UKGTN Testing Criteria

Test name: Skeletal Ciliopathies 23 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 nephrologist	
Consultant in metabolic diseases	
Consultant in retinal diseases	

Minimum criteria required for testing to be appropriate as stated in the Gene Dossier:	
Criteria	**Tick if this patient meets criteria**
At least two of the following:	
- Renal cysts	
- Laterality defect	
- Polydactyly	
- Retinal degeneration	
- Posterior fossa defects/encephalocoele	
- Narrow thorax	

Additional Information:

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.