UKGTN Testing Criteria

Test name: Bardet-Biedl Syndrome 20 Gene Panel	
Approved name and symbol of disorder/condition(s): See website listing	**OMIM number(s):**
Approved name and symbol of gene(s): See website listing	**OMIM number(s):**

Patient name:	**Date of birth:**
Patient postcode:	**NHS number:**
Name of referrer:	
Title/Position:	**Lab ID:**

Referrals will only be accepted from one of the following:	
Referrer	**Tick if this refers to you.**
Consultant Clinical Geneticist	
Consultant Ophthalmologist	
Consultant Nephrologist	

Minimum criteria required for testing to be appropriate as stated in the Gene Dossier:	
Criteria	**Tick if this patient meets criteria**
Four primary features or three primary features and two secondary features	
Primary features:	
- Rod-cone dystrophy	
- Renal abnormalities	
- Obesity	
- Polydactyly	
- Learning difficulties	
- Hypogonadism in males	
Secondary features:	
- Speech disorder/delay	
- Strabismus/cataracts/astigmatism	
- Brachydactyly/syndactyly	
- Developmental delay	
- Polyuria/polydipsia	
- Ataxia/poor coordination/imbalance	
- Mild spasticity (especially lower limbs)	
- Diabetes mellitus	
- Dental crowding/hypodontia/small roots/high arched palate	
- Left ventricularhypertrophy/congenital heart disease	
- Hepatic fibrosis	

continued/…

Additional Information:

All referrals are via specialist NCG multidisciplinary clinics held at 4 Centres.

For panel tests: At risk family members where familial mutation is known do not require a full panel test but should be offered analysis of the known mutation.

If the sample does not fulfil the clinical criteria or you are not one of the specified types of referrer and you still feel that testing should be performed please contact the laboratory to discuss testing of the sample.