AKI GENE PANEL DG 3.1.0 (91 genes)

Releasedate: 23-03-2021

Gene	Agilent V5 covered >10x	Agilent V5 covered >20x	TWIST covered >10x	TWIST covered >20x	Associated Phenotype description and OMIM disease ID
ACTA1	99,6	92,3	100	100	Myopathy, actin, congenital, with cores, 161800 Nemaline myopathy 3, autosomal dominant or recessive, 161800 Myopathy, congenital, with fiber-type disproportion 1, 255310 Myopathy, actin, congenital, with excess of thin myofilaments, 161800 ?Myopathy, scapulohumeroperoneal, 616852
ADCY6	100	100	100	100	Lethal congenital contracture syndrome 8, 616287
ADGRG6	99,9	99	100	100	Lethal congenital contracture syndrome 9, 616503
ALG3	100	99,7	100	100	Congenital disorder of glycosylation, type Id, 601110
ASCC1	93,4	90,3	87,1	87,1	Barrett esophagus/esophageal adenocarcinoma, 614266 Spinal muscular atrophy with congenital bone fractures 2, 616867
BICD2	100	99,7	100	100	Spinal muscular atrophy, lower extremity-predominant, 2B, autosomal dominant, 618291 Spinal muscular atrophy, lower extremity-predominant, 2A, autosomal dominant, 615290
BIN1	99,6	95,7	100	100	Centronuclear myopathy 2, 255200
CACNA1S	100	99,9	100	100	{Malignant hyperthermia susceptibility 5}, 601887 {Thyrotoxic periodic paralysis, susceptibility to, 1}, 188580 {Malignant hyperthermia, susceptibility to, 5}, 601887 Hypokalemic periodic paralysis, type 1, 170400
CHAT	93,5	85,7	100	100	Myasthenic syndrome, congenital, 6, presynaptic, 254210
CHRNA1	100	99,2	100	100	Multiple pterygium syndrome, lethal type, 253290 Myasthenic syndrome, congenital, 1A, slow-channel, 601462 Myasthenic syndrome, congenital, 1B, fast-channel, 608930
CHRNB1	100	99,4	100	100	?Myasthenic syndrome, congenital, 2C, associated with acetylcholine receptor deficiency, 616314 Myasthenic syndrome, congenital, 2A, slow-channel, 616313
CHRND	99,7	97,9	100	100	?Myasthenic syndrome, congenital, 3A, slow-channel, 616321 Myasthenic syndrome, congenital, 3B, fast-channel, 616322 Multiple pterygium syndrome, lethal type, 253290 ?Myasthenic syndrome, congenital, 3C, associated with acetylcholine receptor deficiency, 616323
CHRNE	100	100	100	100	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	Escobar syndrome, 265000 Multiple pterygium syndrome, lethal type, 253290
CHST14	99,9	98,9	100	100	Ehlers-Danlos syndrome, musculocontractural type 1, 601776
CNTNAP1	100	99,8	100	100	Lethal congenital contracture syndrome 7, 616286 Hypomyelinating neuropathy, congenital, 3, 618186
COL6A1	100	99,4	100	100	Ullrich congenital muscular dystrophy 1, 254090 Bethlem myopathy 1, 158810
COL6A2	100	99,8	100	100	Bethlem myopathy 1, 158810 Ullrich congenital muscular dystrophy 1, 254090 ?Myosclerosis, congenital, 255600
COL6A3	100	99,8	100	100	Bethlem myopathy 1, 158810 Dystonia 27, 616411 Ullrich congenital muscular dystrophy 1, 254090
COX15	99,9	98,8	100	100	Mitochondrial complex IV deficiency, nuclear type 6, 615119
CRPPA	98,5	94,8	100	99,4	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 7, 616052 Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 7, 614643
DHCR24	97,7	97,7	97,7	97,7	Desmosterolosis, 602398
DHCR7	100	100	100	100	Smith-Lemli-Opitz syndrome, 270400
DNM2	98,1	94,5	100	100	Lethal congenital contracture syndrome 5, 615368 Charcot-Marie-Tooth disease, axonal type 2M, 606482 Centronuclear myopathy 1, 160150 Charcot-Marie-Tooth disease, dominant intermediate B, 606482
DOK7	95,1	91,6	100	100	Fetal akinesia deformation sequence 3, 618389 Myasthenic syndrome, congenital, 10, 254300
ECEL1	95,4	90	100	100	Arthrogryposis, distal, type 5D, 615065
EGR2	100	100	100	100	Dejerine-Sottas disease, 145900 Hypomyelinating neuropathy, congenital, 1, 605253 Charcot-Marie-Tooth disease, type 1D, 607678
ERBB3	100	99,8	100	100	{?Erythroleukemia, familial, susceptibility to}, 133180 ?Lethal congenital contractural syndrome 2, 607598
ERCC5	100	99,7	100	100	Xeroderma pigmentosum, group G/Cockayne syndrome, 278780 Xeroderma pigmentosum, group G, 278780 Cerebrooculofacioskeletal syndrome 3, 616570
ERCC6	100	100	100	100	{Macular degeneration, age-related, susceptibility to, 5}, 613761 {Lung cancer, susceptibility to}, 211980 Cerebrooculofacioskeletal syndrome 1, 214150 Cockayne syndrome, type B, 133540 Premature ovarian failure 11, 616946

					UV-sensitive syndrome 1, 600630 De Sanctis-Cacchione syndrome, 278800
FBN2	100	99,9	100	100	Contractural arachnodactyly, congenital, 121050 Macular degeneration, early-onset, 616118
FKRP	100	100	100	99,9	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 5, 607155 Muscular dystrophy-dystroglycanopathy (congenital with or without mental retardation), type B, 5, 606612 Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 5, 613153
FLNC	100	99,6	100	100	Cardiomyopathy, familial hypertrophic, 26, 617047 Myopathy, myofibrillar, 5, 609524 Cardiomyopathy, familial restrictive 5, 617047 Myopathy, distal, 4, 614065
FLVCR2	100	100	100	100	Proliferative vasculopathy and hydranencephaly-hydrocephaly syndrome, 225790
GBA	100	100	100	100	Gaucher disease, type III, 231000 {Parkinson disease, late-onset, susceptibility to}, 168600 Gaucher disease, type IIIC, 231005 Gaucher disease, type I, 230800 Gaucher disease, perinatal lethal, 608013 Gaucher disease, type II, 230900 {Lewy body dementia, susceptibility to}, 127750
GBE1	100	99,6	100	100	Polyglucosan body disease, adult form, 263570 Glycogen storage disease IV, 232500
GLDN	94,6	91	100	100	Lethal congenital contracture syndrome 11, 617194
GLE1	100	100	100	100	Congenital arthrogryposis with anterior horn cell disease, 611890 Lethal congenital contracture syndrome 1, 253310
GMPPB	100	100	100	100	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 14, 615350 Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 14, 615351 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 14, 615352
IGHMBP2	98,8	95,1	100	100	Neuronopathy, distal hereditary motor, type VI, 604320 Charcot-Marie-Tooth disease, axonal, type 2S, 616155
KIAA1109	99,8	99,2	100	100	Alkuraya-Kucinskas syndrome, 617822
KIF5C	99,9	98,8	99,8	99,8	Cortical dysplasia, complex, with other brain malformations 2, 615282
KLHL40	100	100	100	100	Nemaline myopathy 8, autosomal recessive, 615348
KLHL41	100	99,9	100	100	Nemaline myopathy 9, 615731
LGI4	99,9	97,9	100	100	Arthrogryposis multiplex congenita 1, neurogenic, with myelin defect, 617468
LMNA	97,4	91,9	100	100	Muscular dystrophy, congenital, 613205 Lipodystrophy, familial partial, type 2, 151660 Charcot-Marie-Tooth disease, type 2B1, 605588 Cardiomyopathy, dilated, 1A, 115200

					Heart-hand syndrome, Slovenian type, 610140 Hutchinson-Gilford progeria, 176670 Restrictive dermopathy, lethal, 275210 Mandibuloacral dysplasia, 248370 Emery-Dreifuss muscular dystrophy 2, autosomal dominant, 181350 Emery-Dreifuss muscular dystrophy 3, autosomal recessive, 616516 Malouf syndrome, 212112
LMOD3	100	99,7	100	100	Nemaline myopathy 10, 616165
MEGF10	100	100	100	100	Myopathy, areflexia, respiratory distress, and dysphagia, early-onset, mild variant, 614399 Myopathy, areflexia, respiratory distress, and dysphagia, early-onset, 614399
MPZ	87,9	84,1	100	100	Charcot-Marie-Tooth disease, type 2J, 607736 Charcot-Marie-Tooth disease, type 1B, 118200 Dejerine-Sottas disease, 145900 Hypomyelinating neuropathy, congenital, 2, 618184 Charcot-Marie-Tooth disease, dominant intermediate D, 607791 Roussy-Levy syndrome, 180800 Charcot-Marie-Tooth disease, type 2I, 607677
MTM1	99	93,3	100	100	Myotubular myopathy, X-linked, 310400
MUSK	100	99,9	100	100	Fetal akinesia deformation sequence 1, 208150 Myasthenic syndrome, congenital, 9, associated with acetylcholine receptor deficiency, 616325
MYBPC1	99,9	99,5	100	100	Arthrogryposis, distal, type 1B, 614335 Myopathy, congenital, with tremor, 618524 Lethal congenital contracture syndrome 4, 614915
MYCN	100	99,9	99,3	96,7	Feingold syndrome 1, 164280
MYH3	99,9	99	100	100	Contractures, pterygia, and spondylocarpotarsal fusion syndrome 1B, 618469 Arthrogryposis, distal, type 2B3 (Sheldon-Hall), 618436 Arthrogryposis, distal, type 2A (Freeman-Sheldon), 193700 Contractures, pterygia, and spondylocarpostarsal fusion syndrome 1A, 178110
MYH8	100	99,6	100	100	Carney complex variant, 608837 Trismus-pseudocamptodactyly syndrome, 158300
NEB	83	82,6	99,9	99,9	Nemaline myopathy 2, autosomal recessive, 256030
NEK9	100	99,6	100	100	Lethal congenital contracture syndrome 10, 617022 Nevus comedonicus, somatic, 617025 ?Arthrogryposis, Perthes disease, and upward gaze palsy, 614262
NUP88	100	100	100	100	Fetal akinesia deformation sequence 4, 618393
PHGDH	99,9	98,8	100	100	Neu-Laxova syndrome 1, 256520 Phosphoglycerate dehydrogenase deficiency, 601815

PIEZO2	100	99,5	100	100	Arthrogryposis, distal, with impaired proprioception and touch, 617146 Arthrogryposis, distal, type 5, 108145 ?Marden-Walker syndrome, 248700 Arthrogryposis, distal, type 3, 114300
PIP5K1C	98	95,8	99,9	99,8	Lethal congenital contractural syndrome 3, 611369
PLOD1	100	98,4	100	100	Ehlers-Danlos syndrome, kyphoscoliotic type, 1, 225400
PSAT1	95,3	81,6	100	100	Neu-Laxova syndrome 2, 616038 ?Phosphoserine aminotransferase deficiency, 610992
PTRH2	100	100	100	100	Infantile-onset multisystem neurologic, endocrine, and pancreatic disease, 616263
RAPSN	100	99,7	100	100	Fetal akinesia deformation sequence 2, 618388 Myasthenic syndrome, congenital, 11, associated with acetylcholine receptor deficiency, 616326
RIPK4	100	99,9	100	100	Popliteal pterygium syndrome, Bartsocas-Papas type, 263650 CHAND syndrome, 214350
RYR1	96,9	93,9	99,4	99	Central core disease, 117000 King-Denborough syndrome, 145600 {Malignant hyperthermia susceptibility 1}, 145600 Minicore myopathy with external ophthalmoplegia, 255320 Neuromuscular disease, congenital, with uniform type 1 fiber, 117000
SCN4A	100	99,6	100	100	Hyperkalemic periodic paralysis, type 2, 170500 Paramyotonia congenita, 168300 Myotonia congenita, atypical, acetazolamide-responsive, 608390 Myasthenic syndrome, congenital, 16, 614198 Hypokalemic periodic paralysis, type 2, 613345
SELENON	84,5	84	87,7	85,1	Muscular dystrophy, rigid spine, 1, 602771 Myopathy, congenital, with fiber-type disproportion, 255310
SLC5A7	100	99,9	100	100	Neuronopathy, distal hereditary motor, type VIIA, 158580 Myasthenic syndrome, congenital, 20, presynaptic, 617143
SLC6A9	100	100	100	100	Glycine encephalopathy with normal serum glycine, 617301
SMN1	99,5	94,7	94,6	94,6	Spinal muscular atrophy-2, 253550 Spinal muscular atrophy-3, 253400 Spinal muscular atrophy-1, 253300 Spinal muscular atrophy-4, 271150
SMPD4	99,4	94,2	100	100	Neurodevelopmental disorder with microcephaly, arthrogryposis, and structural brain anomalies, 618622
STAC3	100	100	100	100	Myopathy, congenital, Baily-Bloch, 255995
SYNE1	98,2	97,8	98,8	98,8	Arthrogryposis multiplex congenita 3, myogenic type, 618484 Spinocerebellar ataxia, autosomal recessive 8, 610743 Emery-Dreifuss muscular dystrophy 4, autosomal dominant, 612998
TBCD	96,2	94,4	100	100	Encephalopathy, progressive, early-onset, with brain atrophy and thin corpus callosum, 617193

TNNI2	100	99,7	100	100	Arthrogryposis, distal, type 2B1, 601680
TNNT3	100	99,7	100	100	Arthrogryposis, distal, type 2B2, 618435
TPM2	100	100	100	100	Nemaline myopathy 4, autosomal dominant, 609285 Arthrogryposis, distal, type 2B4, 108120 Arthrogryposis, distal, type 1A, 108120 CAP myopathy 2, 609285
TPM3	89,2	87,2	100	100	CAP myopathy 1, 609284 Nemaline myopathy 1, autosomal dominant or recessive, 609284 Myopathy, congenital, with fiber-type disproportion, 255310
TRIP4	100	99,1	100	100	Spinal muscular atrophy with congenital bone fractures 1, 616866 ?Muscular dystrophy, congenital, Davignon-Chauveau type, 617066
TRPV4	100	99,9	100	100	Spondylometaphyseal dysplasia, Kozlowski type, 184252 Parastremmatic dwarfism, 168400 SED, Maroteaux type, 184095 Neuronopathy, distal hereditary motor, type VIII, 600175 [Sodium serum level QTL 1], 613508 Scapuloperoneal spinal muscular atrophy, 181405 Metatropic dysplasia, 156530 Digital arthropathy-brachydactyly, familial, 606835 Hereditary motor and sensory neuropathy, type IIc, 606071 Brachyolmia type 3, 113500 ?Avascular necrosis of femoral head, primary, 2, 617383
TTN	98,6	98,1	100	100	Myopathy, myofibrillar, 9, with early respiratory failure, 603689 Cardiomyopathy, familial hypertrophic, 9, 613765 Muscular dystrophy, limb-girdle, autosomal recessive 10, 608807 Cardiomyopathy, dilated, 1G, 604145 Tibial muscular dystrophy, tardive, 600334 Salih myopathy, 611705
TUBA1A	99,9	97	100	100	Lissencephaly 3, 611603
TUBB2B	100	99,5	100	100	Cortical dysplasia, complex, with other brain malformations 7, 610031
UBA1	99,4	98,2	99,8	99	VEXAS syndrome, somatic, 301054 Spinal muscular atrophy, X-linked 2, infantile, 301830
VIPAS39	100	100	100	100	Arthrogryposis, renal dysfunction, and cholestasis 2, 613404
VPS33B	100	100	100	100	Arthrogryposis, renal dysfunction, and cholestasis 1, 208085
WDR62	100	99,5	100	100	Microcephaly 2, primary, autosomal recessive, with or without cortical malformations, 604317
ZC4H2	100	99	100	100	Wieacker-Wolff syndrome, 314580 Wieacker-Wolff syndrome, female-restricted, 301041

ZMPSTE24	100	99,9	100	100	Restrictive dermopathy, lethal, 275210 Mandibuloacral dysplasia with type B lipodystrophy, 608612

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.
Agilent V5 is the default chemistry, and used for all exome analyses apart from the (in-house) TURBO/RAPID WES route.
TWIST is the chemistry used for (in-house) TURBO/RAPID WES analysis.
Covered 10x describes the percentage of a gene's coding sequence that is covered at least 10x.
Covered 20x describes the percentage of a gene's coding sequence that is covered at least 20x.
Genes with coverage denoting NC are non-protein coding genes.
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 23rd , 2021.
This list is accurate for panel version DG 3.1.0
Ad 1. "No OMIM Disease ID" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors