

Inherited Disease Panel

Gene panel Growth Disorders*, version 1, 18-2-2018

HGNC approved gene symbol	Transcript	OMIM gene ID	median depth	% covered >10x	% covered >20x	% covered >30x	% covered >50x
<i>ACAN</i> **	NM_013227	155760	165	100	100	100	100
<i>COMP</i>	NM_000095	600310	95	100	100	99	98
<i>FGFR3</i> **	NM_000142	134934	128	100	100	100	100
<i>GH1</i> **	NM_000515	139250	156	100	100	100	100
<i>GHR</i> **	NM_000163	600946	182	100	100	100	100
<i>GHRH</i> **	NM_001184731	139190	88	100	100	100	100
<i>GHRHR</i> **	NM_000823	139191	106	100	100	100	100
<i>GHSR</i> **	NM_198407	601898	238	100	100	100	100
<i>IGF1</i> **	NM_000618	147440	107	100	100	100	100
<i>IGF1R</i> **	NM_000875	147370	142	100	100	100	100
<i>IGFALS</i> **	NM_004970	601489	***	100	100	100	100
<i>IGFBP3</i> **	NM_001013398	146732	***	100	100	100	100
<i>NPR2</i> **	NM_003995	108961	195	100	100	100	100
<i>PAPSS2</i> **	NM_001015880	603005	133	100	100	100	100
<i>SHOX</i> **	NM_000451	312865	***	100	100	100	100
<i>STAT5B</i> **	NM_012448	604260	145	100	100	100	98

* See also exome sequencing for the larger panel

** Core genes (if necessary additional Sanger sequencing will be performed for 100% coverage)

*** For these genes Sanger sequencing is performed

MLPA for *SHOX*, *GH1*, *LHX4*, *POU1F1*, *HESX1*, *PROP1*, *GHRHR*, *LHX3*, *GHR*, *JAK2*, *IGF1*, *STAT5B*, *IGFR1*, *IGFBP3* and *IGFALS* is included in the gene panel Growth Disorders

