

UKGTN Testing Criteria

Test name: Acrodysostosis with or without Multiple Hormone Resistance	
Approved name and symbol of disorder/condition(s): Acrodysostosis 2, with or without Hormone Resistance; ACRDYS2	OMIM number(s): 614613
Approved name and symbol of gene(s): phosphodiesterase 4D, cAMP-specific; PDE4D	OMIM number(s): 600129

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

Minimum criteria required for testing to be appropriate as stated in the Gene Dossier:	
Criteria	Tick if this patient meets criteria
Clinical diagnosis of acrodysostosis (short stature, severe brachydactyly, facial dysostosis, and nasal hypoplasia) OR	<input type="checkbox"/>
PRKAR1A negative	<input type="checkbox"/>
At risk family members where familial mutation is known.	<input type="checkbox"/>

Additional Information:

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.