AKI GENE PANEL DG 3.2.0 (95 genes)

Releasedate: 16-09-2021

Gene	*Agilent V5 covered >10x*	*Agilent V5 covered >20x*	*TWIST covered >10x*	*TWIST covered >20x*	*Associated Phenotype Description and OMIM disease ID*
ACTA1	98,2	89,5	100	100	?Myopathy, scapulohumeroperoneal, 616852 Nemaline myopathy 3, autosomal dominant or recessive, 161800 Myopathy, actin, congenital, with excess of thin myofilaments, 161800 Myopathy, actin, congenital, with cores, 161800 Myopathy, congenital, with fiber-type disproportion 1, 255310
ADCY6	100	100	100	100	Lethal congenital contracture syndrome 8, 616287
ADGRG6	99,7	98,7	100	100	Lethal congenital contracture syndrome 9, 616503
ALG3	100	99,5	100	100	Congenital disorder of glycosylation, type Id, 601110
ASCC1	92,5	89,4	87,1	87	Spinal muscular atrophy with congenital bone fractures 2, 616867 Barrett esophagus/esophageal adenocarcinoma, 614266
BICD2	99,9	99,1	100	100	Spinal muscular atrophy, lower extremity-predominant, 2B, autosomal dominant, 618291 Spinal muscular atrophy, lower extremity-predominant, 2A, autosomal dominant, 615290
BIN1	99,7	96	100	100	Centronuclear myopathy 2, 255200
CACNA1S	100	99,8	100	100	Hypokalemic periodic paralysis, type 1, 170400
CHAT	93,1	85,1	100	99,9	Myasthenic syndrome, congenital, 6, presynaptic, 254210
CHRNA1	100	99,6	100	100	Myasthenic syndrome, congenital, 1B, fast-channel, 608930 Myasthenic syndrome, congenital, 1A, slow-channel, 601462 Multiple pterygium syndrome, lethal type, 253290
CHRNB1	100	99,5	100	100	?Myasthenic syndrome, congenital, 2C, associated with acetylcholine receptor deficiency, 616314 Myasthenic syndrome, congenital, 2A, slow-channel, 616313
CHRND	99,4	97,4	100	100	?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	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	Multiple pterygium syndrome, lethal type, 253290 Escobar syndrome, 265000
CHST14	99,9	98,8	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	99,7	100	100	Bethlem myopathy 1, 158810 Ullrich congenital muscular dystrophy 1, 254090
COL6A2	100	99,8	100	100	Bethlem myopathy 1, 158810 ?Myosclerosis, congenital, 255600 Ullrich congenital muscular dystrophy 1, 254090
COL6A3	100	99,7	100	100	Ullrich congenital muscular dystrophy 1, 254090 Dystonia 27, 616411 Bethlem myopathy 1, 158810
COX15	99,9	97,8	100	100	Mitochondrial complex IV deficiency, nuclear type 6, 615119
CRPPA	98,4	94,7	100	99,8	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	98,6	93,9	100	100	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	94,9	92	100	100	Fetal akinesia deformation sequence 3, 618389 Myasthenic syndrome, congenital, 10, 254300
ECEL1	95,9	91,8	100	100	Arthrogryposis, distal, type 5D, 615065
EGR2	100	100	100	100	Dejerine-Sottas disease, 145900 Charcot-Marie-Tooth disease, type 1D, 607678 Hypomyelinating neuropathy, congenital, 1, 605253
ERBB3	100	99,3	100	100	?Lethal congenital contractural syndrome 2, 607598 Visceral neuropathy, familial, 1, autosomal recessive, 243180
ERCC5	99,9	99	100	100	Xeroderma pigmentosum, group G, 278780 Cerebrooculofacioskeletal syndrome 3, 616570 Xeroderma pigmentosum, group G/Cockayne syndrome, 278780
ERCC6	100	100	100	100	UV-sensitive syndrome 1, 600630 Cerebrooculofacioskeletal syndrome 1, 214150 Cockayne syndrome, type B, 133540

					De Sanctis-Cacchione syndrome, 278800 Premature ovarian failure 11, 616946
FBN2	100	99,8	100	100	Macular degeneration, early-onset, 616118 Contractural arachnodactyly, congenital, 121050
FKRP	100	100	100	100	Muscular dystrophy-dystroglycanopathy (congenital with or without mental retardation), 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	99,4	100	100	Cardiomyopathy, familial hypertrophic, 26, 617047 Cardiomyopathy, familial restrictive 5, 617047 Myopathy, distal, 4, 614065 Myopathy, myofibrillar, 5, 609524
FLVCR2	100	100	100	100	Proliferative vasculopathy and hydranencephaly-hydrocephaly syndrome, 225790
GBA	100	100	100	100	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
GBE1	99,9	99,7	100	100	Glycogen storage disease IV, 232500 Polyglucosan body disease, adult form, 263570
GLDN	95,8	91,5	100	100	Lethal congenital contracture syndrome 11, 617194
GLE1	100	99,9	100	100	Lethal congenital contracture syndrome 1, 253310 Congenital arthrogryposis with anterior horn cell disease, 611890
GMPPB	100	100	100	100	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 14, 615352 Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 14, 615351 Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 14, 615350
IGHMBP2	99,3	96,9	100	100	Neuronopathy, distal hereditary motor, type VI, 604320 Charcot-Marie-Tooth disease, axonal, type 2S, 616155
KIAA1109	99,8	99	100	100	Alkuraya-Kucinskas syndrome, 617822
KIF5C	99,6	97	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	99,9	99,4	100	100	Nemaline myopathy 9, 615731
LGI4	99,7	97,9	100	100	Arthrogryposis multiplex congenita 1, neurogenic, with myelin defect, 617468
LMNA	96,1	90,6	100	100	Mandibuloacral dysplasia, 248370 Heart-hand syndrome, Slovenian type, 610140 Cardiomyopathy, dilated, 1A, 115200

					Restrictive dermopathy, lethal, 275210 Emery-Dreifuss muscular dystrophy 3, autosomal recessive, 616516 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	99,8	99	100	100	Nemaline myopathy 10, 616165
MAGEL2	94,1	89,1	100	100	Schaaf-Yang syndrome, 615547
MEGF10	100	99,9	100	100	Myopathy, areflexia, respiratory distress, and dysphagia, early-onset, mild variant, 614399 Myopathy, areflexia, respiratory distress, and dysphagia, early-onset, 614399
MPZ	85,6	81,9	100	100	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	98,7	92	100	99,7	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	99,8	99,1	100	99,9	Myopathy, congenital, with tremor, 618524 Lethal congenital contracture syndrome 4, 614915 Arthrogryposis, distal, type 1B, 614335
MYCN	100	100	99,2	96,2	Feingold syndrome 1, 164280
MYH3	99,9	98,4	100	100	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	99,2	100	100	Carney complex variant, 608837 Trismus-pseudocamptodactyly syndrome, 158300
MYLPF	100	100	100	100	Arthrogryposis, distal, type 1C, 619110
NEB	82,9	82,5	99,9	99,8	Nemaline myopathy 2, autosomal recessive, 256030 Arthrogryposis multiplex congenita 6, 619334

NEK9	99,9	99	100	100	?Arthrogryposis, Perthes disease, and upward gaze palsy, 614262 Nevus comedonicus, somatic, 617025 Lethal congenital contracture syndrome 10, 617022
NUP88	99,8	99,8	100	100	Fetal akinesia deformation sequence 4, 618393
PHGDH	99,9	98,2	100	100	Neu-Laxova syndrome 1, 256520 Phosphoglycerate dehydrogenase deficiency, 601815
PIEZO2	99,8	99,2	100	100	Arthrogryposis, distal, type 5, 108145 Arthrogryposis, distal, with impaired proprioception and touch, 617146 Arthrogryposis, distal, type 3, 114300 ?Marden-Walker syndrome, 248700
PIP5K1C	99,2	96,7	99,9	99,2	Lethal congenital contractural syndrome 3, 611369
PLOD1	100	98,2	100	100	Ehlers-Danlos syndrome, kyphoscoliotic type, 1, 225400
PSAT1	92	75,1	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	99,6	100	100	Fetal akinesia deformation sequence 2, 618388 Myasthenic syndrome, congenital, 11, associated with acetylcholine receptor deficiency, 616326
RIPK4	100	99,9	100	100	CHAND syndrome, 214350 Popliteal pterygium syndrome, Bartsocas-Papas type 1, 263650
RYR1	97,1	94	99,4	99	Neuromuscular disease, congenital, with uniform type 1 fiber, 117000 Central core disease, 117000 King-Denborough syndrome, 145600 Minicore myopathy with external ophthalmoplegia, 255320
SCN4A	99,9	99,4	100	100	Paramyotonia congenita, 168300 Hypokalemic periodic paralysis, type 2, 613345 Myotonia congenita, atypical, acetazolamide-responsive, 608390 Myasthenic syndrome, congenital, 16, 614198 Hyperkalemic periodic paralysis, type 2, 170500
SCYL2	96,2	88	100	100	Arthrogryposis multiplex congenita 4, neurogenic, with agenesis of the corpus callosum, 618766
SELENON	84,3	84	87,8	85,1	Myopathy, congenital, with fiber-type disproportion, 255310 Muscular dystrophy, rigid spine, 1, 602771
SLC5A7	100	100	100	100	Neuronopathy, distal hereditary motor, type VIIA, 158580 Myasthenic syndrome, congenital, 20, presynaptic, 617143
SLC6A9	100	99,6	100	100	Glycine encephalopathy with normal serum glycine, 617301
SMN1	99,7	96,1	94,6	94,6	Spinal muscular atrophy-2, 253550 Spinal muscular atrophy-4, 271150

					Spinal muscular atrophy-3, 253400 Spinal muscular atrophy-1, 253300
SMPD4	99,6	95	100	100	Neurodevelopmental disorder with microcephaly, arthrogryposis, and structural brain anomalies, 618622
STAC3	100	100	100	100	Myopathy, congenital, Baily-Bloch, 255995
SYNE1	98,1	97,5	98,8	98,8	Arthrogryposis multiplex congenita 3, myogenic type, 618484 Emery-Dreifuss muscular dystrophy 4, autosomal dominant, 612998 Spinocerebellar ataxia, autosomal recessive 8, 610743
TBCD	95,5	93,3	100	100	Encephalopathy, progressive, early-onset, with brain atrophy and thin corpus callosum, 617193
TNNI2	100	99,9	100	100	Arthrogryposis, distal, type 2B1, 601680
TNNT3	100	99,6	100	100	Arthrogryposis, distal, type 2B2, 618435
TOR1A	91,3	91,3	91,7	91,3	Arthrogryposis multiplex congenita 5, 618947 Dystonia-1, torsion, 128100
TPM2	100	99,8	100	99,9	Arthrogryposis, distal, type 2B4, 108120 Arthrogryposis, distal, type 1A, 108120 Nemaline myopathy 4, autosomal dominant, 609285 CAP myopathy 2, 609285
TPM3	87,7	84,3	100	100	CAP myopathy 1, 609284 Myopathy, congenital, with fiber-type disproportion, 255310 Nemaline myopathy 1, autosomal dominant or recessive, 609284
TRIP4	99,8	99	100	100	?Muscular dystrophy, congenital, Davignon-Chauveau type, 617066 Spinal muscular atrophy with congenital bone fractures 1, 616866
TRPV4	100	99,9	100	100	Spondylometaphyseal dysplasia, Kozlowski type, 184252 Digital arthropathy-brachydactyly, familial, 606835 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 Neuronopathy, distal hereditary motor, type VIII, 600175 Parastremmatic dwarfism, 168400 Brachyolmia type 3, 113500
TTN	98,5	98	100	100	Muscular dystrophy, limb-girdle, autosomal recessive 10, 608807 Cardiomyopathy, familial hypertrophic, 9, 613765 Tibial muscular dystrophy, tardive, 600334 Salih myopathy, 611705

					Cardiomyopathy, dilated, 1G, 604145 Myopathy, myofibrillar, 9, with early respiratory failure, 603689
TUBA1A	99,5	93,2	100	100	Lissencephaly 3, 611603
TUBB2B	100	99,7	100	100	Cortical dysplasia, complex, with other brain malformations 7, 610031
UBA1	99,2	97,3	99,9	99,3	Spinal muscular atrophy, X-linked 2, infantile, 301830 VEXAS syndrome, somatic, 301054
VIPAS39	100	100	100	100	Arthrogryposis, renal dysfunction, and cholestasis 2, 613404
VPS33B	100	99,9	100	100	Arthrogryposis, renal dysfunction, and cholestasis 1, 208085
WDR62	100	99,9	100	100	Microcephaly 2, primary, autosomal recessive, with or without cortical malformations, 604317
ZC4H2	100	98,1	100	100	Wieacker-Wolff syndrome, 314580 Wieacker-Wolff syndrome, female-restricted, 301041
ZMPSTE24	99,6	99,4	100	99,9	Mandibuloacral dysplasia with type B lipodystrophy, 608612 Restrictive dermopathy, lethal, 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.
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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 : September 16th , 2021.
This list is accurate for panel version DG 3.2.0
Ad 1. "No OMIM Disease ID" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors