CLINICAL CHECKLIST - KABUKI SYNDROME

Patient:

First name : Referring physician:

Last name : Referring Center:

Date of Birth: Sex: M ☐ F ☐

Pregnancy – Birth – Neonatal period information:

	Yes	No	NE	
Pregnancy :				
Cystic hygroma:	☐	☐	☐	
Two-vessel cord:	☐	☐	☐	
Birth :				
Premature	☐	☐	☐ (	weeks)
Weight < P3	☐	☐	☐ (	kg)
Length < P3	☐	☐	☐ (	cm)
Head circumference < P3	☐	☐	☐ (	cm)
Neonatal period :				
Hypoglycemia	☐	☐	☐	

CRANIUM

Head circumference:

cm at age y m

	Yes	No	NE
Microcephaly	☐	☐	☐
Craniosynostosis	☐	☐	☐
Malar hypoplasia	☐	☐	☐
Eyebrows:			
Arched	☐	☐	☐
Lateral sparseness	☐	☐	☐
Notched	☐	☐	☐
Orbital region:			
Long palpebral fissures	☐	☐	☐
Long eyelashes	☐	☐	☐
Everted lower lids	☐	☐	☐
Upper lids: heavy, thick	☐	☐	☐
Ptosis	☐	☐	☐
Epicanthic folds	☐	☐	☐
Lagophtalmos *(sleep with eyes opened)*	☐	☐	☐
Eyes:			
Blue sclera	☐	☐	☐
Coloboma	☐	☐	☐
Congenital glaucoma	☐	☐	☐
Optic nerve hypoplasia	☐	☐	☐
Strabismus	☐	☐	☐
Astigmatism	☐	☐	☐
Nystagmus	☐	☐	☐
Nose:			
Broad tip	☐	☐	☐
Depressed tip	☐	☐	☐
Short columella	☐	☐	☐

	Yes	No	NE
Lips:			
Broad, prominent philtrum	☐	☐	☐
Tented upper lip	☐	☐	☐
Cleft upper lip	☐	☐	☐
Prominent, droopy lower lip	☐	☐	☐
Pits	☐	☐	☐
Palate:			
Cleft	☐	☐	☐
High arched	☐	☐	☐
Narrow	☐	☐	☐
Velopharyngeal dysfunction	☐	☐	☐
Teeth, tongue:			
Neonatal teeth	☐	☐	☐
Malocclusion	☐	☐	☐
Hypodontia	☐	☐	☐
Abnormal tooth shape	☐	☐	☐
Fissured tongue	☐	☐	☐
Ears:			
Prominent	☐	☐	☐
Cupped	☐	☐	☐
Simple in form	☐	☐	☐
Large auricle	☐	☐	☐
Large lobe	☐	☐	☐
Dimple, pit, fistula	☐	☐	☐
Recurrent otitis	☐	☐	☐
Hearing loss	☐	☐	☐
Inner ear abnormalities	☐	☐	☐

	Yes	No	NE
GROWTH AND FEEDING			
Feeding difficulties	☐	☐	☐
Reflux	☐	☐	☐
Aspiration pneumonia	☐	☐	☐
Failure to thrive	☐	☐	☐
Gastrostomy	☐	☐	☐
Start:			
Stop:			
Short stature (postnatal)	☐	☐	☐
Height: cm at age			
Delayed bone age	☐	☐	☐
Growth hormone deficiency	☐	☐	☐
Obesity	☐	☐	☐
Weight: kg at age			
CARDIOVASCULAR			
Malformation	☐	☐	☐
ASD	☐	☐	☐
VSD	☐	☐	☐
Left sided obstruction	☐	☐	☐
Other:			
GASTROINTESTINAL			
Anal malformation	☐	☐	☐
Intestinal malrotation	☐	☐	☐
Chronic diarrhea	☐	☐	☐
Constipation	☐	☐	☐
GENITOURINARY			
Kidney			
Renal dysplasia/agenesis	☐	☐	☐
Horseshoe/ ectopic kidney	☐	☐	☐
Hydronephrosis/ vesico-ureteric reflux	☐	☐	☐
Genital			
Hypospadias	☐	☐	☐
Cryptorchidism	☐	☐	☐
Micropenis	☐	☐	☐
Labia hypoplasia	☐	☐	☐
PUBERTY			
Areolar fullness in infancy	☐	☐	☐
Age of first menstruation:		years	
Gonadotropin abnormality	☐	☐	☐

	Yes	No	NE
MUSCULOSKELETAL AND SKIN			
Persistent finger pads	☐	☐	☐
Single palmar crease	☐	☐	☐
Brachydactyly	☐	☐	☐
Clinodactyly of 5th finger	☐	☐	☐
Small feet	☐	☐	☐
Long hallux	☐	☐	☐
Vertebral anomaly	☐	☐	☐
Scoliosis	☐	☐	☐
Sacral sinus or dimple	☐	☐	☐
Clavicle hypo/aplasia	☐	☐	☐
Hip dysplasia	☐	☐	☐
Patellar dislocation	☐	☐	☐
Chest deformities	☐	☐	☐
Hyperlaxity	☐	☐	☐
Beighton score /9			
Small/hypoplastic/ deep set nails	☐	☐	☐
Hirsutism	☐	☐	☐
Pilomatrixoma	☐	☐	☐
IMMUNITY			
Autoimmune condition	☐	☐	☐
Recurrent infection	☐	☐	☐
Comment:			
Hypogammaglobulinemia	☐	☐	☐
CANCER	☐	☐	☐
Comment:			
DEVELOPMENT AND BEHAVIOUR			
Development delay: mild☐ moderate☐ severe☐			
IQ:			
Autism	☐	☐	☐
Hypotonia	☐	☐	☐
Seizure	☐	☐	☐
Expressionless face	☐	☐	☐
Dysarthria	☐	☐	☐
Behavior trouble	☐	☐	☐
Comment:			