

資料2 ジュベール症候群、ジュベール症候群関連疾患の原因遺伝子

略名	染色体	病型	遺伝子名
INPP5E	9q34.3.	JBTS1	inositol polyphosphate-5-phosphatase
TMEM216	11q13	JBTS2	TMEM216 transmembrane protein 216
AHI1	6q23	JBTS3	Abelson helper integration site 1
NPHP1	2q13	JBTS4	nephronophthisis 1 (juvenile)
CEP290	12q21.32	JBTS5	centrosomal protein 290kDa
TMEM67	8q21	JBTS6/COACH	transmembrane protein 67
RPGRIP1L	16q12.2	JBTS7/COACH	RPGRIP1-like
ARL13B	3q11.2	JBTS8	ADP-ribosylation factor-like 13B
CC2D2A	4p15.3	JBTS9/COACH	coiled-coil and C2 domain containing 2A
CXORF5(OFD1)	Xp22.3	JBTS10	oral-facial-digital syndrome 1
TTC21B	2q24	JBTS11	tetratricopeptide repeat domain 21B
KIF7	15q26	JBTS12	kinesin family member 7
TCTN1	12q24	JBTS13	tectonic family member 1
TMEM237	2q33	JBTS14	transmembrane protein 237
CEP41	7q32	JBTS15	centrosomal protein 41kDa
TMEM138	11q	JBTS16	transmembrane protein 138
C5ORF42	5p13	JBTS17	chromosome 5 open reading frame 42
TCTN3	10q24	JBTS18	tectonic family member 3
ZNF423	16	JBTS19	zinc finger protein 423
TMEM231	16q23.1	JBTS20?	transmembrane protein 231
EXOC8	1q42.2	JBTS21?	exocyst complex component 8
NPHP4	1p36.31	SLS4	nephronophthisis 4
IQCB1	3q21	SLS5	IQ motif containing B1
CEP290	12q21	SLS6	Centrosomal protein 290kDa
SDCCAG8	1q44	SLS7	serologically defined colon cancer antigen 8

JBTS: ジュベール (Joubert) 症候群、COACH: COACH症候群、SLS: セニール・ローケン (Senior-Loken) 症候群。