UKGTN Testing Criteria

Approved name and symbol of disease/condition(s):	**OMIM number(s):**
Aarskog-Scott Syndrome; AAS	305400
Approved name and symbol of gene(s):	**OMIM number(s):**
fyve, rhogef, and ph domain-containing 1; FGD1	300546

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	

Minimum criteria required for testing to be appropriate as stated in the Gene Dossier:	
Criteria	**Tick if this patient meets criteria**
Three or more signs from: • moderate short stature, • distinct craniofacial abnormalities, (including hypertelorism, down-slanting palpebral fissures and short nose with upturned nares), • short and characteristic hands (brachydactyly with inter digital webbing) • shawl scrotum • cryptorchidism • hypermetropia	
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