

UKGTN Testing criteria

Name of Disease(s): ACHONDROGENESIS, TYPE II; ACG2 (200610)
Name of gene(s): collagen, type II, alpha 1; COL2A1 (120140)

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	<input type="checkbox"/>
Perinatal Pathologist	<input type="checkbox"/>

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

Criteria	Tick if this patient meets criteria
Characteristic postnatal radiographic features (if delivered): <ul style="list-style-type: none"> • underossified vertebral bodies • unossified pubic and ischial bones • small iliac bones • short tubular bones with metaphyseal cupping lateral bowing of femora • short ribs 	<input type="checkbox"/>

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.