UKGTN Testing Criteria

Test name: **3-Beta-Hydroxysteroid Dehydrogenase, Type II, Deficiency Of**	
Approved name and symbol of disorder/condition(s): 3-Beta-Hydroxysteroid Dehydrogenase, Type II, Deficiency of	**OMIM number(s):** 201810
Approved name and symbol of gene(s): hydroxy-delta-5-steroid dehydrogenase, 3 beta- and steroid delta-isomerase 2; HSD3B2	**OMIM number(s):** 613890

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 Biochemist/Chemical Pathologist	
Consultant Paediatric or adult Endocrinologist	

Minimum criteria required for testing to be appropriate as stated in the Gene Dossier:	
Criteria	**Tick if this patient meets criteria**
Ambiguous genitalia **AND** at least one of the following: • Urine steroid profile suggestive of 3βHSD • Renal salt loss • abnormal ratio of DHA/androstenedione • failure of testosterone to increase on hCG test	
OR At risk family members where familial mutation is known.	

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.