

UKGTN Testing Criteria

Test name: Microcephaly and Microcephalic Dwarfism Spectrum 50 Gene Panel	
Approved name and symbol of disorder/condition(s): <i>See website listing</i>	OMIM number(s):
Approved name and symbol of gene(s): <i>See website listing</i>	OMIM number(s):

Referrals will only be accepted from one of the following:	
Referrer	Tick if this refers to you
Consultant Clinical Geneticist	<input type="checkbox"/>
Consultant Neurologist, Paediatric	<input type="checkbox"/>
Consultant Endocrinologist, Paediatric	<input type="checkbox"/>
Consultant Endocrinologist, Adult	<input type="checkbox"/>

Minimum criteria required for testing to be appropriate as stated in the Gene Dossier:	
Criteria	Tick if this patient meets criteria
Inclusion criteria	<input type="checkbox"/>
<ul style="list-style-type: none"> • Occipitofrontal circumference (OFC) >2 standard deviations (SD) below mean at birth AND progressive microcephaly to >3SD, OR • OFC >3SD below mean at birth, OR • OFC >3SD below mean at birth AND length >3SD below mean at birth 	<input type="checkbox"/>
Exclusion criteria	<input type="checkbox"/>
<ul style="list-style-type: none"> • A known genetic cause • Dysmorphic physical features or MRI brain indicative of an alternative diagnosis (e.g. a neuronal migration disorder) • Evidence of an environmental cause (e.g. infection, teratogens, birth hypoxia) 	<input type="checkbox"/>

Additional Information:

Please see the care pathway appended to the end of this document.

For panel tests:

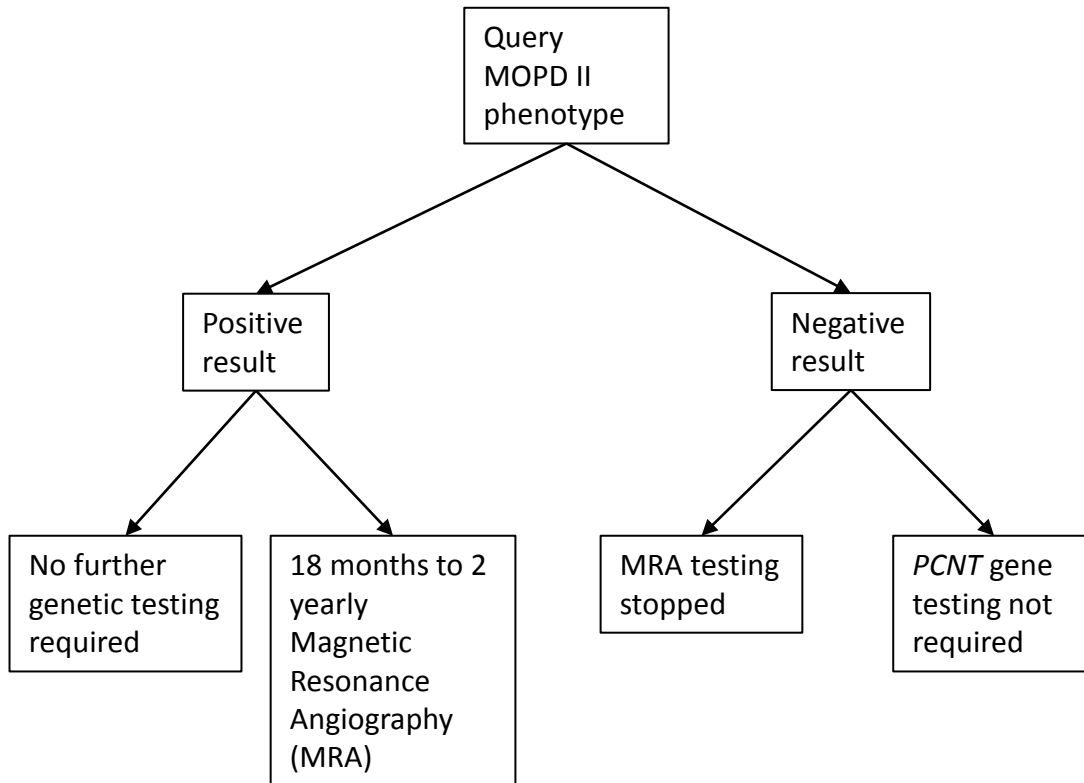
At risk family members where familial mutation is known do not require a full panel test but should be offered analysis of the known mutation.

A proforma is required to be completed for this testing, please access this from the laboratory website at:

http://www.nhslothian.scot.nhs.uk/Services/A-Z/ClinicalGeneticsService/GeneticLaboratoryServices/molecular_genetics/DNA_Tests/Pages/Molecular-genetic-test-information-sheets.aspx.

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.

Gene panel available



Gene panel **not** available

