TALL STATURE PANEL WITH GENOME WIDE CNV ANALYSIS DG-5.2.0 (40 GENES)

Gene symbols used follow HGNC guidelines: Gray KA, Yates B, Seal RL, Wright MW, Bruford EA. Nucleic Acids Res. 2015 Jan 43(Database issue):D1079-85.
OMIM disease identifiers and descriptions : October 25th, 2026.
This list is accurate for panel version DG 5.2.0
non-protein coding genes are covered, but as coverage statistics are based on protein coding regions, statistics could not be generated.
Ad 1. Blank field signifies a gene without a current OMIM association
Ad 2. OMIM phenotype descriptions between {} signify risk factors
*Ad * This value indicates the percentage of a gene's coding sequence covered at or above the specified Genotype Quality (GQ) threshold, based on alignment against the GRCh38 reference genome. The GQ score is a measure of the confidence of the genotype call and is calculated based on both the quantity and quality of the DNA reads.*
- GQ 20: 99% confidence in the genotype call
- GQ 30: 99.9% confidence in the genotype call

Gene	WES GQ>20 | GQ>30*	srWGS GQ>20 | GQ>30*	lrWGS GQ>20 | GQ>30*	Associated Phenotype description and OMIM disease ID
ABCC9	95% | 95%	98% | 98%	100% | 100%	Cardiomyopathy, dilated, 1O, 608569;Hypertrichotic osteochondrodysplasia (Cantu syndrome), 239850;?Atrial fibrillation, familial, 12, 614050;Intellectual disability and myopathy syndrome, 619719
AKT2	96% | 95%	96% | 96%	99% | 99%	Diabetes mellitus, type II, 125853;Hypoinsulinemic hypoglycemia with hemihypertrophy, 240900
APC2	100% | 100%	95% | 95%	97% | 96%	Cortical dysplasia, complex, with other brain malformations 10, 618677;Intellectual developmental disorder, autosomal recessive 74, 617169
BRWD3	100% | 100%	99% | 99%	100% | 100%	Intellectual developmental disorder, X-linked 93, 300659
CBS	82% | 81%	95% | 95%	98% | 98%	Thrombosis, hyperhomocysteinemic, 236200;Homocystinuria, B6-responsive and nonresponsive types, 236200
CDKN1C	99% | 99%	91% | 91%	97% | 97%	IMAGE syndrome, 614732;Beckwith-Wiedemann syndrome, 130650
CHD8	100% | 100%	97% | 97%	99% | 99%	Intellectual developmental disorder with autism and macrocephaly, 615032
CYP19A1	100% | 100%	98% | 98%	100% | 100%	Aromatase deficiency, 613546
DIS3L2	99% | 99%	97% | 97%	99% | 99%	Perlman syndrome, 267000
DNMT3A	100% | 100%	96% | 96%	99% | 99%	Tatton-Brown-Rahman syndrome, 615879;Acute myeloid leukemia, somatic, 601626;Heyn-Sproul-Jackson syndrome, 618724
EED	99% | 99%	96% | 96%	99% | 99%	Cohen-Gibson syndrome, 617561
EZH2	100% | 100%	97% | 97%	99% | 99%	Weaver syndrome, 277590
FBN1	100% | 100%	98% | 98%	100% | 100%	Geleophysic dysplasia 2, 614185;Weill-Marchesani syndrome 2, dominant, 608328;Ectopia lentis, familial, 129600;MASS syndrome, 604308;Marfan lipodystrophy syndrome, 616914;Acromicric dysplasia, 102370;Marfan syndrome, 154700;Stiff skin syndrome, 184900
FBN2	99% | 99%	98% | 98%	99% | 99%	Macular degeneration, early-onset, 616118;Contractural arachnodactyly, congenital, 121050

FGFR3	100% | 100%	95% | 95%	98% | 96%	Muenke syndrome, 602849;SADDAN, 616482;Hypochondroplasia, 146000;Thanatophoric dysplasia, type II, 187601;Nevus, epidermal, somatic, 162900;CATSHL syndrome, 610474;Thanatophoric dysplasia, type I, 187600;Spermatocytic seminoma, somatic, 273300;Bladder cancer, somatic, 109800;LADD syndrome 2, 620192;Achondroplasia, 100800;Cervical cancer, somatic, 603956;Colorectal cancer, somatic, 114500;Crouzon syndrome with acanthosis nigricans, 612247
FIBP	95% | 95%	95% | 95%	99% | 99%	Thauvin-Robinet-Faivre syndrome, 617107
GPC3	94% | 94%	98% | 98%	100% | 100%	Wilms tumor, somatic, 194070;Simpson-Golabi-Behmel syndrome, type 1, 312870
GPR101	97% | 97%	98% | 98%	100% | 100%	Pituitary adenoma 2, GH-secreting, 300943
H19				
HERC1	100% | 100%	98% | 98%	99% | 99%	Macrocephaly, dysmorphic facies, and psychomotor retardation, 617011
IGF1R	100% | 100%	97% | 97%	99% | 99%	Insulin-like growth factor I, resistance to, 270450
KCNQ1OT1				Beckwith-Wiedemann syndrome, 130650
MED12	99% | 99%	98% | 98%	100% | 100%	Lujan-Fryns syndrome, 309520;Ohdo syndrome, X-linked, 300895;Hardikar syndrome, 301068;Opitz-Kaveggia syndrome, 305450
MTOR	100% | 100%	97% | 97%	99% | 99%	Focal cortical dysplasia, type II, somatic, 607341;Smith-Kingsmore syndrome, 616638
NFIX	100% | 100%	96% | 96%	98% | 98%	Marshall-Smith syndrome, 602535;Malan syndrome, 614753
NKAP	99% | 99%	98% | 98%	100% | 100%	Intellectual developmental disorder, X-linked syndromic, Hackman-Di Donato type, 301039
NPR2	100% | 100%	97% | 97%	99% | 99%	Epiphyseal chondrodysplasia, Miura type, 615923;Short stature with nonspecific skeletal abnormalities, 616255;Acromesomelic dysplasia 1, Maroteaux type, 602875
NPR3	100% | 100%	96% | 96%	99% | 99%	Boudin-Mortier syndrome, 619543
NSD1	100% | 100%	97% | 97%	99% | 99%	Sotos syndrome, 117550
PDGFRB	97% | 97%	97% | 97%	99% | 99%	Premature aging syndrome, Penttinen type, 601812;?Ocular pterygium-digital keloid dysplasia syndrome, 621091;Kosaki overgrowth syndrome, 616592;Myofibromatosis, infantile, 1, 228550;Basal ganglia calcification, idiopathic, 4, 615007
RNF125	100% | 100%	97% | 97%	99% | 98%	Tenorio syndrome, 616260
RNF135	97% | 97%	97% | 97%	99% | 99%	
SETD2	100% | 100%	98% | 98%	99% | 99%	Luscan-Lumish syndrome, 616831;Intellectual developmental disorder, autosomal dominant 70, 620157;Rabin-Pappas syndrome, 620155
SMAD2	86% | 86%	97% | 97%	100% | 100%	Loeys-Dietz syndrome 6, 619656;Congenital heart defects, multiple types, 8, with or without heterotaxy, 619657

SMAD3	100% | 100%	96% | 96%	99% | 99%	Loeys-Dietz syndrome 3, 613795
SUZ12	96% | 96%	97% | 97%	99% | 99%	Imagawa-Matsumoto syndrome, 618786
TGFB2	100% | 100%	97% | 97%	99% | 99%	Loeys-Dietz syndrome 4, 614816;Camurati-Engelmann disease 2, 606631
TGFB3	88% | 88%	97% | 97%	99% | 99%	Arrhythmogenic right ventricular dysplasia 1, 107970;Loeys-Dietz syndrome 5, 615582
TGFBR1	100% | 100%	97% | 97%	100% | 100%	{Multiple self-healing squamous epithelioma, susceptibility to}, 132800;Loeys-Dietz syndrome 1, 609192
TGFBR2	100% | 100%	97% | 97%	100% | 100%	Loeys-Dietz syndrome 2, 610168;Colorectal cancer, hereditary nonpolyposis, type 6, 614331;Esophageal cancer, somatic, 133239