TALL STATURE PANEL WITH GENOME WIDE CNV ANALYSIS DG-3.9.0 (40 GENES)

Gene	*Twist X2 covered >10x*	*Twist X2 covered >20x*	*WGS covered >10x*	*WGS covered >20x*	*Associated Phenotype description and OMIM disease ID*
ABCC9	100.0%	100.0%	100.0%	98.4%	Cardiomyopathy, dilated, 1O, 608569;Hypertrichotic osteochondrodysplasia (Cantu syndrome), 239850;?Atrial fibrillation, familial, 12, 614050;Intellectual disability and myopathy syndrome, 619719
AKT2	100.0%	100.0%	100.0%	98.6%	Diabetes mellitus, type II, 125853;Hypoinsulinemic hypoglycemia with hemihypertrophy, 240900
APC2	100.0%	100.0%	100.0%	96.8%	Cortical dysplasia, complex, with other brain malformations 10, 618677;Intellectual developmental disorder, autosomal recessive 74, 617169
BRWD3	100.0%	99.7%	97.7%	71.0%	Intellectual developmental disorder, X-linked 93, 300659

CBS	100.0%	100.0%	100.0%	99.5%	Thrombosis, hyperhomocysteinemic, 236200;Homocystinuria, B6-responsive and nonresponsive types, 236200
CDKN1C	100.0%	100.0%	100.0%	92.1%	IMAGE syndrome, 614732;Beckwith-Wiedemann syndrome, 130650
CHD8	100.0%	100.0%	100.0%	98.4%	Intellectual developmental disorder with autism and macrocephaly, 615032
CYP19A1	100.0%	99.9%	100.0%	98.8%	Aromatase deficiency, 613546;Aromatase excess syndrome, 139300
DIS3L2	100.0%	100.0%	100.0%	98.7%	Perlman syndrome, 267000
DNMT3A	100.0%	100.0%	100.0%	99.4%	Tatton-Brown-Rahman syndrome, 615879;Acute myeloid leukemia, somatic, 601626;Heyn-Sproul-Jackson syndrome, 618724
EED	100.0%	100.0%	99.9%	95.0%	Cohen-Gibson syndrome, 617561
EZH2	100.0%	100.0%	100.0%	99.0%	Weaver syndrome, 277590

FBN1	100.0%	100.0%	100.0%	99.1%	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	100.0%	100.0%	100.0%	99.4%	Macular degeneration, early-onset, 616118;Contractural arachnodactyly, congenital, 121050

FGFR3	100.0%	100.0%	100.0%	99.8%	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	100.0%	100.0%	100.0%	98.7%	Thauvin-Robinet-Faivre syndrome, 617107
GPC3	99.6%	98.9%	97.7%	68.3%	Wilms tumor, somatic, 194070;Simpson-Golabi-Behmel syndrome, type 1, 312870
GPR101	100.0%	100.0%	97.9%	69.4%	Pituitary adenoma 2, GH-secreting, 300943
H19					

HERC1	100.0%	100.0%	100.0%	99.2%	Macrocephaly, dysmorphic facies, and psychomotor retardation, 617011
IGF1R	100.0%	100.0%	100.0%	99.1%	Insulin-like growth factor I, resistance to, 270450
KCNQ1OT1					Beckwith-Wiedemann syndrome, 130650
MED12	100.0%	99.8%	97.5%	69.0%	Lujan-Fryns syndrome, 309520;Ohdo syndrome, X-linked, 300895;Hardikar syndrome, 301068;Opitz-Kaveggia syndrome, 305450
MTOR	100.0%	100.0%	100.0%	99.3%	Focal cortical dysplasia, type II, somatic, 607341;Smith-Kingsmore syndrome, 616638
NFIX	100.0%	99.7%	99.7%	96.9%	Marshall-Smith syndrome, 602535;Malan syndrome, 614753
NKAP	100.0%	100.0%	96.4%	67.3%	Intellectual developmental disorder, X-linked syndromic, Hackman-Di Donato type, 301039
NPR2	100.0%	100.0%	100.0%	99.1%	Epiphyseal chondrodysplasia, Miura type, 615923;Short stature with nonspecific skeletal abnormalities, 616255;Acromesomelic dysplasia 1, Maroteaux type, 602875

NPR3	100.0%	100.0%	100.0%	98.7%	Boudin-Mortier syndrome, 619543
NSD1	100.0%	100.0%	100.0%	98.6%	Sotos syndrome, 117550
PDGFRB	100.0%	100.0%	100.0%	99.2%	Premature aging syndrome, Penttinen type, 601812;Kosaki overgrowth syndrome, 616592;Myofibromatosis, infantile, 1, 228550;Basal ganglia calcification, idiopathic, 4, 615007;Myeloproliferative disorder with eosinophilia, 131440
RNF125	100.0%	100.0%	100.0%	99.0%	Tenorio syndrome, 616260
RNF135	100.0%	100.0%	100.0%	98.8%	
SETD2	100.0%	100.0%	100.0%	97.9%	Luscan-Lumish syndrome, 616831;Intellectual developmental disorder, autosomal dominant 70, 620157;Rabin-Pappas syndrome, 620155
SMAD2	100.0%	100.0%	100.0%	99.0%	Loeys-Dietz syndrome 6, 619656;Congenital heart defects, multiple types, 8, with or without heterotaxy, 619657
SMAD3	100.0%	100.0%	99.9%	96.8%	Loeys-Dietz syndrome 3, 613795
SUZ12	100.0%	100.0%	100.0%	94.8%	Imagawa-Matsumoto syndrome, 618786

TGFB2	100.0%	100.0%	100.0%	98.4%	Loeys-Dietz syndrome 4, 614816
TGFB3	100.0%	100.0%	100.0%	99.5%	Arrhythmogenic right ventricular dysplasia 1, 107970;Loeys-Dietz syndrome 5, 615582
TGFBR1	100.0%	100.0%	100.0%	96.7%	{Multiple self-healing squamous epithelioma, susceptibility to}, 132800;Loeys-Dietz syndrome 1, 609192
TGFBR2	100.0%	100.0%	100.0%	98.5%	Loeys-Dietz syndrome 2, 610168;Colorectal cancer, hereditary nonpolyposis, type 6, 614331;Esophageal cancer, somatic, 133239

Gene symbols used follow HGCN 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.
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.
srWGS GRCh38 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 GRCh38 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 : March 17th, 2023.
This list is accurate for panel version DG 3.9.0