Table SI. Causative genes for primary ciliopathies.

OMIM ID	Gene name	Cytogenetic location	Disease association
600294	ADCY6	12q13.12	Lethal congenital contracture syndrome; arthrogryposis multiplex congenita syndrome
608894	AHI1	6q23.3	JBTS
606844	ALMS1	2p13.1	ALMS
615370	ANKS6	9q22.33	NPHP; NPHP with cardiovascular abnormalities, situs inversus and liver fibrosis
608922	ARL13B	3q11.1-q11.2	JBTS; JBTS with retinal degeneration, obesity
615407	ARL2BP	16q13	RP
604695	ARL3	10q24.32	RP; JBTS
608845	ARL6	3q11.2	BBS; BBS modifier; RP
611150	ATXN10	22q13.31	JBTS; NPHP; spinocerebellar ataxia
614144	B9D1	17p11.2	MKS; JBTS
611951	B9D2	19q13.2	MKS; JBTS
613605	BBIP1	10q25.2	BBS
209901	BBS1	11q13.2	BBS
610148	BBS10	12q21.2	BBS
610683	BBS12	4q27	BBS
606151	BBS2	16q13	BBS
600374	BBS4	15q24.1	BBS
603650	BBS5	2q31.1	BBS
607590	BBS7	4q27	BBS
607968	PTHB1	7p14.3	BBS
603191	C21ORF2	21q22.3	CRD; axial spondylometaphyseal dysplasia
615944	C2CD3	11q13.4	OFD
613425	C2ORF71	2p23.2	RP
614571	C5ORF42	5p13.2	JBTS; OFD; monomelic amyotrophy
614477	C8ORF37	8q22.1	RP; CRD; BBS
612013	CC2D2A	4p15.32	MKS; JBTS; COACH syndrome
610162	CCDC28B	1p35.2	BBS modifier; JBTS
600236	CENPF	1q41	Stromme syndrome
616690	CEP104	1p36.32	JBTS
613446	CEP120	5q23.2	SRTD; JBTS
614848	CEP164	11q23.3	NPHP with retinal degeneration; probably SLSN, MKS, JBTS
615586	CEP19	3q29	Morbid obesity and spermatogenic failure (MOSPGF)
610142	CEP290	12q21.32	MKS; BBS; JBTS; LCA; SLSN
610523	CEP41	7q32.2	JBTS; autism spectrum disorder
617110	CEP78	9q21.2	CRD; hearing loss
615847	CEP83	12q22	NPHP; intellectual disability
616787	CLUAP1	16p13.3	LCA
123825	CNGA1	4p12	RP
300338	CNGA2	Xq28	Isolated congenital anosmia
600724	CNGB1	16q21	RP
602225	CRX	19q13.33	LCA; CRD
611654	CSPP1	8q13.1-q13.2	MKS; JBTS; SRTD
605755	DCDC2	6p22.3	NPHP; non-syndromic recessive deafness; neonatal sclerosing cholangitis
615464	DDX59	1q32.1	OFD
603297	DYNC2H1	11q22.3	SRTD
617083	DYNC2LI1	2p21	SRTD
617610	DZIP2	3q22.3	ARPKD
608815	EFHC1	6p12.2	Juvenile myoclonic epilepsy
604831	EVC	4p16.2	EVC; Weyers acrofacial dysostosis
607261	EVC2	4p16.2	EVC; Weyers acrofacial dysostosis

Table SI. Continued.

OMIM ID	Gene name	Cytogenetic location	Disease association
618413	FAM149B1(KIAA0974)	10q22.2	JBTS
613596	FAM161A	2p15	RP
300708	FAM58A	Xq28	STAR syndrome
607273	FLCN	17p11.2	Birt-Hogg-Dubé syndrome
165230	GLI2	2q14.2	Holoprosencephaly; Culler-Jones syndrome
165240	GLI3	7p14.1	Greig cephalopolysyndactyly syndrome; Pallister-Hall syndrome
608539	GLIS2	16p13.3	NPHP
612250	GPR161	1q24.2	Pituitary stalk interruption syndrome
610693	HYLS1	11q24.2	Hydrolethalus; JBTS (OKA2016)
612325	ICK	6p12.1	Lethal endocrine-cerebro-osteodysplasia syndrome; SRTD
606045	IFT122	3q21.3-q22.1	CED
614620	IFT140	16p13.3	SRTD; RP; LCA; skeletal ciliopathy with early progressive kidney disease
607386	IFT172	2p23.3	BBS; RP; NPHP; SRTD; Jeune syndrome
615870	IFT27	22q12.3	BBS
614068	IFT43	14q24.3	CED; SRTD
617094	IFT52	20q13.12	SRTD
606621	IFT57	3q13.12-q13.13	OFD with short stature and brachymesophalangia
608040	IFT74	9p21.2	BBS
611177	IFT80	3q25.33	SRTD
605489	IFT81	12q24.11	NPHP; polydactyly; SRTD
613037	INPP5E	9q34.3	JBTS; MORM syndrome
610621	INTU	4q28.1	OFD; SRTD
243305	INVS	9q31.1	NPHP; SLSN; situs inversus
609237	IQCB1	3q13.33	LCA; SLSN; NPHP
616650	KIAA0556	16p12.1	JBTS
610178	KIAA0586	14q23.1	JBTS; hydrolethalus; SRTD
617112	KIAA0753	17p13.1	JBTS; OFD
617612	KIAA1868 (ARMC9)	2q37.1	JBTS
611254	KIF7	15q26.1	Hydrolethalus; scrocallosal syndromes; JBTS; Al-Gazali syndrome
615757	KIZ	20p11.23	CRD
611408	LCA5	6q14.1	LCA
606568	LZTFL1	3p21.31	BBS
154235	MAK	6p24.2	RP
604896	MKKS	20p12.2	MKKS; BBS
609883	MKS1	17q22	BBS, MKS; JBTS
616786	MAPKBP1	15q15.1	NRHP
604588	NEK1	4q33	SRTD; OFD
604043	NEK2	1q32.3	RP
609799	NEK8	17q11.2	NPHP; renal-hepatic-pancreatic dysplasia
609798	NEK9	14q24.3	SRTD; lethal congenital contracture syndrome
607100	NPHP1	2q13	NPHP; JBTS; SLSN; BBS
608002	NPHP3	3q22.1	NPHP; MKS; renal-hepatic-pancreatic dysplasia
607215	NPHP4	1p36.31	NPHP; SLSN
300170	OFD1	Xp22.2	RP; JBTS; OFD; Simpson-Golabi-Behmel syndrome
602676	PDE6D	2q37.1	JBTS
607532	PIBF1	13q21.3-q22.1	JBTS
602610	PIK3R4	3q22.1	RP with NPHP and developmental anomalies
601313	PKD1	16p13.3	ADPKD
173910	PKD2	4q22.1	ADPKD
606702	PKHD1	6p12.3-p12.2	ARPKD
614784	POC1B	12q21.33	CRD; LCA; JBTS
606822	POMGNT1	1p34.1	Non-syndromic RP; muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies)
601309	PTCH1	9q22.32	Holoprosencephaly; basal cell nevus syndrome

Table SI. Continued.

OMIM ID	Gene name	Cytogenetic location	Disease association
607968	PTHB1	7p14.3	BBS
606144	RAB23	6p12.1-p11.2	Carpenter syndrome; acrocephalopolysyndactyly type 2
612994	RAB28	4p15.33	CRD
603937	RP1	8q11.2-q12.1	RP
608581	RP1L1	8p23.1	RP; occult macular dystrophy
300757	RP2	Xp11.3	RP
312610	RPGR	Xp11.4	CRD; RP; RP with PCD; sinorespiratory infections, with or without deafness
605446	RPGRIP1	14q11.2	CRD; LCA
610937	RPGRIP1L	16q12.2	MKS; JBTS; COACH syndrome
611399	SCLT1	4q28.2	OFD
613524	SDCCAG8	1q43-q44	BBS; SLSN; NPHP
601500	SMO	7q32.1	Curry-Jones syndrome; basal cell carcinoma; medulloblastoma
609868	SPATA7	14q31.3	LCA; RP
607035	SUFU	10q24.32	JBTS; basal cell carcinoma; medulloblastoma; meningioma
612758	TAPT1	4p15.32	Complex lethal osteochondrodysplasia
615867	TBC1D32	6q22.31	OFD; polydactyly, absent pituitary and congenital heart disease
617353	TCTEX1D2	3q29	SRTD
609863	TCTN1	12q24.11	JBTS, MKS, OFD
613846	TCTN2	12q24.31	MKS; NPHP; JBTS
613847	TCTN3	10q24.1	Mohr syndrome; JBTS; OFD
616183	TMEM107	17p13.1	JBTS; OFD; MKS
614459	TMEM138	11q12.2	JBTS; OFD
614950	TMEM17	2p15	OFD
613277	TMEM216	11q12.2	MKS; JBTS; OFD
614949	TMEM231	16q23.1	OFD; MKS; JBTS
614423	TMEM237	2q33.1	JBTS; MKS
609884	TMEM67	8q22.1	MKS; JBTS; NPHP; COACH; modifier of BBS
609507	TOPORS	9p21.1	RP
607380	TRAF3IP1	2q37.3	SLSN
602290	TRIM32	9q33.1	BBS; limb-girdle muscular dystrophy type 2H
611695	TTBK2	15q15.2	Spinocerebellar ataxia
612014	TTC21B	2q24.3	NPHP; SRTD; focal segmental glomerulosclerosis (FSGS)
608132	TTC8	14q31.3	BBS; RP
612268	TTLL5	14q24.3	CRD
601197	TUB	11p15.4	Retinal dystrophy and obesity
602280	TULP1	6p21.31	RP, LCA
604011	UNC119	17q11.2	CRD
300072	USP9X	Xp11.4	A novel syndrome with multiple congenital malformations and developmental delay
613580	WDPCP	2p15	BBS; OFD; heart defect-tongue hamartoma-polysyndactyly syndrome
608151	WDR19	4p14	CED; SLSN; RP; SRTD; NPHP
613363	WDR34	9q34.11	SRTD
613602	WDR35	2p24.1	CED; EVC; SRTD
615462	WDR60	7q36.3	SRTD; JATD
613553	XPNPEP3	22q13.2	NPHP-like nephropathy
603073	ZIC2	13q32.3	Holoprosencephaly
604557	ZNF423	16q12.1	NPHP; JBTS

ALMS, Alström syndrome; ADPKD, autosomal dominant polycystic kidney disease; ARPKD, autosomal recessive polycystic kidney disease; BBS, Bardet Biedl Syndrome; CED, cranioectodermal dysplasia; CRD, con-rode dystrophy; EVC, Ellis-van Creveld syndrome; JATD, Jeune asphyxiating thoracic dystrophy; JBTS, Joubert Syndrome; LCA, Leber congenital amaurosis; MKS, Meckel-Gruber syndrome; NPHP, nephronophthisis; OFD, orofaciodigital syndromes; RP, retinitis pigmentosa; SLSN, Senior Loken syndrome; SRTD, short-rib thoracic dysplasia.