

UKGTN Testing criteria

Name of Disease(s): 17-@BETA HYDROXYSTEROID DEHYDROGENASE III DEFICIENCY (264300)

Name of gene(s): Hydroxysteroid (17-beta) dehydrogenase 3; HSD17B3 (605573)

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	
Paediatric/Adult Endocrinologist	
Adolescent Gynaecologist	

Minimum criteria required for testing to be appropriate as stated in the Gene Dossier:

Criteria	Tick if this patient meets criteria
Ratio of testosterone to androstenedione < 0.8 AND	
Inguinal swellings in apparent newborn female AND	
Some newborn clitoromegaly OR	
Virilisation of female at puberty	

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.