

## Joubert syndrome and Related Disorders – clinical questionnaire

### General data and clinical history

name and surname: \_\_\_\_\_

date of birth: \_\_\_\_\_

gender: M  F

pregnancy (relevant events): \_\_\_\_\_

delivery (relevant events): \_\_\_\_\_

development steps (age): sitting: \_\_\_\_\_ walking: \_\_\_\_\_

first words: \_\_\_\_\_ complete language: \_\_\_\_\_

scholarship: normal  aided  impossible

### Referral source/physician:

### Family history

Father: name: \_\_\_\_\_ place of birth/ethnicity: \_\_\_\_\_

Mother: name: \_\_\_\_\_ place of birth/ethnicity: \_\_\_\_\_

consanguineous parents: yes  no  possibly

affected sibs: yes  no  if yes, how many: \_\_\_\_\_ age: \_\_\_\_\_

healthy sibs yes  no  if yes, how many: \_\_\_\_\_ age: \_\_\_\_\_

other affected family members: yes  no  if yes, please attach pedigree

family history of other genetic disorders: \_\_\_\_\_

blood for genetic study: yes  no

*if yes, please specify on the pedigree which family members have been blood-sampled. Please note any intra-familial phenotypic differences or fill out additional questionnaires for each affected family member*

### Central Nervous System

breathing abnormalities: yes  no

*if yes, please describe:* \_\_\_\_\_

ataxia: yes  no

hypotonia: yes  no

tremor: yes  no

mental retardation: yes  no

oculomotor apraxia: yes  no

nystagmus: yes  no

other (please describe): \_\_\_\_\_

\_\_\_\_\_

\_\_\_\_\_

**Kidney**

polyuria: yes  no   
 enuresis: yes  no   
 renal ultrasound: date \_\_\_\_\_ normal  abnormal  not performed   
*if abnormal, please describe (cysts, cortico-medullar differentiation, kidney longitudinal diameter, hyperintensity, nephrocalcinosis etc):* \_\_\_\_\_

nephronophthisis: yes  no   
 renal failure: yes  no  if yes, age: \_\_\_\_\_  
 serum creatinine: \_\_\_\_\_ date: \_\_\_\_\_ not evaluated   
 urinary specific gravity: \_\_\_\_\_ date: \_\_\_\_\_ not evaluated   
 U Osm max (DDAVP concentration test): \_\_\_\_\_ date: \_\_\_\_\_ not performed   
 other (please describe): \_\_\_\_\_

**Eyes**

roving eye movements: yes  no   
 fundus oculi: date \_\_\_\_\_ normal  abnormal  not evaluated   
*if abnormal, please describe (retinal abnormalities, colobomas etc):* \_\_\_\_\_

visus reduction: yes  no   
*if yes, please specify RE: \_\_\_\_/10 LE: \_\_\_\_/10*  
 visual evoked potentials (VEP): date \_\_\_\_\_ normal  abnormal  absent  not performed   
*if abnormal, specify: amplitude: \_\_\_\_\_ latency: \_\_\_\_\_*  
 electroretinogram (ERG): date \_\_\_\_\_ normal  abnormal  absent  not performed   
*if abnormal, please describe:* \_\_\_\_\_

other (please describe): \_\_\_\_\_

**Liver**

serum liver enzymes: date \_\_\_\_\_ normal  abnormal  not evaluated   
*if abnormal, please specify:* \_\_\_\_\_  
 liver ultrasound: date \_\_\_\_\_ normal  abnormal  not performed   
*if abnormal, please describe (cysts etc):* \_\_\_\_\_

hepatic fibrosis: yes  no   
 other (please describe): \_\_\_\_\_

**Other features**

polydactyly: absent  preaxial  postaxial

tongue amartomas/tumors: yes  no

multiple lingual frenula: yes  no

cleft lip: yes  no

cleft palate: yes  no

other tumors: yes  no

pituitary abnormality: yes  no

seizures: yes  no

diabetes: yes  no

heart problems: yes  no

facial dysmorphic features: yes  no

*if yes to any of the above, please describe:* \_\_\_\_\_

other (please describe): \_\_\_\_\_

**Behavioral Heath**

bipolar disorder yes  no

autistic-like behavior (for children over the developmental age of two only)

## 1. social interaction:

impaired non-verbal communication (eye to eye gaze, facial expression, gestures) yes  no

failure to form developmentally appropriate peer relationships yes  no

lack of spontaneous interest in sharing with others (showing, bringing, pointing) yes  no

lack of emotional reciprocity yes  no

## 2. communication:

spoken language delay (in the absence attempts to use gestures or mime to communicate) yes  no

inability to initiate or sustain a conversation (assuming adequate speech) yes  no

stereotyped/repetitive use of language yes  no

lack of developmentally appropriate spontaneous, varied make-believe or imitative play yes  no

## 3. repetitive, stereotyped behavior/interests:

preoccupation with stereotyped/restricted interests, abnormal in intensity or focus yes  no

inflexible adherence to routines yes  no

stereotyped/repetitive motor actions (i.e. hand twisting) yes  no

preoccupation with objects yes  no

**Brain MRI**

date \_\_\_\_\_

not performed

vermis aplasia:

no

partial

total

*if partial, please describe:* \_\_\_\_\_

“molar tooth” sign:

yes

no

encephalo-meningocele

yes

no

hydrocephalus

yes

no

Dandy-Walker malformation:

yes

no

corpus callosum hypoplasia:

yes

no

other malformations posterior cranial fossa:

yes

no

*if yes, please describe:* \_\_\_\_\_

other abnormalities (please describe): \_\_\_\_\_

\_\_\_\_\_

originals/copy available:                      yes                       no                       if yes from whom? \_\_\_\_\_

**Patient’s measurements (if appropriate)**

\_\_\_\_\_

**Other exams**

\_\_\_\_\_  
\_\_\_\_\_  
\_\_\_\_\_  
\_\_\_\_\_

**Comments and notes**

\_\_\_\_\_  
\_\_\_\_\_  
\_\_\_\_\_  
\_\_\_\_\_

\_\_\_\_\_  
date

\_\_\_\_\_  
physician’s signature