Inherited Disease Panel
Gene panel Growth Disorders, version 1.1, 11-09-2019

Leids Universitair Medisch Centrum

HGNC approved gene symbol	Transcript	OMIM gene ID	median depth	% covered >10x	% covered >20x	% covered >30x	% covered >50x
*ACAN***	NM_001135	155760	186	90	90	90	89
COMP	NM_000095	600310	95	100	100	99	97
*FGFR3***	NM_000142	134934	128	100	99	99	99
*GH1***	NM_000515	139250	156	100	100	100	100
*GHR***	NM_000163	600946	182	100	100	100	100
*GHRH***	NM_001184731	139190	88	100	99	91	90
*GHRHR***	NM_000823	139191	106	100	100	100	100
*GHSR***	NM_198407	601898	238	100	100	100	100
*IGF1***	NM_000618	147440	107	100	100	100	100
*IGF1R***	NM_000875	147370	142	100	100	100	99
*IGFALS****	NM_004970	601489	Sanger sequencing is performed				
*IGFBP3****	NM_001013398	146732	Sanger sequencing is performed				
*NPR2***	NM_003995	607072	195	100	100	100	100
*PAPSS2***	NM_001015880	603005	133	100	100	100	100
*SHOX****	NM_000451	312865	Sanger sequencing is performed				
*STAT5B***	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* is included in the gene panel Growth Disorders