Supplementary Table 2. Imaging and genetics of patients with Joubert syndrome

	Patient									Total (*n* [%])
	1	2	3	4	5	6	7	8	9	
MRI brain										
MTS	+	+	+	+	+	+	+	+	+	9 (100)
Deep IF	+	+	+	+	+	+	+	+	+	9 (100)
Vermis hypoplasia	-	+	+	+	+	+	+	+	+	8 (88.9)
SCP thickening	+	+	+	+	+	+	+	+	+	9 (100)
Batwing appearance	+	+	+	+	+	+	+	+	+	9 (100)
Orientation of SCP (type)	C	A	P	C	P	C	P	P	P	A: 1 (11.1) C: 3 (33.3) P: 5 (55.6)
Retro cerebellar collection	-	-	-	-	-	+	+	-	+	3 (33.3)
Callosal atrophy	+	+	-	-	-	-	-	-	-	2 (22.2)
Pachygyria	-	-	-	-	-	-	+	-	-	1 (11.1)
DTI—Lack of decussation of SCP	NA	NA	+	+	NA	NA	+	NA	+	4 (100)
DTI—Horizontal SCP	NA	NA	+	+	NA	NA	-	NA	+	3 (75)
2D-ECHO	N	N	N	N	N	N	N	NA	N	x
USG—Abdomen	N	N	N	N	N	N	N	NA	N	x
Genetic abnormality										
Variant detail	NA	NA	*CPLANE1* p.R2467Kfs*7; p.W24R	NA	*MKS1* p.S297del	*MKS1* p.S297del	NA	NA	*PIBF1* p.N658Kfs*15; p.R405Q	x
Zygosity	NA	NA	Compound heterozygous	NA	Homozygous	Homozygous	NA	NA	Compound heterozygous	x
Variant classification according to ACMG criteria	NA	NA	PVS1, PM2, PP3—Pathogenic/PM2, PM3, PP3—Variant of unknown significance	NA	PM2, PM4, PP1, PP3—Likely pathogenic	PM2, PM4, PP1, PP3—Likely pathogenic	NA	NA	PVS1, PM2, PP3-Pathogenic/PM1, PM3, PP3, PP5—Likely pathogenic	x

ACMG, American College of Medical Genetics; A, superior cerebellar peduncles in axial image oriented like letter “A”; C, superior cerebellar peduncles in axial image curved and oriented like letter “C”; DTI, diffusion tensor imaging; ECHO, echocardiography; IF, interpeduncular fossa; MRI, magnetic resonance imaging; MTS, molar tooth sign; N, normal; NA, not available; P, superior cerebellar peduncles in axial image oriented “Parallelly”; SCP, superior cerebellar peduncle; USG, ultrasonography; +, present; -, absent; x, not applicable.