UKGTN Testing Criteria

Test name: Microphthalmia, Anophthalmia and Coloboma (MAC) Spectrum and Aniridia 40 Gene Exome Panel	
Approved name and symbol of disorder/condition(s): *For panel tests: See website listing*	**OMIM number(s):**
Approved name and symbol of gene(s): *For panel tests: See website listing*	**OMIM number(s):**

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

Minimum criteria required for testing to be appropriate as stated in the Gene Dossier:	
Criteria	**Tick if this patient meets criteria**
Normal Karyotype or array CGH **AND** one or more of the following eye phenotype as a *prominent feature* of the clinical presentation:	
Anophthalmia **(please specify-Left or Right eye or Both)**	
Microphthalmia **(please specify-Left or Right eye or Both)**	
Coloboma **(please specify-Left or Right eye or Both)**	
Aniridia (typical of Classical Aniridia MIM #106210) **(please specify-Left or Right eye or Both)**	
Iris hypoplasia (typical of Gillespie syndrome MIM #206700) **(please specify-Left or Right eye or Both)**	

Additional Information:
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.