UKGTN Testing Criteria

Approved name and symbol of disease/condition(s): 46,XY Sex Reversal 3; SRXY3	**OMIM number(s):** 612965
Approved name and symbol of gene(s): nuclear receptor subfamily 5, group A, member 1 NR5A1	**OMIM number(s):** 184757

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.**
Clinical geneticist	
Endocrinologist (paediatric/adult)	

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

Criteria	**Tick if this patient meets criteria**
46,XY karyotype **AND**	
evidence of gonadal dysgenesis (uterus, ↑FSH, low testosterone, low AMH, gonadal histology if available).	
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