

Joubert Syndrome Clinical Features Checklist

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THIS FORM MUST BE COMPLETED IN ORDER FOR TESTING TO BEGIN

Patient name:	Patient ID Code:
Contact person/relationship:	
Email and phone:	
Patient Sex (M/F):	Patient DOB:
Consanguinity (Y/N):	Ethnic background:
Name of person completing this form:	

CLINICAL FEATURE	Documented present	Documented absent	Unknown	Not applicable/ not done	Comments
CNS:					
Molar tooth sign (MRI scan)					
Cerebellar vermis hypoplasia (CT scan)					
Dandy-Walker malformation/cyst					
Encephalocele					
Agenesis of corpus callosum					
Polymicrogyria					
Heterotopias					
Hypotonia					
Ataxia					
Developmental delay					
Speech apraxia					
Breathing abnormality-apnea					
Breathing abnormality-tachypnea					
Seizure disorder					
Lab studies:					
Karyotype (normal/abnormal)					
Abnormal gene test/where tested?					
Abnormal LFT's					
Eyes:					
Abnormal eye movements					
Nystagmus					
Oculomotor apraxia					
Colobomas					
Ptosis					
Strabismus/amblyopia (eyes malaligned)					

Retinal dystrophy					
Severe visual impairment (blindness from birth)					
Abdomen:					
Cystic dysplastic kidney(s)					
Nephronophthisis					
Kidney failure					
Hepatic fibrosis/cirrhosis					
Situs inversus/dextrocardia					
Other:					
Anosmia (no sense of smell)					
Pituitary dysfunction					
Micropenis (males)					
Congenital heart defect					
Obstructive sleep apnea					
Polydactyly-postaxial (pinkie side)					
Polydactyly-preaxial (thumb side)					
Polydactyly-mesaxial (middle fingers)					
Tongue tumors/oral frenulae					
Abnormal prenatal imaging					
Abnormal fetal MRI					