TALL STATURE PANEL WITH GENOME WIDE CNV ANALYSIS DG-5.1.0 (40 GENES)

Gene	*Twist X2 covered 10x*	*Twist X2 covered 20x*	*srWGS covered 10x*	*srWGS covered 15x*	*srWGS covered 20x*	*Associated Phenotype description and OMIM disease ID*
ABCC9	96%	96%	100%	100%	99.8%	Cardiomyopathy, dilated, 1O, 608569;Hypertrichotic osteochondrodysplasia (Cantu syndrome), 239850;?Atrial fibrillation, familial, 12, 614050;Intellectual disability and myopathy syndrome, 619719
AKT2	98.3%	95%	100%	100%	99.4%	Diabetes mellitus, type II, 125853;Hypoinsulinemic hypoglycemia with hemihypertrophy, 240900
APC2	100%	100%	100%	100%	99%	Cortical dysplasia, complex, with other brain malformations 10, 618677;Intellectual developmental disorder, autosomal recessive 74, 617169
BRWD3	100%	100%	99.3%	91.7%	73.7%	Intellectual developmental disorder, X-linked 93, 300659
CBS	87.7%	85.5%	100%	100%	99.5%	Thrombosis, hyperhomocysteinemic, 236200;Homocystinuria, B6-responsive and nonresponsive types, 236200
CDKN1C	100%	100%	100%	100%	99%	IMAGE syndrome, 614732;Beckwith-Wiedemann syndrome, 130650

CHD8	100%	100%	100%	100%	99.5%	Intellectual developmental disorder with autism and macrocephaly, 615032
CYP19A1	100%	100%	100%	100%	99.8%	Aromatase deficiency, 613546
DIS3L2	100%	100%	100%	99.9%	99.5%	Perlman syndrome, 267000
DNMT3A	100%	100%	100%	100%	99.5%	Tatton-Brown-Rahman syndrome, 615879;Acute myeloid leukemia, somatic, 601626;Heyn-Sproul-Jackson syndrome, 618724
EED	99.1%	99.1%	100%	99.6%	97.9%	Cohen-Gibson syndrome, 617561
EZH2	100%	100%	100%	100%	99.7%	Weaver syndrome, 277590
FBN1	100%	100%	100%	100%	99.8%	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	99.2%	99.2%	100%	100%	99.6%	Macular degeneration, early-onset, 616118;Contractural arachnodactyly, congenital, 121050

FGFR3	100%	100%	100%	99.9%	99.1%	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	98.7%	95%	100%	100%	99.5%	Thauvin-Robinet-Faivre syndrome, 617107
GPC3	94%	94%	98.6%	88.7%	70.8%	Wilms tumor, somatic, 194070;Simpson-Golabi-Behmel syndrome, type 1, 312870
GPR101	100%	100%	98.4%	88.9%	67.5%	Pituitary adenoma 2, GH-secreting, 300943
H19						
HERC1	100%	100%	100%	100%	99.7%	Macrocephaly, dysmorphic facies, and psychomotor retardation, 617011
IGF1R	100%	100%	100%	100%	99.7%	Insulin-like growth factor I, resistance to, 270450
KCNQ1OT1						Beckwith-Wiedemann syndrome, 130650

MED12	100%	99.9%	98.1%	87.5%	68.9%	Lujan-Fryns syndrome, 309520;Ohdo syndrome, X-linked, 300895;Hardikar syndrome, 301068;Opitz-Kaveggia syndrome, 305450
MTOR	100%	100%	100%	100%	99.6%	Focal cortical dysplasia, type II, somatic, 607341;Smith-Kingsmore syndrome, 616638
NFIX	100%	100%	100%	100%	98.8%	Marshall-Smith syndrome, 602535;Malan syndrome, 614753
NKAP	100%	100%	99.1%	89.5%	71%	Intellectual developmental disorder, X-linked syndromic, Hackman-Di Donato type, 301039
NPR2	100%	100%	100%	100%	99.5%	Epiphyseal chondrodysplasia, Miura type, 615923;Short stature with nonspecific skeletal abnormalities, 616255;Acromesomelic dysplasia 1, Maroteaux type, 602875
NPR3	100%	100%	100%	100%	99.1%	Boudin-Mortier syndrome, 619543
NSD1	100%	100%	100%	100%	99.5%	Sotos syndrome, 117550
PDGFRB	99.3%	97.4%	100%	100%	99.2%	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%	100%	100%	99.5%	Tenorio syndrome, 616260
RNF135	96%	96%	100%	100%	99.9%	
SETD2	100%	100%	100%	100%	99.7%	Luscan-Lumish syndrome, 616831;Intellectual developmental disorder, autosomal dominant 70, 620157;Rabin-Pappas syndrome, 620155
SMAD2	86.7%	86.7%	100%	100%	99.8%	Loeys-Dietz syndrome 6, 619656;Congenital heart defects, multiple types, 8, with or without heterotaxy, 619657
SMAD3	100%	100%	100%	100%	99.4%	Loeys-Dietz syndrome 3, 613795
SUZ12	95.6%	95.6%	100%	100%	99.7%	Imagawa-Matsumoto syndrome, 618786
TGFB2	100%	100%	100%	99.8%	99.4%	Loeys-Dietz syndrome 4, 614816;Camurati-Engelmann disease 2, 606631
TGFB3	88.3%	88.3%	100%	100%	99.7%	Arrhythmogenic right ventricular dysplasia 1, 107970;Loeys-Dietz syndrome 5, 615582
TGFBR1	100%	100%	100%	100%	99.6%	{Multiple self-healing squamous epithelioma, susceptibility to}, 132800;Loeys-Dietz syndrome 1, 609192
TGFBR2	100%	100%	100%	100%	99.6%	Loeys-Dietz syndrome 2, 610168;Colorectal cancer, hereditary nonpolyposis, type 6, 614331;Esophageal cancer, somatic, 133239

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.

TWIST X2 covered 10x describes the percentage of a gene's coding sequence that is covered at least 10x when analyzed by WES using TWIST X2 chemistry mapped against GRCh38.
TWIST X2 covered 20x describes the percentage of a gene's coding sequence that is covered at least 20x when analyzed by WES using TWIST X2 chemistry mapped against

GRCh38.

srWGS covered 10x describes the percentage of a gene's coding sequence that is covered at least 10x when analyzed by WGS mapped against GRCh38.

srWGS covered 15x describes the percentage of a gene's coding sequence that is covered at least 15x when analyzed by WGS mapped against GRCh38.

srWGS covered 20x describes the percentage of a gene's coding sequence that is covered at least 20x when analyzed by WGS mapped against GRCh38.

non-protein coding genes are covered, but as coverage statistics are based on protein coding regions, statistics could not be generated.

OMIM release used for OMIM disease identifiers and descriptions : November 25th, 2024.

This list is accurate for panel version DG 5.1.0

Ad 1. Blank field signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors