

**Supplementary Table 1.** Clinical features of patients with Joubert syndrome

	Patient									Total
	1	2	3	4	5	6	7	8	9	
Age (years)	2.5	0.8	15	13	24	13	7	5	12.5	10.3 ± 6.82 (10 m–24 y)
Gender	F	M	M	F	F	M	M	F	F	4:5
Family history	-	-	-	-	+	+	-	-	-	2 (22.2)
Consanguinity	+	+	-	-	-	-	-	-	-	2 (22.2)
Developmental delay	+	+	+	+	+	+	+	+	+	9 (100)
Apnoea	+	+	+	-	-	+	+	+	+	7 (77.8)
Facial dysmorphism	+	+	+	+	+	+	+	+	+	9 (100)
Trapezoid shaped mouth	+	-	-	+	-	-	+	+	-	4 (44.4)
Everted lower lip	-	-	-	+	-	-	+	+	-	3 (33.3)
Broad nasal bridge	+	+	+	+	-	-	-	-	+	5 (55.5)
Low set ears	-	+	+	+	+	+	-	-	-	5 (55.5)
Frontal bossing	-	+	-	-	-	-	+	+	-	3 (33.3)
Dolicocephaly	-	-	-	-	-	-	+	-	-	1 (11.1)
Arachnodactyly	-	-	-	-	+	+	-	-	-	2 (22.2)
Kyphosis	-	-	-	-	+	+	-	-	-	2 (22.2)
Speech abnormalities*	Not attained	Not attained	Scanning	Scanning	Hypo-kinetic	Mixed	Anarthria	Anarthria	Normal	6 (85.7)
Eye abnormalities	+	+	+	+	+	+	+	+	+	9 (100)
Nystagmus	Vertical	-	-	Horizontal	Torsional	Torsional	-	-	-	4 (44.4)
Saccadic abnormalities	-	-	+	+	+	-	-	-	-	3 (33.3)
Exotropia	-	-	-	-	+	+	-	+	-	3 (33.33)
Esotropia	-	+	-	-	-	-	-	-	-	1 (11.1)
COMA	-	-	+	+	+	-	+	-	-	4 (44.4)
OTR	+	+	+	+	+	+	-	-	-	6 (66.7)
ASD	+	+	+	+	+	-	-	-	-	6 (66.7)
Iris coloboma	-	-	-	-	-	+	-	-	-	1 (11.1)
Fundus abnormality	+	-	-	-	-	-	-	-	-	1 (11.1)
Head tilt	-	-	+	+	+	+	+	-	-	5 (55.5)
Hypotonia	+	+	+	+	+	-	+	+	-	7 (77.8)
Spasticity	-	-	-	-	-	+	-	-	-	1 (11.1)
Dystonia	-	-	-	+	+	+	-	-	+	4 (44.4)
Myoclonus	-	-	+	-	-	-	-	-	-	1 (11.1)
Limb ataxia	+	NA	+	+	+	+	+	+	+	8 (100)
Ataxia*	Not attained	Not attained	+	+	+	+	Not attained	Not attained	+	5 (100)

Values are presented as *n* (%) unless otherwise indicated. \*two patients did not attain language milestones and four patients did not attain walking milestones. ASD, alternate skew deviation; COMA, congenital oculomotor apraxia; F, female; M, male; m, months; NA, not available; OTR, ocular tilt reaction; y, years; +, present; -, absent.