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TALL STATURE PANEL WITH GENOME WIDE CNV ANALYSIS DG-5.2.0 (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	n.t.b					Cardiomyopathy, dilated, 1O, 608569;Hypertrichotic osteochondrodysplasia (Cantu syndrome), 239850;?Atrial fibrillation, familial, 12, 614050;Intellectual disability and myopathy syndrome, 619719
AKT2	n.t.b					Diabetes mellitus, type II, 125853;Hypoinsulinemic hypoglycemia with hemihypertrophy, 240900
APC2	n.t.b					Cortical dysplasia, complex, with other brain malformations 10, 618677;Intellectual developmental disorder, autosomal recessive 74, 617169
BRWD3	n.t.b					Intellectual developmental disorder, X-linked 93, 300659
CBS	n.t.b					Thrombosis, hyperhomocysteinemic, 236200;Homocystinuria, B6-responsive and nonresponsive types, 236200
CDKN1C	n.t.b					IMAGE syndrome, 614732;Beckwith-Wiedemann syndrome, 130650

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CHD8	n.t.b					Intellectual developmental disorder with autism and macrocephaly, 615032
CYP19A1	n.t.b					Aromatase deficiency, 613546
DIS3L2	n.t.b					Perlman syndrome, 267000
DNMT3A	n.t.b					Tatton-Brown-Rahman syndrome, 615879;Acute myeloid leukemia, somatic, 601626;Heyn-Sproul-Jackson syndrome, 618724
EED	n.t.b					Cohen-Gibson syndrome, 617561
EZH2	n.t.b					Weaver syndrome, 277590
FBN1	n.t.b					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	n.t.b					Macular degeneration, early-onset, 616118;Contractural arachnodactyly, congenital, 121050

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FGFR3	n.t.b					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	n.t.b					Thauvin-Robinet-Faivre syndrome, 617107
GPC3	n.t.b					Wilms tumor, somatic, 194070;Simpson-Golabi-Behmel syndrome, type 1, 312870
GPR101	n.t.b					Pituitary adenoma 2, GH-secreting, 300943
H19	n.t.b					
HERC1	n.t.b					Macrocephaly, dysmorphic facies, and psychomotor retardation, 617011
IGF1R	n.t.b					Insulin-like growth factor I, resistance to, 270450
KCNQ1OT1	n.t.b					Beckwith-Wiedemann syndrome, 130650

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MED12	n.t.b					Lujan-Fryns syndrome, 309520;Ohdo syndrome, X-linked, 300895;Hardikar syndrome, 301068;Opitz-Kaveggia syndrome, 305450
MTOR	n.t.b					Focal cortical dysplasia, type II, somatic, 607341;Smith-Kingsmore syndrome, 616638
NFIX	n.t.b					Marshall-Smith syndrome, 602535;Malan syndrome, 614753
NKAP	n.t.b					Intellectual developmental disorder, X-linked syndromic, Hackman-Di Donato type, 301039
NPR2	n.t.b					Epiphyseal chondrodysplasia, Miura type, 615923;Short stature with nonspecific skeletal abnormalities, 616255;Acromesomelic dysplasia 1, Maroteaux type, 602875
NPR3	n.t.b					Boudin-Mortier syndrome, 619543
NSD1	n.t.b					Sotos syndrome, 117550
PDGFRB	n.t.b					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

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RNF125	n.t.b					Tenorio syndrome, 616260
RNF135	n.t.b					
SETD2	n.t.b					Luscan-Lumish syndrome, 616831;Intellectual developmental disorder, autosomal dominant 70, 620157;Rabin-Pappas syndrome, 620155
SMAD2	n.t.b					Loeys-Dietz syndrome 6, 619656;Congenital heart defects, multiple types, 8, with or without heterotaxy, 619657
SMAD3	n.t.b					Loeys-Dietz syndrome 3, 613795
SUZ12	n.t.b					Imagawa-Matsumoto syndrome, 618786
TGFB2	n.t.b					Loeys-Dietz syndrome 4, 614816;Camurati-Engelmann disease 2, 606631
TGFB3	n.t.b					Arrhythmogenic right ventricular dysplasia 1, 107970;Loeys-Dietz syndrome 5, 615582
TGFBR1	n.t.b					{Multiple self-healing squamous epithelioma, susceptibility to}, 132800;Loeys-Dietz syndrome 1, 609192
TGFBR2	n.t.b					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.

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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