Appendix

Composition of the short stature panel v4

Gene	Ref transcript	Avg. % ≥ 20x	Gene	Ref transcript	Avg. % ≥ 20x	Gene	Ref transcript	Avg. % ≥ 20x
ACAN	NM_013227	98,9	FGFR3	NM_000142	99	ORC1	NM_004153	94,7
ANKRD11	NM_001256182	93,1	FLNA	NM_001456	99,4	ORC4	NM_001190879	80,9
ARID1A	NM_006015	89,2	GH1	NM_000515	99,9	ORC6	NM_014321	99,8
ATR	NM_001184	96,3	GHR	NM_000163	99,8	PCNT	NM_006031	94,4
CCDC8	NM_032040	100	GHRHR	NM_000823	99,3	POC1A	NM_015426	99,6
CDC45	NM_001178010	97,4	GHSR	NM_198407	96,5	PTPN11	NM_002834	84,4
CDC6	NM_001254	99,4	GMNN	NM_015895	84,6	RBBP8	NM_002894	95,6
CDT1	NM_030928	91,9	IGF1	NM_001111283	100	ROR2	NM_004560	98,2
CENPJ	NM_018451	97,4	IGF1R	NM_000875	98,8	SHOX*	NM_000451	75,7
CEP152	NM_001194998	94,4	KMT2A	NM_001197104	98,4	SRCAP	NM_006662	98,4
CEP63	NM_025180	90	NIN	NM_020921	97,7	STAT5B	NM_012448	92,3
CHD4	NM_001273	99,8	NPPC	NM_024409	88,5	THRA	NM_003250	99,6
CUL7	NM_001168370	97,4	NPR2	NM_003995	98,8	TRAIP	NM_005879	99,7
DNA2	NM_001080449	96,2	NPR3	NM_001204375	99,9	TRIM37	NM_001005207	97,2
FGD1	NM_004463	85	OBSL1	NM_015311	96,1	WNT5A	NM_003392	99,3

A normal result does not exclude a genetic defect causing short stature completely, as not all analyzed genes are covered 100%. Moreover, not all genes correlated with short stature have been identified. Copy number variations (CNVs), mutations outside the exons and splice sites are not detected with the currently used method. Detailed information regarding the coverage of the genes in the short stature panel for your patient can be provided upon request.

* If a mutation is expected in the *SHOX* gene, then an additional Sanger sequence analysis of the *SHOX* gene is recommended.