

## UKGTN Testing Criteria

<b>Test name:</b> Acrodysostosis with or without Multiple Hormone Resistance	
<b>Approved name and symbol of disease/condition(s):</b> Acrodysostosis 1, with or without hormone resistance; ACRDYS1	<b>OMIM number(s):</b> 101800
<b>Approved name and symbol of gene(s):</b> protein kinase, cAMP-dependent, regulatory, type I, alpha; PRKAR1A	<b>OMIM number(s):</b> 188830

<b>Patient name:</b>	<b>Date of birth:</b>
<b>Patient postcode:</b>	<b>NHS number:</b>
<b>Name of referrer:</b>	
<b>Title/Position:</b>	<b>Lab ID:</b>

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) <b>AND</b>	<input type="checkbox"/>
Resistance to hormones including PTH and other hormones that bind GPCR's <b>OR</b>	<input type="checkbox"/>
PDE4D 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.