FETAL AKINESIA PANEL DG-4.2.0 (103 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*
ACTA1	100%	100%	100%	99.8%	97.2%	Congenital myopathy 2B, severe infantile, autosomal recessive, 620265;?Myopathy, scapulohumeroperoneal, 616852;Congenital myopathy 2C, severe infantile, autosomal dominant, 620278;Congenital myopathy 2A, typical, autosomal dominant, 161800
ADAMTS15	100%	100%	100%	100%	99.1%	Arthrogryposis, distal, type 12, 620545
ADCY6	100%	100%	100%	100%	99.4%	Lethal congenital contracture syndrome 8, 616287
ADGRG6	100%	100%	100%	100%	99.6%	Lethal congenital contracture syndrome 9, 616503
ALG3	100%	100%	100%	100%	98.9%	Congenital disorder of glycosylation, type Id, 601110

ASCC1	86.6%	86.6%	100%	100%	99.3%	Spinal muscular atrophy with congenital bone fractures 2, 616867;Barrett esophagus/esophageal adenocarcinoma, 614266
ATP1A2	100%	100%	100%	100%	98.9%	Developmental and epileptic encephalopathy 98, 619605;Fetal akinesia, respiratory insufficiency, microcephaly, polymicrogyria, and dysmorphic facies, 619602;Alternating hemiplegia of childhood 1, 104290;Migraine, familial basilar, 602481;Migraine, familial hemiplegic, 2, 602481
BICD2	100%	100%	100%	100%	99.5%	Spinal muscular atrophy, lower extremity-predominant, 2B, autosomal dominant, 618291;Spinal muscular atrophy, lower extremity-predominant, 2A, autosomal dominant, 615290

BIN1	100%	100%	100%	99.8%	98.8%	Centronuclear myopathy 2, 255200
BLTP1	100%	100%	100%	100%	99.4%	Alkuraya-Kucinskas syndrome, 617822
CACNA1S	100%	100%	100%	100%	99.1%	{Thyrotoxic periodic paralysis, susceptibility to, 1}, 188580;Congenital myopathy 18 due to dihydropyridine receptor defect, 620246;Hypokalemic periodic paralysis, type 1, 170400;{Malignant hyperthermia susceptibility 5}, 601887
CHAT	100%	99.9%	100%	100%	99.3%	Myasthenic syndrome, congenital, 6, presynaptic, 254210
CHRNA1	100%	100%	100%	100%	99.2%	Myasthenic syndrome, congenital, 1B, fast-channel, 608930;Myasthenic syndrome, congenital, 1A, slow-channel, 601462;Multiple pterygium syndrome, lethal type, 253290

CHRNB1	100%	100%	100%	99.9%	98%	?Myasthenic syndrome, congenital, 2C, associated with acetylcholine receptor deficiency, 616314;Myasthenic syndrome, congenital, 2A, slow-channel, 616313
CHRND	100%	100%	100%	100%	98.9%	?Myasthenic syndrome, congenital, 3C, associated with acetylcholine receptor deficiency, 616323;Multiple pterygium syndrome, lethal type, 253290;Myasthenic syndrome, congenital, 3B, fast-channel, 616322;?Myasthenic syndrome, congenital, 3A, slow-channel, 616321

CHRNE	100%	100%	100%	100%	98.9%	Myasthenic syndrome, congenital, 4A, slow-channel, 605809;Myasthenic syndrome, congenital, 4C, associated with acetylcholine receptor deficiency, 608931;Myasthenic syndrome, congenital, 4B, fast-channel, 616324
CHRNG	100%	100%	100%	100%	99%	Multiple pterygium syndrome, lethal type, 253290;Escobar syndrome, 265000
CHST14	100%	100%	100%	99.8%	97.5%	Ehlers-Danlos syndrome, musculocontractural type 1, 601776
CNTNAP1	100%	100%	100%	99.9%	98.7%	Lethal congenital contracture syndrome 7, 616286;Hypomyelinating neuropathy, congenital, 3, 618186
COL12A1	100%	100%	100%	100%	99.6%	Bethlem myopathy 2, 616471;?Ullrich congenital muscular dystrophy 2, 616470

COL6A1	100%	100%	100%	99.8%	98.8%	Ullrich congenital muscular dystrophy 1A, 254090;Bethlem myopathy 1A, 158810
COL6A2	100%	100%	100%	100%	98.9%	?Myosclerosis, congenital, 255600;Ullrich congenital muscular dystrophy 1B, 620727;Bethlem myopathy 1B, 620725
COL6A3	100%	100%	100%	99.9%	99.4%	Bethlem myopathy 1C, 620726;Ullrich congenital muscular dystrophy 1C, 620728;Dystonia 27, 616411
COX15	100%	100%	100%	100%	99.1%	Mitochondrial complex IV deficiency, nuclear type 6, 615119
CRPPA	100%	100%	100%	100%	99.6%	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 7, 616052;Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 7, 614643
DHCR24	100%	100%	100%	99.8%	98.7%	Desmosterolosis, 602398
DHCR7	96.2%	96.2%	100%	99.9%	99.2%	Smith-Lemli-Opitz syndrome, 270400

DNM2	100%	100%	100%	100%	99.2%	Centronuclear myopathy 1, 160150;Charcot-Marie-Tooth disease, axonal type 2M, 606482;Charcot-Marie-Tooth disease, dominant intermediate B, 606482;Lethal congenital contracture syndrome 5, 615368
DOK7	100%	100%	100%	100%	98.5%	Fetal akinesia deformation sequence 3, 618389;Myasthenic syndrome, congenital, 10, 254300
ECEL1	100%	100%	100%	99.9%	98.8%	Arthrogryposis, distal, type 5D, 615065
EGR2	100%	100%	100%	100%	98.8%	Dejerine-Sottas disease, 145900;Charcot-Marie-Tooth disease, type 1D, 607678;Hypomyelinating neuropathy, congenital, 1, 605253

ERBB3	100%	100%	100%	99.9%	98.3%	?Lethal congenital contractural syndrome 2, 607598;{?Erythroleukemia, familial, susceptibility to}, 133180;Visceral neuropathy, familial, 1, autosomal recessive, 243180
ERCC5	100%	100%	100%	99.9%	99.3%	Xeroderma pigmentosum, group G, 278780;Cerebrooculofacioskeletal syndrome 3, 616570;Xeroderma pigmentosum, group G/Cockayne syndrome, 278780
ERCC6	100%	100%	100%	100%	99.3%	UV-sensitive syndrome 1, 600630;Cerebrooculofacioskeletal syndrome 1, 214150;?De Sanctis-Cacchione syndrome, 278800;Cockayne syndrome, type B, 133540;{Macular degeneration, age-related, susceptibility to, 5}, 613761;Premature ovarian failure 11, 616946;{Lung cancer, susceptibility to}, 211980

FBN2	99.2%	99.2%	100%	100%	99.4%	Macular degeneration, early-onset, 616118;Contractural arachnodactyly, congenital, 121050
FKRP	100%	100%	100%	99.9%	98.6%	Muscular dystrophy-dystroglycanopathy (congenital with or without impaired intellectual development), type B, 5, 606612;Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 5, 607155;Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 5, 613153
FLNC	100%	100%	100%	100%	99%	Cardiomyopathy, familial hypertrophic, 26, 617047;Arrhythmogenic right ventricular dysplasia, familial, 617047;Cardiomyopathy, familial restrictive 5, 617047;Myopathy, distal, 4, 614065;Myopathy, myofibrillar, 5, 609524

FLVCR2	100%	100%	100%	99.9%	99.4%	Proliferative vasculopathy and hydranencephaly-hydrocephaly syndrome, 225790
GBA1	100%	100%	100%	100%	99.3%	{Lewy body dementia, susceptibility to}, 127750;Gaucher disease, type II, 230900;Gaucher disease, type IIIC, 231005;Gaucher disease, type III, 231000;Gaucher disease, type I, 230800;Gaucher disease, perinatal lethal, 608013;{Parkinson disease, late-onset, susceptibility to}, 168600
GBE1	100%	100%	100%	100%	99.5%	Glycogen storage disease IV, 232500;Polyglucosan body disease, adult form, 263570
GLDN	100%	100%	100%	100%	99.4%	Lethal congenital contracture syndrome 11, 617194

GLE1	100%	100%	100%	100%	99.1%	Lethal congenital contracture syndrome 1, 253310;Congenital arthrogryposis with anterior horn cell disease, 611890
GMPPB	100%	100%	100%	100%	99.2%	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 14, 615352;Muscular dystrophy-dystroglycanopathy (congenital with impaired intellectual development), type B, 14, 615351;Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 14, 615350
IGHMBP2	100%	100%	100%	100%	99.6%	Charcot-Marie-Tooth disease, axonal, type 2S, 616155;Neuronopathy, distal hereditary motor, autosomal recessive 1, 604320

KIDINS220	100%	100%	100%	100%	99.5%	Spastic paraplegia, intellectual disability, nystagmus, and obesity, 617296;Ventriculomegaly and arthrogryposis, 619501
KIF5C	100%	100%	100%	99.9%	99.4%	Cortical dysplasia, complex, with other brain malformations 2, 615282
KLHL40	100%	100%	100%	99.9%	98.8%	Nemaline myopathy 8, autosomal recessive, 615348
KLHL41	100%	100%	100%	100%	99.8%	Nemaline myopathy 9, 615731
LGI4	100%	100%	100%	99.9%	98.4%	Arthrogryposis multiplex congenita 1, neurogenic, with myelin defect, 617468

LMNA	100%	100%	100%	99.9%	99.1%	Mandibuloacral dysplasia, 248370;Heart-hand syndrome, Slovenian type, 610140;Cardiomyopathy, dilated, 1A, 115200;Emery-Dreifuss muscular dystrophy 3, autosomal recessive, 616516;Restrictive dermopathy 2, 619793;Charcot-Marie-Tooth disease, type 2B1, 605588;Emery-Dreifuss muscular dystrophy 2, autosomal dominant, 181350;Hutchinson-Gilford progeria, 176670;Lipodystrophy, familial partial, type 2, 151660;Muscular dystrophy, congenital, 613205;Malouf syndrome, 212112
LMOD3	100%	100%	99.9%	99.5%	97.4%	Nemaline myopathy 10, 616165
MAGEL2	100%	100%	100%	99.9%	98.7%	Schaaf-Yang syndrome, 615547

MEGF10	100%	100%	100%	100%	99.2%	Congenital myopathy 10A, severe variant, 614399;Congenital myopathy 10B, mild variant, 620249
MPZ	100%	100%	100%	100%	98.6%	Charcot-Marie-Tooth disease, type 2I, 607677;Dejerine-Sottas disease, 145900;Charcot-Marie-Tooth disease, type 1B, 118200;Roussy-Levy syndrome, 180800;Charcot-Marie-Tooth disease, dominant intermediate D, 607791;Hypomyelinating neuropathy, congenital, 2, 618184;Charcot-Marie-Tooth disease, type 2J, 607736
MTM1	100%	100%	99.4%	89.9%	69.2%	Myopathy, centronuclear, X-linked, 310400
MUSK	100%	100%	100%	100%	98.9%	Fetal akinesia deformation sequence 1, 208150;Myasthenic syndrome, congenital, 9, associated with acetylcholine receptor deficiency, 616325

MYBPC1	100%	100%	100%	100%	99.5%	Congenital myopathy 16, 618524;Lethal congenital contracture syndrome 4, 614915;Arthrogryposis, distal, type 1B, 614335
MYCN	100%	99.4%	100%	99.8%	98.6%	Feingold syndrome 1, 164280;Megalencephaly-polydactyly syndrome, 620748
MYH3	100%	100%	100%	100%	99.3%	Contractures, pterygia, and spondylocarpostarsal fusion syndrome 1A, 178110;Contractures, pterygia, and spondylocarpotarsal fusion syndrome 1B, 618469;Arthrogryposis, distal, type 2B3 (Sheldon-Hall), 618436;Arthrogryposis, distal, type 2A (Freeman-Sheldon), 193700
MYH8	100%	100%	100%	100%	99.5%	Carney complex variant, 608837;Trismus-pseudocamptodactyly syndrome, 158300
MYL11	100%	100%	100%	99.8%	98.3%	Arthrogryposis, distal, type 1C, 619110

MYMK	100%	100%	100%	99.9%	98.6%	Carey-Fineman-Ziter syndrome, 254940
MYOD1	100%	100%	100%	100%	98.5%	Congenital myopathy 17, 618975
NEB	99.9%	99.8%	99.7%	99.2%	98%	Nemaline myopathy 2, autosomal recessive, 256030;Arthrogryposis multiplex congenita 6, 619334
NEK9	100%	100%	100%	99.9%	99.5%	?Arthrogryposis, Perthes disease, and upward gaze palsy, 614262;Nevus comedonicus, somatic, 617025;Lethal congenital contracture syndrome 10, 617022
NUP88	93.4%	93.4%	100%	100%	99.5%	Fetal akinesia deformation sequence 4, 618393
PHGDH	100%	100%	100%	100%	99.4%	Neu-Laxova syndrome 1, 256520;Phosphoglycerate dehydrogenase deficiency, 601815

PIEZO2	100%	100%	100%	99.9%	99.1%	Arthrogryposis, distal, type 5, 108145;Arthrogryposis, distal, with impaired proprioception and touch, 617146;Arthrogryposis, distal, type 3, 114300;?Marden-Walker syndrome, 248700
PIP5K1C	100%	100%	100%	100%	98.8%	Lethal congenital contractural syndrome 3, 611369
PLOD1	100%	100%	100%	99.8%	97.7%	Ehlers-Danlos syndrome, kyphoscoliotic type, 1, 225400
PSAT1	100%	100%	100%	100%	99.8%	Neu-Laxova syndrome 2, 616038;Phosphoserine aminotransferase deficiency, 610992
PTRH2	100%	100%	100%	100%	99.6%	Infantile-onset multisystem neurologic, endocrine, and pancreatic disease, 616263

RAPSN	100%	100%	100%	99.8%	98.3%	Fetal akinesia deformation sequence 2, 618388;Myasthenic syndrome, congenital, 11, associated with acetylcholine receptor deficiency, 616326
RIPK4	100%	99.9%	100%	99.9%	99.3%	CHAND syndrome, 214350;Popliteal pterygium syndrome, Bartsocas-Papas type 1, 263650
RYR1	99.9%	99.6%	100%	99.9%	98%	Congenital myopathy 1B, autosomal recessive, 255320;Congenital myopathy 1A, autosomal dominant, with susceptibility to malignant hyperthermia, 117000;King-Denborough syndrome, 619542;{Malignant hyperthermia susceptibility 1}, 145600

SCN4A	100%	100%	100%	100%	98.9%	Paramyotonia congenita, 168300;Hyperkalemic periodic paralysis, 170500;Congenital myopathy 22B, severe fetal, 620369;Hypokalemic periodic paralysis, type 2, 613345;Myotonia congenita, atypical, acetazolamide-responsive, 608390;Myasthenic syndrome, congenital, 16, 614198;Congenital myopathy 22A, classic, 620351
SCYL2	100%	100%	100%	100%	99.5%	Arthrogryposis multiplex congenita 4, neurogenic, with agenesis of the corpus callosum, 618766
SELENON	90.4%	86.7%	100%	99.8%	98.3%	Congenital myopathy 3 with rigid spine, 602771
SLC35A3	97.6%	90%	100%	100%	99.4%	Arthrogryposis, impaired intellectual development, and seizures, 615553

SLC5A7	100%	100%	100%	99.9%	98.9%	Neuronopathy, distal hereditary motor, autosomal dominant 7, 158580;Myasthenic syndrome, congenital, 20, presynaptic, 617143
SLC6A9	100%	100%	100%	99.9%	98.6%	Glycine encephalopathy with normal serum glycine, 617301
SMPD4	100%	100%	100%	99.9%	99.2%	Neurodevelopmental disorder with microcephaly, arthrogryposis, and structural brain anomalies, 618622
SRPK3	100%	99.3%	98.4%	86.4%	66%	Intellectual developmental disorder, X-linked 114, 301134
STAC3	100%	100%	100%	100%	99.2%	Congenital myopathy 13, 255995
SYNE1	100%	100%	100%	100%	99.5%	Arthrogryposis multiplex congenita 3, myogenic type, 618484;Emery-Dreifuss muscular dystrophy 4, autosomal dominant, 612998;Spinocerebellar ataxia, autosomal recessive 8, 610743

TBCD	91.2%	90.6%	100%	99.9%	98.5%	Encephalopathy, progressive, early-onset, with brain atrophy and thin corpus callosum, 617193
TNNI2	100%	100%	100%	100%	99.6%	Arthrogryposis, distal, type 2B1, 601680
TNNT3	100%	100%	100%	99.9%	99%	Arthrogryposis, distal, type 2B2, 618435
TOR1A	93.1%	90.8%	100%	100%	99.2%	{Dystonia-1, modifier of};Arthrogryposis multiplex congenita 5, 618947;Dystonia-1, torsion, 128100
TPM2	100%	100%	100%	99.9%	98.8%	Arthrogryposis, distal, type 2B4, 108120;Arthrogryposis, distal, type 1A, 108120;Congenital myopathy 23, 609285
TPM3	100%	100%	100%	100%	99%	Congenital myopathy 4A, autosomal dominant, 255310;Congenital myopathy 4B, autosomal recessive, 609284

TRIP4	100%	100%	100%	100%	99.2%	?Muscular dystrophy, congenital, Davignon-Chauveau type, 617066;Spinal muscular atrophy with congenital bone fractures 1, 616866
TRPV4	100%	100%	100%	99.9%	98.5%	Neuronopathy, distal hereditary motor, autosomal dominant 8, 600175;Spondylometaphyseal dysplasia, Kozlowski type, 184252;Digital arthropathy-brachydactyly, familial, 606835;[Sodium serum level QTL 1], 613508;SED, Maroteaux type, 184095;Metatropic dysplasia, 156530;Scapuloperoneal spinal muscular atrophy, 181405;Hereditary motor and sensory neuropathy, type IIc, 606071;?Avascular necrosis of femoral head, primary, 2, 617383;Parastremmatic dwarfism, 168400;Brachyolmia type 3, 113500

TTN	100%	100%	100%	99.9%	99.5%	Muscular dystrophy, limb-girdle, autosomal recessive 10, 608807;Cardiomyopathy, familial hypertrophic, 9, 613765;Congenital myopathy 5 with cardiomyopathy, 611705;Tibial muscular dystrophy, tardive, 600334;Cardiomyopathy, dilated, 1G, 604145;Myopathy, myofibrillar, 9, with early respiratory failure, 603689
TUBA1A	100%	100%	100%	100%	99.2%	Lissencephaly 3, 611603
TUBB2B	100%	100%	100%	100%	99.2%	Cortical dysplasia, complex, with other brain malformations 7, 610031
UBA1	100%	99.7%	98.9%	87.8%	68%	Spinal muscular atrophy, X-linked 2, infantile, 301830;VEXAS syndrome, somatic, 301054
VAMP1	100%	100%	100%	100%	98.6%	Myasthenic syndrome, congenital, 25, 618323;Spastic ataxia 1, autosomal dominant, 108600

VIPAS39	100%	100%	100%	100%	99.4%	Arthrogryposis, renal dysfunction, and cholestasis 2, 613404
VPS33B	100%	100%	100%	99.9%	98.6%	Keratoderma-ichthyosis-deafness syndrome, autosomal recessive, 620009;Cholestasis, progressive familial intrahepatic, 12, 620010;Arthrogryposis, renal dysfunction, and cholestasis 1, 208085
WDR62	98.8%	98.7%	100%	100%	99.3%	Microcephaly 2, primary, autosomal recessive, with or without cortical malformations, 604317
ZC4H2	100%	99.7%	98.9%	89.8%	69.6%	Wieacker-Wolff syndrome, 314580;Wieacker-Wolff syndrome, female-restricted, 301041
ZMPSTE24	100%	100%	100%	100%	99.5%	Mandibuloacral dysplasia with type B lipodystrophy, 608612;Restrictive dermopathy 1, 275210

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 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 4.2.0

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