

UKGTN Testing criteria

Name of Disease(s): ACHONDROPLASIA; ACH (100800)

Name of gene(s): fibroblast growth factor receptor 3; FGFR3 (134934)

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	
Paediatrician	
Obstetrician	
Perinatal pathologist	

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

Criteria	Tick if this patient meets criteria
Rhizomelic limb shortening with onset from the second trimester	
and relative macrocephaly	
and characteristic facial features	
and characteristic radiographic features (if delivered, e.g. trident shaped acetabulum, proximal femoral translucency, failure of interpedicular distances to widen)	

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.