	Renal tubular dysgenesis	267430
Acvr2b	Activin A Receptor Type 2B	9242489	Heterotaxy, visceral, 4, autosomal	613751
Adamts1	ADAM Metallopeptidase With Thrombospondin Type 1 Motif 1	10811842	-	-
Adamts3	ADAM Metallopeptidase With Thrombospondin Type 1 Motif 3	J:239583	Hennekam lymphangiectasia-lymphedema syndrome 3	618154
Adamts18	ADAM Metallopeptidase With Thrombospondin Type 1 Motif 18	31242448	Microcornea, myopic chorioretinal atrophy, and telecanthus	615458
Agt	Angiotensinogen	8675666	Renal tubular dysgenesis	267430
Agtr1a	Angiotensin II Receptor, Type 1a	10024874	Renal tubular dysgenesis	267430
Agtr1b	Angiotensin II Receptor, Type 1b	10024874	Renal tubular dysgenesis	267430
Agtr2	Angiotensin II Receptor Type 2	10024874	-	-
Aldh1a2	Aldehyde Dehydrogenase 1 Family Member A2	20040494	-	-
Amer1	APC Membrane Recruitment Protein 1	21571217	Osteopathia striata with cranial sclerosis	300373
Anp32b	Acidic Nuclear Phosphoprotein 32 Family Member B	21636789	-	-
Aprt	Adenine Phosphoribosyltransferase	8864750	Adenine phosphoribosyl-transferase deficiency	614723
Aqp2	Aquaporin 2	3184310	Diabetes insipidus, nephrogenic	125800
Arhgap1	Rho GTPase Activating Protein 1	17227869	-	-
Arhgap35	Rho GTPase Activating Protein 35	26859289	-	-
Arid5b	AT-Rich Interaction Domain 5B	17143286	-	-
Arl3	ADP Ribosylation Factor Like GTPase 3	16565502	-	-
Atmin	ATM Interactor	24852369	-	-
Atp6ap2	ATPase, H+ Transporting, Lysosomal Accessory Protein 2	23704941	?Parkinsonism with spasticity, X-linked; Congenital disorder of glycosylation, type IIr; Intellectual developmental disorder, X-linked syndromic, Hedera type	300911; 301045; 300423

Atp7a	ATPase Copper Transporting Alpha	11534785	Menkes disease; Occipital horn syndrome; Spinal muscular atrophy, distal, X-linked 3	309400; 304150; 300489
Axin1	Axin 1	17246824 13340237	Hepatocellular carcinoma, somatic	114550
B9d2	B9 Protein Domain 2	J:239583	Joubert syndrome 34; ?Meckel syndrome 10	614175
Bag6	BCL2 Associated Athanogene 6	16287848	-	-
Bcl2	BCL2, Apoptosis Regulator	8623928	Leukemia/ lymphoma, B-cell, 2	n/a
Bdkrb2	Bradykinin Receptor, Beta 2	11015607 12560214	-	-
Bicc1	BicC Family RNA Binding Protein 1	8887273	{Renal dysplasia, cystic, susceptibility to}	601331
Bmp4	Bone Morphogenetic Protein 4	10749566	Microphthalmia, syndromic 6	607932
Bmp5	Bone Morphogenetic Protein 5	5692092	-	-
Bmp7	Bone Morphogenetic Protein 7	7590254	-	-
Bmper	BMP Binding Endothelial Regulator	17035289	Diaphano-spondylo-dysostosis	608022
Btbd1	BTB (POZ) Domain Containing 1	J:211773	-	-
Btbd7	BTB (POZ) Domain Containing 7	28506999	-	-
Brca1	Breast Cancer 1, Early Onset	17653086	Fanconi anemia, complementation group S; {Breast-ovarian cancer, familial, 1}; {Pancreatic cancer, susceptibility to, 4}	617883; 604370; 614320
Cbs	Cystathionine Beta-Synthase	7878023	Thrombosis, hyperhomocysteinemic; Homocystinuria, B6-responsive and nonresponsive types	236200
Cbwd1	COBW Domain Containing 1	31862704	-	-
Cc2d2a	Coiled-Coil And C2 Domain Containing 2A	J:175213	COACH syndrome; Joubert syndrome 9; Meckel syndrome 6	216360; 612285; 612284
Cdc42	Cell Division Cycle 42	23555292	Takenouchi-Kosaki syndrome	616737
Cdh4	Cadherin 4	11839813	-	-
Cdh6	Cadherin 6	10864459	-	-
Cdk13	Cyclin-Dependent Kinase 13	31440507	Congenital heart defects, dysmorphic facial features, and intellectual developmental disorder	617360
Cdkn1c	Cyclin-Dependent Kinase Inhibitor 1C (P57)	19276117	Beckwith-Wiedemann syndrome; IMAGE syndrome	130650; 614732
Chrm3	Cholinergic Receptor Muscarinic 3	10944224	Prune belly syndrome	100100
Chst11	Carbohydrate Sulfotransferase 11	J:239583	?Osteochondrodysplasia, brachydactyly, and overlapping malformed digits	618167
Chtop	Chromatin Target of PRMT1	J:239583	-	-
Cilk1	Ciliogenesis Associated Kinase 1	24853502	Endocrine-cerebroosteodysplasia; {Epilepsy, juvenile myoclonic, susceptibility to, 10}	612651; 617924
Cir1	Corepressor Interacting With RBPJ, 1	J:239583	-	-
Cited2	Cbp/p300-Interacting Transactivator, With Glu/Asp-rich Carboxy-terminal Domain, 2	17615577	Atrial septal defect 8; Ventricular septal defect 2	614433; 614431
Cldn4	Claudin 4	23284964	-	-
Clmp	CXADR-like Membrane Protein	29361518	Congenital short bowel syndrome	615237
Cntrl	Centriolin	J:175213	-	-
Col4a1	Collagen, Type IV, Alpha 1	26260163	Angiopathy, hereditary, with nephropathy, aneurysms, and muscle cramps	611773
Crb3	Crumbs 3, Cell Polarity Complex Component	26631503	-	-
Crim1	Cysteine Rich Transmembrane BMP Regulator 1	22511315	-	-
Ctdnep1	CTD Nuclear Envelope Phosphatase 1	23360989	-	-
Ctnnb1	Catenin Beta 1	20454682	Colorectal cancer, somatic; Hepatocellular carcinoma, somatic; Medullo-blastoma, somatic; Mental retardation, autosomal dominant 19; Ovarian cancer, somatic; Pilomatricoma, somatic	114500; 114550; 155255; 615075; 167000; 132600
Ctnnbip1	Catenin Beta Interacting Protein 1	17803964	-	-
Ctnnd1	Catenin (Cadherin Associated Protein), Delta 1	21521738	Blepharocheilodontic syndrome 2	617681
Cxcr4	C-X-C Motif Chemokine Receptor 4	J:175213	WHIM syndrome	193670
Cyfip2	Cytoplasmic FMR1 Interacting Protein 2	J:239583	Developmental and epileptic encephalopathy 65	618008
Cyp26a1	Cytochrome P450 Family 26 Subfamily A Member 1	11157778	-	-
Dact1	Dishevelled Binding Antagonist Of Beta Catenin 1	20145239	-	-

Dchs1	Dachsous Cadherin-Related 1	21303848	Mitral valve prolapse 2; Van Maldergem syndrome 1	607829; 601390
Dhcr7	7-Dehydrocholesterol Reductase	11230174	Smith-Lemli-Opitz syndrome	270400
Dis3l2	DIS3 Like 3'-5' Exoribonuclease 2	29950491	Perlman syndrome	267000
Dlg1	Discs Large MAGUK Scaffold Protein 1	17172448	-	-
Dlg5	Discs Large MAGUK Scaffold Protein 5	17765678	{Inflammatory bowel disease 20}	612288
Dnah11	Dynein Axonemal Heavy Chain 11	J:175213	Ciliary dyskinesia, primary, 7, with or without situs inversus	611884
Dnah5	Dynein Axonemal Heavy Chain 5	J:175213	Ciliary dyskinesia, primary, 3, with or without situs inversus	608644
Drosha	Drosha, Ribonuclease Type III	30367465	-	-
Dspp	Dentin Sialophosphoprotein	16937369	Deafness, autosomal dominant 39, with dentinogenesis; Dentin dysplasia, type II; Dentinogenesis imperfecta, Shields type II; Dentinogenesis imperfecta, Shields type III	605594; 125420; 125490; 125500
Dym	Dymeclin	18852472	Dyggve-Melchior-Clausen disease; Smith-McCort dysplasia	223800; 607326
Dync2h1	Dynein Cytoplasmic 2 Heavy Chain 1	J:175213	Short-rib thoracic dysplasia 3 with or without polydactyly	613091
Edc3	Enhancer Of mRNA Decapping 3	J:211773	?Intellectual developmental disorder, autosomal recessive 50	616460
Efnb2	Ephrin B2	15223334	-	-
Emx2	Empty Spiracles Homeobox 2	9165114	Schizencephaly	269160
Esrrg	Estrogen Related Receptor Gamma	21138943	-	-
Etl4/Etn2	Early transposon element insertion site 2	23436999	Epilepsy, familial temporal lobe, 4	611631
Etv4	ETS Variant 4	19898483	-	-
Etv5	ETV Variant 5	19898483	-	-
Exoc5	Exocyst Complex Component 5	26046524	-	-
Eya1	EYA Transcriptional Coactivator And Phosphatase 1	10471511	Branchiooto-renal syndrome 1, with or without cataracts; Branchiootic syndrome 1; Anterior segment anomalies with or without cataract; Otofaciocervical syndrome	113650; 602588; 602588; 166780
Fat4	FAT Atypical Cadherin 4	21303848	Van Maldergem syndrome 2; Hennekam lymph-angiectasia-lymphedema syndrome 2	615546; 616006
Fbxw15	F-Box And WD-40 Domain Protein 15	J:211773	-	-
Fgf10	Fibroblast Growth Factor 10	11062007	Aplasia of lacrimal and salivary glands; LADD syndrome	180920; 149730
Fgf7	Fibroblast Growth Factor 7	9876183	-	-
Fgf8	Fibroblast Growth Factor 8	16049111	Hypo-gonadotropic hypogonadism 6 with or without anosmia	612702
Fgfr2	Fibroblast Growth Factor Receptor 2	15843416	Antley-Bixler syndrome; Apert syndrome; Beare-Stevenson cutis gyrata syndrome; Bent bone dysplasia syndrome; Craniofacial-skeletal-dermatologic dysplasia; Crouzon syndrome; Gastric cancer, somatic; Jackson-Weiss syndrome; LADD syndrome; Pfeiffer syndrome; Saethre-Chotzen syndrome; Scaphocephaly, maxillary retrusion, and mental retardation	207410; 101200; 123790; 614592; 101600; 123500; 613659; 123150; 149730; 101600; 101400; 609579
Fgfrl1	Fibroblast Growth Factor Receptor-Like 1	19715689	-	-
Fmn1	Formin 1	7517224	-	-
Foxc1	Forkhead Box C1	5500588	Anterior segment dysgenesis 3, multiple subtypes; Axenfeld-Rieger syndrome, type 3	601631; 602482
Foxc2	Forkhead Box C2	16498405	Lymphedema-distichiasis syndrome with renal disease and diabetes mellitus	153400
Foxd1	Forkhead Box D1	8666231	-	-
Foxd2	Forkhead Box D2	10648626	-	-
Foxg1	Forkhead Box G1	16109771	Rett syndrome, congenital variant	613454
Fras1	Fraser Extracellular Matrix Complex Subunit 1	12766769	Fraser syndrome	219000
Frem1	FRAS1 Related Extracellular Matrix 1	12766769	Bifid nose with or without anorectal and renal anomalies; Manitoba oculotrichoanal syndrome; Trigonocephaly 2	608980; 248450; 614485
Frem2	FRAS1 Related Extracellular Matrix Protein 2	12766769	Fraser syndrome	219000

Fryl	FRY Like Transcription Coactivator	29409347	-	-
Fstl1	Follistatin Like 1	22485132	-	-
Fzd4	Frizzled Class Receptor 4	21343368	Exudative vitreoretinopathy 1; Retinopathy of prematurity	133780; 133780
Fzd8	Frizzled Class Receptor 8	21343368	-	-
Galc	Galactosylceramidase	6319815	Krabbe disease	245200
Gas2l2	Growth Arrest-Specific 2 Like 2	J:239583	?Ciliary dyskinesia, primary, 41	618449
Gata2	GATA Binding Protein 2	18233958	Emberger syndrome; Immunodeficiency 21	614038; 614172
Gata3	GATA Binding Protein 3	16319112	Hypopara-thyroidism, sensorineural deafness, and renal dysplasia	146255
Gdf11	Growth Differentiation Factor 11	12729564	-	-
Gdnf	Glial Cell Derived Neurotrophic Factor	11422733	Central hypoventilation syndrome	209880
Gen1	GEN1, Holliday Junction 5' Flap Endonuclease	29483821	-	-
Gfra1	GDNF Family Receptor Alpha 1	23542432	-	-
Glce	Glucuronic Acid Epimerase	12788935	-	-
Gli3	GLI Family Zinc Finger 3	11978771	Greig cephalopoly-syndactyly syndrome; Pallister-Hall syndrome; Polydactyly, postaxial, types A1 and B; Polydactyly, preaxial, type IV	175700; 146510; 174200; 174700
Gpc3	Glypican 3	10402475	Simpson-Golabi-Behmel syndrome, type 1; Wilms tumor, somatic	312870; 194070
Greb1l	Growth Regulation By Estrogen In Breast Cancer-Like	29100091	Deafness, autosomal dominant 80; Renal hypodysplasia/aplasia 3	619274; 617805
Grem1	Gremlin 1, DAN Family BMP Antagonist	15201225	-	-
Grip1	Glutamate Receptor Interacting Protein 1	10974668	Fraser syndrome	219000
H13	Histocompatibility 13	J:239583	-	-
Hnf1b	HNF1 Homeobox B	23362348	Diabetes mellitus, noninsulin-dependent; Renal cysts and diabetes syndrome	125853; 137920
Hoxa11	Homeobox A11	7596412	Radioulnar synostosis with amegakaryo-cytic thrombocyto-penia 1	605432
Hoxd11	Homeobox D11	12050119	-	-
Hoxa13	Homeobox A13	12783783	Guttmacher syndrome; Hand-foot-uterus syndrome	176305; 140000
Hoxc10	Homeobox C10	19623272	-	-
Hoxc11	Homeobox C11	12050119	-	-
Hpse2	Heparanase 2	25510506	Urofacial syndrome 1	236730
Hs2st1	Heparan Sulfate 2-O-Sulfotransferase 1	9637690	-	-
Hsd17b2	Hydroxysteroid 17-Beta Dehydrogenase 2	18048640	-	-
Hspa4l	Heat Shock Protein Family A (Hsp70) Member 4 Like	16923965	-	-
Htr3a	5-Hydroxytryptamine Receptor 3A	15201326	-	-
Id2	Inhibitor Of DNA Binding 2, HLH Protein	15569159	-	-
Ilk	Integrin Linked Kinase	19829382	-	-
Itga3	Integrin Subunit Alpha 3	10433923	Interstitial lung disease, nephrotic syndrome, and epidermolysis bullosa, congenital	614748
Itga6	Integrin Subunit Alpha 6	10433923	Epidermolysis bullosa, junctional, with pyloric stenosis	226730
Itga8	Integrin Subunit Alpha 8	9054500 17537792	Renal hypodysplasia/ aplasia 1	191830
Itgb1	Integrin Subunit Beta 1	19439520	-	-
Kavh	Kidney Adysplasia And Variable Hydronephrosis	25834070	-	-
Kif26b	Kinesin Family Member 26B	20439720	-	-
Krt33b	Keratin 33B	J:211773	-	-
Lama5	Laminin Subunit Alpha 5	10625553	-	-
Lamc1	Laminin Subunit Gamma 1	12015298	-	-
Lgr4	Leucine Rich Repeat Containing G Protein-Coupled Receptor 4	21523854 22738954	{Bone mineral density, low, susceptibility to}	615311
Lhx1	Luteinizing Hormone/Choriogonadotropin Receptor	16216236	-	-
Lin7c	Lin-7 Homolog C, Crumbs Cell Polarity Complex Component	17923534	-	-
Lifr	LIF receptor alpha	28334964	Stuve-Wiedemann syndrome/Schwartz-Jampel type 2 syndrome	601559
Lrp4	LDL Receptor Related Protein 4	20454682	Sclerosteosis 2; Cenani-Lenz syndactyly syndrome; ?Myasthenic syndrome, congenital, 17	614305; 212780; 616304
Lzts2	Leucine Zipper Tumor Suppressor 2	21949185	-	-

Maz	MYC-Associated Zinc Finger Protein (Purine-Binding Transcription Factor)	29432158	-	-
Megf8	Multiple EGF Like Domains 8	18043505	Carpenter syndrome 2	614976
Mks1	Meckel Syndrome, Type 1	21045211	Meckel syndrome 1	249000
Mmp14	Matrix Metallopeptidase 14	20727881	?Winchester syndrome	277950
Mmp17	Matrix Metallopeptidase 17	21347258	-	-
Morc2a	Microrchidia 2A	J:239583	-	-
Mybphl	Myosin Binding Protein H-like	J:239583	-	-
Mycn	V-Myc Avian Myelocytomatosis Viral Oncogene Neuroblastoma Derived Homolog	1459449	Feingold syndrome 1	164280
Ncf1	Neutrophil Cytosolic Factor 1	J:211773	Chronic granulomatous disease 1, autosomal recessive	233700
Ndst1	N-Deacetylase And N-Sulfotransferase 1		Mental retardation, autosomal recessive 46	616116
Nf1	Neurofibromin 1	7926784	Neurofibromatosis, type 1	162200
Nfia	Nuclear Factor I A	17530927	-	-
Nmnat2	Nicotinamide Nucleotide Adenylyltransferase 2	23082226	-	-
Nog	Noggin	18028901	Brachydactyly, type B2; Multiple synostoses syndrome 1; Stapes ankylosis with broad thumb and toes; Symphalangism, proximal, 1A; Tarsal-carpal coalition syndrome	611377; 186500; 184460; 185800; 186570
Notch2	Notch 2	20299358	Alagille syndrome 2; Hajdu-Cheney syndrome	610205; 102500
Notum	Notum Palmitoleoyl-Protein Carboxylesterase	26926082	-	-
Npat	Nuclear Protein In The AT Region	J:239583	-	-
Npnt	Nephronectin	17537792	-	-
Osr1	Odd-Skipped Related Transciption Factor 1	16790474	-	-
Parva	Parvin Alpha	19829382	-	-
Pax2	Paired Box 2	8575306	Papillorenal syndrome; Glomerulo-sclerosis, focal segmental, 7	120330; 616002
Pax8	Paired Box 8	12435636	Hypothyroidism, congenital, due to thyroid dysgenesis or hypoplasia	218700
Pbx1	PBX Homeobox 1	12591246	Leukemia, acute pre-B-cell	176310
Pcnt	Pericentrin (Kendrin)	25220058	Microcephalic osteodysplastic primordial dwarfism, type II	210720
Pcsk5	Proprotein Convertase Subtilisin/Kexin Type 5	18519639	-	-
Pdgfra	Platelet Derived Growth Factor Receptor Alpha	19217431	Gastrointestinal stromal tumor, somatic; Hyper-eosinophilic syndrome, idiopathic, resistant to imatinib	606764; 607685
Pds5a	PDS5 Cohesin Associated Factor A	19412548	-	-
Plxnb1	Plexin B1	18799546	-	-
Plxnb2	Plexin B2	21035938	-	-
Plxnd1	Plexin D1	J:175213	-	-
Ppp3r1	Protein Phosphatase 3 Regulatory Subunit B, Alpha	15057312	-	-
Prickle1	Prickle Planar Cell Polarity Protein 1	25190059	Epilepsy, progressive myoclonic 1B	612437
Ptch1	Patched 1	22792366	Basal cell carcinoma, somatic; Basal cell nevus syndrome; Holopros-encephaly 7	605462; 109400; 610828
Pten	Phosphatase And Tensin Homolog	17540362	Bannayan-Riley-Ruvalcaba syndrome; Cowden syndrome 1; Endometrial carcinoma, somatic; Lhermitte-Duclos syndrome; Macrocephaly/autism syndrome; Malignant melanoma, somatic; Squamous cell carcinoma, head and neck, somatic; VATER association with macrocephaly and ventriculo-megaly	153480; 158350; 608089; 158350; 605309; 155600; 275355; 276950
Ptpn14	Protein Tyrosine Phosphatase, Non-Receptor Type 14	J:211773	Choanal atresia and lymphedema	613611
Ptprf	Protein Tyrosine Phosphatase, Receptor Type F	19273906	?Breasts and/or nipples, aplasia or hypoplasia of, 2	616001
Pygo1	Pygopus Family PHD Finger 1	17425782	-	-
Pygo2	Pygopus Family PHD Finger 2	17425782	-	-
Rala	V-Ral Simian Leukemia Viral Oncogene A (Ras Related)	J:239583	Hiatt-Neu-Cooper neurodevelopmental syndrome	619311
Rara	Retinoic Acid Receptor Alpha	9376317	Leukemia, acute promyelocytic	612376
Rdh10	Retinol Dehydrogenase 10 (All-Trans)	21930923 17473173	-	-

Rere	Arginine-Glutamic Acid Dipeptide Repeats	23451234	Neurodevelopmental disorder with or without anomalies of the brain, eye, or heart	616975
Ret	Ret Proto-Oncogene	16452504	Central hypoventilation syndrome, congenital; Medullary thyroid carcinoma; Multiple endocrine neoplasia IIA; Multiple endocrine neoplasia IIB; Pheochromo-cytoma	209880; 155240; 171400; 162300; 171300
Rnf223	Ring Finger 223	J:211773	-	-
Robo1	Roundabout Guidance Receptor 1	J:175213	-	-
Robo2	Roundabout Guidance Receptor 2	17357069	Vesicoureteral reflux 2	610878
Rr2	Regulatory Region 2	17803355	-	-
Rspo2	R-Spondin 2	17904116 12782276	-	-
Sall1	Spalt Like Transcription Factor 1	11688560	Townes-Brocks syndrome; Townes-Brocks branchio-otorenal-like syndrome	107480
Sall4	Spalt Like Transcription Factor 4	17216607	Duane-radial ray syndrome; IVIC syndrome	607323; 147750
Sc5d	Sterol-C5-Desaturase	J:175213	Lathosterolosis	607330
Scarb2	Scavenger Receptor Class B Member 2	12620969	Epilepsy, progressive myoclonic 4, with or without renal failure	254900
Sema3a	Semaphorin 3A	18249526	{Hypogonado-tropic hypogonadism 16 with or without anosmia}	614897
Sestd1	SEC14 And Spectrin Domain Containing 1	23696638	-	-
Sh3pxd2a	SH3 And PX Domains 2A	J:239583	-	-
Shh	Sonic Hedgehog	12399320	Holopros-encephaly 3; Microphthalmia with coloboma 5; Schizencephaly; Single median maxillary central incisor	142945; 611638; 269160; 147250
Six1	SIX Homeobox 1	14695375	Branchiootic syndrome 3; Deafness, autosomal dominant 23	608389; 605192
Six2	SIX Homeobox 2	17036046	-	-
Smad4	SMAD Family Member 4	15342483	Juvenile polyposis/hereditary hemorrhagic telangiectasia syndrome; Myhre syndrome; Pancreatic cancer, somatic; Polyposis, juvenile intestinal	175050; 139210; 260350; 174900
Slc25a20	Solute Carrier Family 25 (Mitochondrial Carnitine/Acylcarnitine Translocase), Member 20	J:239583	Carnitine-acylcarnitine translocase deficiency	212138
Slit2	Slit Guidance Ligand 2	15130495	-	-
Slit3	Slit Guidance Ligand 3	14550534	-	-
Smg9	SMG9 Nonsense Mediated mRNA Decay Factor	J:239583	Heart and brain malformation syndrome; Neurodevelopmental disorder with intention tremor, pyramidal signs, dyspraxia, and ocular anomalies	616920; 619995
Smpd4	Sphingomyelin Phosphodiesterase 4	J:239583	Neurodevelopmental disorder with microcephaly, arthrogryposis, and structural brain anomalies	618622
Sox4	SRY-Box 4	16109771	-	-
Sox9	SRY-Box 9	20881014	Acampomelic campomelic dysplasia; Campomelic dysplasia; Campomelic dysplasia with autosomal sex reversal	114290
Spink13	Serine Peptidase Inhibitor, Kazal Type 13	J:211773	-	-
Spry1	Sprouty RTK Signaling Antagonist 1	15691764	-	-
Sulf1	Sulfatase 1/ Sulfatase 2	17593974	-	-
Sulf2	Sulfatase 1/ Sulfatase 2	17593974	-	-
Tbc1d32	TBC1 Domain Family Member 32	J:175213	-	-
Tbx18	T-Box 18	24016759	Congenital anomalies of kidney and urinary tract 2	143400
Tbx6	T-Box 6	4073528	Spondylocostal dysostosis 5	122600
Tcf21	Transcription Factor 21	10572052	-	-
Tfcp2l1	Transcription Factor CP2-Like 1	17079272	-	-
Tgfb2	Transforming Growth Factor Beta 2	9217007	Loeys-Dietz syndrome 4	614816
Timp2	Tissue Inhibitor Of Metalloproteinase 2	J:211773	-	-
Trp53	Transformation related protein 53	11780111	Adrenal cortical carcinoma; Breast cancer; Choroid plexus papilloma; Colorectal cancer; Hepatocellular carcinoma; Li-Fraumeni	202300; 114480; 260500; 114500;

			syndrome; Nasopharyngeal carcinoma; Osteosarcoma; Pancreatic cancer	114550; 151623; 607107; 259500; 260350
Trps1	Transcriptional Repressor GATA Binding 1	19820125	Trichorhino-phalangeal syndrome, type I; Trichorhino-phalangeal syndrome, type III	190350; 190351
Tshz3	Teashirt Zinc Finger Homeobox 3	18776146	-	-
Tyr	Tyrosinase	J:179802	-	-
Umod	Uromodulin	15611339	Medullary cystic kidney disease 2; Hyperuricemic nephropathy, familial juvenile 1; Glomerulocystic kidney disease with hyperuricemia and isosthenuria	603860; 162000; 609886
Unk	Unkempt Family Zinc Finger	J:239583	-	-
Upk3a	Uroplakin 3A	11085999	-	-
Vamp5	Vesicle-Associated Membrane Protein 5	29330887	-	-
Vstm2a	V-Set And Transmembrane Domain Containing 2A	J:211773	-	-
Wasl	WAS/WASL Interacting Protein Family Member 1	23555292	-	-
Wdpcp	WD Repeat Containing Planar Cell Polarity Effector	24302887	?Congenital heart defects, hamartomas of tongue, and polysyndactyly	217085
Wnt11	Wnt Family Member 11	12783789	-	-
Wnt4	Wnt Family Member 4	7990960	Mullerian aplasia and hyperandro-genism; ?SERKAL syndrome	158330; 611812
Wnt5a	Wnt Family Member 5A	J:175213	Robinow syndrome, autosomal dominant 1	180700
Wnt7b	Wnt Family Member 7B	19060336	-	-
Wnt9b	Wnt Family Member 9B	16054034	-	-
Wt1	Wilms Tumor 1	18040647	Denys-Drash syndrome; Frasier syndrome; Meacham syndrome; Mesothelioma, somatic; Nephrotic syndrome, type 4; Wilms tumor, type 1	194080; 136680; 608978; 156240; 256370; 194070
Xpl	X-linked polydactyly	7391545	Orofaciodigital syndrome I	311200
Yap1	Yes Associated Protein 1	23555292	Coloboma, ocular; Coloboma, ocular, with or without hearing impairment, cleft lip/palate, and/or mental retardation	120433
Zbtb14	Zinc Finger And BTB Domain Containing 14	J:175213	-	-
Zc3h8	Zinc Finger CCCH Type Containing 8	J:211773	-	-

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