Inherited Disease Panel
Gene panel Growth Disorders, version 2, 22-03-2022

Leids Universitair Medisch Centrum

HGNC approved gene symbol	Transcript	OMIM gene ID	median depth	% covered >10x	% covered >20x	% covered >30x	% covered >50x
*ACAN***	NM_013227	155760	291	87	87	86	86
COMP	NM_000095	600310	140	100	100	100	98
*FGFR3***	NM_000142	134934	216	100	100	99	97
*GH1***	NM_000515	139250	258	100	100	100	100
*GHR***	NM_000163	600946	319	100	100	100	100
*GHRH***	NM_021081	139190	90	100	100	98	98
*GHRHR***	NM_000823	139191	140	100	100	100	100
*GHSR***	NM_198407	601898	391	100	100	100	100
HMGA2	NM_003483	600698	174	100	94	79	66
*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				
IHH	NM_002181	600726	261	100	100	100	100
*NPR2***	NM_003995	607072	284	100	100	100	100
*PAPSS2***	NM_001015880	603005	170	100	100	100	100
PTPN11	NM_002834	176876	195	100	100	99	99
*SHOX***	NM_000451	312865	Sanger sequencing is performed				
*STAT5B***	NM_012448	604260	141	100	100	100	99

* See also exome sequencing for the larger panel
** Core genes (if necessary additional Sanger sequencing will be performed for 100% coverage)
For some genes Sanger sequencing is performed
MLPA for SHOX is included in the gene panel Growth Disorders