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FETAL AKINESIA PANEL DG-5.2.0 (120 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	n.t.b					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	n.t.b					Arthrogryposis, distal, type 12, 620545
ADCY6	n.t.b					Lethal congenital contracture syndrome 8, 616287
ADGRG6	n.t.b					Lethal congenital contracture syndrome 9, 616503
ALG3	n.t.b					Congenital disorder of glycosylation, type Id, 601110
ASCC1	n.t.b					Spinal muscular atrophy with congenital bone fractures 2, 616867;Barrett esophagus/esophageal adenocarcinoma, 614266

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ATP1A2	n.t.b					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	n.t.b					Spinal muscular atrophy, lower extremity-predominant, 2B, autosomal dominant, 618291;Spinal muscular atrophy, lower extremity-predominant, 2A, autosomal dominant, 615290
BIN1	n.t.b					Centronuclear myopathy 2, 255200
BLTP1	n.t.b					Alkuraya-Kucinskas syndrome, 617822
CACNA1E	n.t.b					Developmental and epileptic encephalopathy 69, 618285
CACNA1S	n.t.b					{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

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CHAT	n.t.b					Myasthenic syndrome, congenital, 6, presynaptic, 254210
CHRNA1	n.t.b					Myasthenic syndrome, congenital, 1B, fast-channel, 608930;Myasthenic syndrome, congenital, 1A, slow-channel, 601462;Multiple pterygium syndrome, lethal type, 253290
CHRNB1	n.t.b					?Myasthenic syndrome, congenital, 2C, associated with acetylcholine receptor deficiency, 616314;Myasthenic syndrome, congenital, 2A, slow-channel, 616313
CHRND	n.t.b					?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	n.t.b					Myasthenic syndrome, congenital, 4A, slow-channel, 605809;Myasthenic syndrome, congenital, 4C, associated with acetylcholine receptor deficiency, 608931;Myasthenic syndrome, congenital, 4B, fast-channel, 616324

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CHRNG	n.t.b					Multiple pterygium syndrome, lethal type, 253290;Escobar syndrome, 265000
CHST14	n.t.b					Ehlers-Danlos syndrome, musculocontractural type 1, 601776
CNTNAP1	n.t.b					Lethal congenital contracture syndrome 7, 616286;Hypomyelinating neuropathy, congenital, 3, 618186
COL12A1	n.t.b					Bethlem myopathy 2, 616471;?Ullrich congenital muscular dystrophy 2, 616470
COL6A1	n.t.b					Ullrich congenital muscular dystrophy 1A, 254090;Bethlem myopathy 1A, 158810
COL6A2	n.t.b					?Myosclerosis, congenital, 255600;Ullrich congenital muscular dystrophy 1B, 620727;Bethlem myopathy 1B, 620725
COL6A3	n.t.b					Bethlem myopathy 1C, 620726;Ullrich congenital muscular dystrophy 1C, 620728;Dystonia 27, 616411
COX15	n.t.b					Mitochondrial complex IV deficiency, nuclear type 6, 615119

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CRPPA	n.t.b					Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 7, 616052;Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 7, 614643
DHCR24	n.t.b					Desmosterolosis, 602398
DHCR7	n.t.b					Smith-Lemli-Opitz syndrome, 270400
DNM2	n.t.b					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	n.t.b					Fetal akinesia deformation sequence 3, 618389;Myasthenic syndrome, congenital, 10, 254300
DPAGT1	n.t.b					Myasthenic syndrome, congenital, 13, with tubular aggregates, 614750;Congenital disorder of glycosylation, type Ij, 608093
ECEL1	n.t.b					Arthrogryposis, distal, type 5D, 615065
EGR2	n.t.b					Dejerine-Sottas disease, 145900;Charcot-Marie-Tooth disease, type 1D, 607678;Hypomyelinating neuropathy, congenital, 1, 605253

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ERBB3	n.t.b					?Lethal congenital contractural syndrome 2, 607598;{?Erythroleukemia, familial, susceptibility to}, 133180;Visceral neuropathy, familial, 1, autosomal recessive, 243180
ERCC5	n.t.b					Xeroderma pigmentosum, group G, 278780;Cerebrooculofacioskeletal syndrome 3, 616570;Xeroderma pigmentosum, group G/Cockayne syndrome, 278780
ERCC6	n.t.b					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
EXOSC3	n.t.b					Pontocerebellar hypoplasia, type 1B, 614678
FBN2	n.t.b					Macular degeneration, early-onset, 616118;Contractural arachnodactyly, congenital, 121050

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FHL1	n.t.b					Myopathy, X-linked, with postural muscle atrophy, 300696;Emery-Dreifuss muscular dystrophy 6, X-linked, 300696;?Uruguay faciocardiomusculoskeletal syndrome, 300280;Scapuloperoneal myopathy, X-linked dominant, 300695;Reducing body myopathy, X-linked 1b, with late childhood or adult onset, 300718;Reducing body myopathy, X-linked 1a, severe, infantile or early childhood onset, 300717
FKBP10	n.t.b					Osteogenesis imperfecta, type XI, 610968;Bruck syndrome 1, 259450
FKRP	n.t.b					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

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FKTN	n.t.b					Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 4, 611588;Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 4, 253800;Cardiomyopathy, dilated, 1X, 611615;Muscular dystrophy-dystroglycanopathy (congenital without impaired intellectual development), type B, 4, 613152
FLNC	n.t.b					Cardiomyopathy, familial dilated, 1PP, 617047;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	n.t.b					Proliferative vasculopathy and hydranencephaly-hydrocephaly syndrome, 225790

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GBA1	n.t.b					{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	n.t.b					Glycogen storage disease IV, 232500;Polyglucosan body disease, adult form, 263570
GLDN	n.t.b					Lethal congenital contracture syndrome 11, 617194
GLE1	n.t.b					Lethal congenital contracture syndrome 1, 253310;Congenital arthrogryposis with anterior horn cell disease, 611890
GMPPB	n.t.b					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

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IGHMBP2	n.t.b					Charcot-Marie-Tooth disease, axonal, type 2S, 616155;Neuronopathy, distal hereditary motor, autosomal recessive 1, 604320
KAT6B	n.t.b					SBBYSS syndrome, 603736;Genitopatellar syndrome, 606170
KIDINS220	n.t.b					Spastic paraplegia, intellectual disability, nystagmus, and obesity, 617296;Ventriculomegaly and arthrogryposis, 619501
KIF5C	n.t.b					Cortical dysplasia, complex, with other brain malformations 2, 615282
KLHL40	n.t.b					Nemaline myopathy 8, autosomal recessive, 615348
KLHL41	n.t.b					Nemaline myopathy 9, 615731
LGI4	n.t.b					Arthrogryposis multiplex congenita 1, neurogenic, with myelin defect, 617468

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LMNA	n.t.b					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	n.t.b					Nemaline myopathy 10, 616165
MAGEL2	n.t.b					Schaaf-Yang syndrome, 615547
MEGF10	n.t.b					Congenital myopathy 10A, severe variant, 614399;Congenital myopathy 10B, mild variant, 620249

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MPZ	n.t.b					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	n.t.b					Myopathy, centronuclear, X-linked, 310400
MTOR	n.t.b					Focal cortical dysplasia, type II, somatic, 607341;Smith-Kingsmore syndrome, 616638
MUSK	n.t.b					Fetal akinesia deformation sequence 1, 208150;Myasthenic syndrome, congenital, 9, associated with acetylcholine receptor deficiency, 616325
MYBPC1	n.t.b					Congenital myopathy 16, 618524;Lethal congenital contracture syndrome 4, 614915;Arthrogryposis, distal, type 1B, 614335
MYCN	n.t.b					Feingold syndrome 1, 164280;Megalencephaly-polydactyly syndrome, 620748
MYH2	n.t.b					Congenital myopathy 6 with ophthalmoplegia, 605637

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MYH3	n.t.b					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
MYH7	n.t.b					Laing distal myopathy, 160500;Cardiomyopathy, hypertrophic, 1, 192600;Left ventricular noncompaction 5, 613426;Cardiomyopathy, dilated, 1S, 613426;Congenital myopathy 7B, myosin storage, autosomal recessive, 255160;Congenital myopathy 7A, myosin storage, autosomal dominant, 608358
MYH8	n.t.b					Carney complex variant, 608837;Trismus-pseudocamptodactyly syndrome, 158300
MYL11	n.t.b					Arthrogryposis, distal, type 1C, 619110
MYMK	n.t.b					Carey-Fineman-Ziter syndrome, 254940
MYOD1	n.t.b					Congenital myopathy 17, 618975

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NALCN	n.t.b					Congenital contractures of the limbs and face, hypotonia, and developmental delay, 616266;Hypotonia, infantile, with psychomotor retardation and characteristic facies 1, 615419
NEB	n.t.b					Nemaline myopathy 2, autosomal recessive, 256030;Arthrogryposis multiplex congenita 6, 619334
NEK9	n.t.b					?Arthrogryposis, Perthes disease, and upward gaze palsy, 614262;Nevus comedonicus, somatic, 617025;Lethal congenital contracture syndrome 10, 617022
NUP88	n.t.b					Fetal akinesia deformation sequence 4, 618393
PHGDH	n.t.b					Neu-Laxova syndrome 1, 256520;Phosphoglycerate dehydrogenase deficiency, 601815
PIEZO2	n.t.b					Arthrogryposis, distal, type 5, 108145;Arthrogryposis, distal, with impaired proprioception and touch, 617146;Arthrogryposis, distal, type 3, 114300;?Marden-Walker syndrome, 248700
PIP5K1C	n.t.b					Lethal congenital contractural syndrome 3, 611369;Neurodevelopmental disorder with intellectual, visual, and language impairment, 621635

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PLOD1	n.t.b					Ehlers-Danlos syndrome, kyphoscoliotic type, 1, 225400
PLOD2	n.t.b					Bruck syndrome 2, 609220
PMM2	n.t.b					Congenital disorder of glycosylation, type Ia, 212065
PPP3CA	n.t.b					Arthrogryposis, cleft palate, craniosynostosis, and impaired intellectual development, 618265;Developmental and epileptic encephalopathy 91, 617711
PSAT1	n.t.b					Neu-Laxova syndrome 2, 616038;Phosphoserine aminotransferase deficiency, 610992
PTRH2	n.t.b					Infantile-onset multisystem neurologic, endocrine, and pancreatic disease, 616263
RAPSN	n.t.b					Fetal akinesia deformation sequence 2, 618388;Myasthenic syndrome, congenital, 11, associated with acetylcholine receptor deficiency, 616326
RIPK4	n.t.b					CHAND syndrome, 214350;Popliteal pterygium syndrome, Bartsocas-Papas type 1, 263650

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RYR1	n.t.b					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	n.t.b					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	n.t.b					Arthrogryposis multiplex congenita 4, neurogenic, with agenesis of the corpus callosum, 618766
SELENON	n.t.b					Congenital myopathy 3 with rigid spine, 602771
SLC35A3	n.t.b					Arthrogryposis, impaired intellectual development, and seizures, 615553
SLC5A7	n.t.b					Neuronopathy, distal hereditary motor, autosomal dominant 7, 158580;Myasthenic syndrome, congenital, 20, presynaptic, 617143

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SLC6A9	n.t.b					Glycine encephalopathy with normal serum glycine, 617301;{Scoliosis, isolated, susceptibility to, 6}, 621428
SMPD4	n.t.b					Neurodevelopmental disorder with microcephaly, arthrogryposis, and structural brain anomalies, 618622
SRPK3	n.t.b					Intellectual developmental disorder, X-linked 114, 301134
STAC3	n.t.b					Congenital myopathy 13, 255995
SYNE1	n.t.b					Arthrogryposis multiplex congenita 3, myogenic type, 618484;Emery-Dreifuss muscular dystrophy 4, autosomal dominant, 612998;Spinocerebellar ataxia, autosomal recessive 8, 610743
TBCD	n.t.b					Encephalopathy, progressive, early-onset, with brain atrophy and thin corpus callosum, 617193
TNNI2	n.t.b					Arthrogryposis, distal, type 2B1, 601680
TNNT1	n.t.b					Nemaline myopathy 5C, autosomal dominant, 620389;Nemaline myopathy 5A, autosomal recessive, severe infantile, 605355;Nemaline myopathy 5B, autosomal recessive, childhood-onset, 620386

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TNNT3	n.t.b					Arthrogryposis, distal, type 2B2, 618435
TOR1A	n.t.b					{Dystonia-1, modifier of};Arthrogryposis multiplex congenita 5, 618947;Dystonia-1, torsion, 128100
TPM2	n.t.b					Arthrogryposis, distal, type 2B4, 108120;Arthrogryposis, distal, type 1A, 108120;Congenital myopathy 23, 609285
TPM3	n.t.b					Congenital myopathy 4A, autosomal dominant, 255310;Congenital myopathy 4B, autosomal recessive, 609284
TRIP4	n.t.b					?Muscular dystrophy, congenital, Davignon-Chauveau type, 617066;Spinal muscular atrophy with congenital bone fractures 1, 616866

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TRPV4	n.t.b					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
TSEN54	n.t.b					Pontocerebellar hypoplasia type 2A, 277470;Pontocerebellar hypoplasia type 4, 225753;?Pontocerebellar hypoplasia type 5, 610204
TTN	n.t.b					Muscular dystrophy, limb-girdle, autosomal recessive 10, 608807;Congenital myopathy 5 with cardiomyopathy, 611705;Tibial muscular dystrophy, tardive, 600334;Cardiomyopathy, dilated, 1G, 604145;?Cardiomyopathy, familial hypertrophic, 9, 613765;Myopathy myofibrillar, 9, with early respiratory failure, 603689

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TUBA1A	n.t.b					Cortical dysplasia, complex, with other brain malformations 16, 611603
TUBB2B	n.t.b					Cortical dysplasia, complex, with other brain malformations 7, 610031
UBA1	n.t.b					Spinal muscular atrophy, X-linked 2, infantile, 301830;VEXAS syndrome, somatic, 301054
VAMP1	n.t.b					Myasthenic syndrome, congenital, 25, 618323;Spastic ataxia 1, autosomal dominant, 108600
VIPAS39	n.t.b					Arthrogryposis, renal dysfunction, and cholestasis 2, 613404
VPS33B	n.t.b					Keratoderma-ichthyosis-deafness syndrome, autosomal recessive, 620009;Cholestasis, progressive familial intrahepatic, 12, 620010;Arthrogryposis, renal dysfunction, and cholestasis 1, 208085
VRK1	n.t.b					Pontocerebellar hypoplasia type 1A, 607596;Neuronopathy, distal hereditary motor, autosomal recessive 10, 620542
WDR62	n.t.b					Microcephaly 2, primary, autosomal recessive, with or without cortical malformations, 604317

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ZC4H2	n.t.b					Wieacker-Wolff syndrome, 314580;Wieacker-Wolff syndrome, female-restricted, 301041
ZMPSTE24	n.t.b					Mandibuloacral dysplasia with type B lipodystrophy, 608612;Restrictive dermopathy 1, 275210

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