WES TALL STATURE DG 3.8.1

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%	99.4%	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%	99.7%	Diabetes mellitus, type II, 125853;Hypoinsulinemic hypoglycemia with hemihypertrophy, 240900
APC2	100.0%	100.0%	100.0%	99.7%	Cortical dysplasia, complex, with other brain malformations 10, 618677;Intellectual developmental disorder, autosomal recessive 74, 617169
BRWD3	100.0%	99.7%	98.5%	74.1%	Intellectual developmental disorder, X-linked 93, 300659
CBS	100.0%	100.0%	100.0%	100.0%	Thrombosis, hyperhomocysteinemic, 236200;Homocystinuria, B6-responsive and nonresponsive types, 236200
CDKN1C	100.0%	100.0%	100.0%	99.8%	IMAGE syndrome, 614732;Beckwith-Wiedemann syndrome, 130650
CHD8	100.0%	100.0%	100.0%	99.3%	Intellectual developmental disorder with autism and macrocephaly, 615032

Gene				Disorder
CYP19A1	100.0%	99.9%	100.0%	99.4%	Aromatase deficiency, 613546;Aromatase excess syndrome, 139300
DIS3L2	100.0%	100.0%	100.0%	99.5%	Perlman syndrome, 267000
DNMT3A	100.0%	100.0%	100.0%	99.8%	Tatton-Brown-Rahman syndrome, 615879;Acute myeloid leukemia, somatic, 601626;Heyn-Sproul-Jackson syndrome, 618724
EED	100.0%	100.0%	100.0%	98.4%	Cohen-Gibson syndrome, 617561
EZH2	100.0%	100.0%	100.0%	99.6%	Weaver syndrome, 277590
FBN1	100.0%	100.0%	100.0%	99.6%	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%	99.7%	Macular degeneration, early-onset, 616118;Contractural arachnodactyly, congenital, 121050

FGFR3	100.0%	100.0%	100.0%	100.0%	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%	99.8%	Thauvin-Robinet-Faivre syndrome, 617107
GPC3	99.6%	98.9%	98.1%	72.5%	Wilms tumor, somatic, 194070;Simpson-Golabi-Behmel syndrome, type 1, 312870
GPR101	100.0%	100.0%	98.5%	71.5%	Pituitary adenoma 2, GH-secreting, 300943
H19					
HERC1	100.0%	100.0%	100.0%	99.7%	Macrocephaly, dysmorphic facies, and psychomotor retardation, 617011
IGF1R	100.0%	100.0%	100.0%	99.7%	Insulin-like growth factor I, resistance to, 270450
KCNQ1OT1					Beckwith-Wiedemann syndrome, 130650
MED12	100.0%	99.8%	98.2%	71.9%	Lujan-Fryns syndrome, 309520;Ohdo syndrome, X-linked, 300895;Hardikar syndrome, 301068;Opitz-Kaveggia syndrome, 305450

MTOR	100.0%	100.0%	100.0%	99.7%	Focal cortical dysplasia, type II, somatic, 607341;Smith-Kingsmore syndrome, 616638
NFIX	100.0%	99.7%	99.9%	98.6%	Marshall-Smith syndrome, 602535;Malan syndrome, 614753
NKAP	100.0%	100.0%	98.5%	73.7%	Intellectual developmental disorder, X-linked syndromic, Hackman-Di Donato type, 301039
NPR2	100.0%	100.0%	100.0%	99.8%	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%	99.8%	Boudin-Mortier syndrome, 619543
NSD1	100.0%	100.0%	100.0%	99.3%	Sotos syndrome, 117550
PDGFRB	100.0%	100.0%	100.0%	99.8%	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%	99.9%	Tenorio syndrome, 616260
RNF135	100.0%	100.0%	100.0%	99.6%	
SETD2	100.0%	100.0%	100.0%	99.1%	Luscan-Lumish syndrome, 616831;Intellectual developmental disorder, autosomal dominant 70, 620157;Rabin-Pappas syndrome, 620155
SMAD2	100.0%	100.0%	100.0%	99.8%	Loeys-Dietz syndrome 6, 619656;Congenital heart defects, multiple types, 8, with or without heterotaxy, 619657

Gene	TWIST X2 Covered 10x	TWIST X2 Covered 20x	srWGS GRCh38 Covered 10x	srWGS GRCh38 Covered 20x	OMIM phenotype
SMAD3	100.0%	100.0%	100.0%	99.1%	Loeys-Dietz syndrome 3, 613795
SUZ12	100.0%	100.0%	100.0%	97.8%	Imagawa-Matsumoto syndrome, 618786
TGFB2	100.0%	100.0%	100.0%	99.5%	Loeys-Dietz syndrome 4, 614816
TGFB3	100.0%	100.0%	100.0%	99.8%	Arrhythmogenic right ventricular dysplasia 1, 107970;Loeys-Dietz syndrome 5, 615582
TGFBR1	100.0%	100.0%	100.0%	99.4%	{Multiple self-healing squamous epithelioma, susceptibility to}, 132800;Loeys-Dietz syndrome 1, 609192
TGFBR2	100.0%	100.0%	100.0%	99.1%	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 3.8.1

Ad 1. "No OMIM phenotype" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors