AKI GENE PANEL DG 2.18 (86 genes)

Releasedate: 20-04-2020

Gene	Agilent V5 covered >10x	Agilent V5 covered > 20x	TWIST covered >10x	TWIST covered >20x	Associated Phenotype description and OMIM disease ID
ACTA1	99,60%	92,30%	100,00%	100,00%	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	100,00%	100,00%	100,00%	100,00%	?Lethal congenital contracture syndrome 8, 616287
ADGRG6	99,90%	99,00%	100,00%	100,00%	Lethal congenital contracture syndrome 9, 616503
ALG3	100,00%	99,70%	100,00%	100,00%	Congenital disorder of glycosylation, type Id, 601110
ASCC1	97,70%	94,40%	91,10%	91,10%	Barrett esophagus/esophageal adenocarcinoma, 614266 Spinal muscular atrophy with congenital bone fractures 2, 616867
BICD2	100,00%	99,70%	100,00%	100,00%	Spinal muscular atrophy, lower extremity-predominant, 2B, autosomal dominant, 618291 Spinal muscular atrophy, lower extremity-predominant, 2A, autosomal dominant, 615290
BIN1	99,60%	95,70%	100,00%	100,00%	Centronuclear myopathy 2, 255200
CHAT	93,50%	85,70%	100,00%	100,00%	Myasthenic syndrome, congenital, 6, presynaptic, 254210
CHRNA1	94,70%	94,00%	100,00%	100,00%	Myasthenic syndrome, congenital, 1A, slow-channel, 601462 Myasthenic syndrome, congenital, 1B, fast-channel, 608930 Multiple pterygium syndrome, lethal type, 253290
CHRND	99,70%	97,90%	100,00%	100,00%	?Myasthenic syndrome, congenital, 3C, associated with acetylcholine receptor deficiency, 616323 ?Myasthenic syndrome, congenital, 3A, slow-channel, 616321 Myasthenic syndrome, congenital, 3B, fast-channel, 616322 Multiple pterygium syndrome, lethal type, 253290
CHRNE	100,00%	100,00%	100,00%	100,00%	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	100,00%	100,00%	100,00%	100,00%	Escobar syndrome, 265000 Multiple pterygium syndrome, lethal type, 253290
CHST14	99,90%	98,90%	100,00%	100,00%	Ehlers-Danlos syndrome, musculocontractural type 1, 601776
CNTNAP1	100,00%	99,80%	100,00%	100,00%	Lethal congenital contracture syndrome 7, 616286 Hypomyelinating neuropathy, congenital, 3, 618186

COL6A1	100,00%	99,40%	100,00%	100,00%	Ullrich congenital muscular dystrophy 1, 254090 Bethlem myopathy 1, 158810
COL6A2	100,00%	99,80%	100,00%	100,00%	Bethlem myopathy 1, 158810 ?Myosclerosis, congenital, 255600 Ullrich congenital muscular dystrophy 1, 254090
COL6A3	100,00%	99,80%	100,00%	100,00%	Bethlem myopathy 1, 158810 Dystonia 27, 616411 Ullrich congenital muscular dystrophy 1, 254090
COX15	99,90%	98,80%	100,00%	100,00%	Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency 2, 615119 Leigh syndrome due to cytochrome c oxidase deficiency, 256000
DHCR24	100,00%	100,00%	100,00%	100,00%	Desmosterolosis, 602398
DHCR7	100,00%	100,00%	100,00%	100,00%	Smith-Lemli-Opitz syndrome, 270400
DNM2	98,10%	94,50%	100,00%	100,00%	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	94,10%	91,30%	100,00%	100,00%	Myasthenic syndrome, congenital, 10, 254300 Fetal akinesia deformation sequence 3, 618389
ECEL1	95,40%	90,00%	100,00%	100,00%	Arthrogryposis, distal, type 5D, 615065
EGR2	100,00%	100,00%	100,00%	100,00%	Dejerine-Sottas disease, 145900 Hypomyelinating neuropathy, congenital, 1, 605253 Charcot-Marie-Tooth disease, type 1D, 607678
ERBB3	100,00%	99,80%	100,00%	100,00%	?Lethal congenital contractural syndrome 2, 607598
ERCC5	100,00%	99,60%	100,00%	100,00%	Xeroderma pigmentosum, group G/Cockayne syndrome, 278780 Xeroderma pigmentosum, group G, 278780 Cerebrooculofacioskeletal syndrome 3, 616570
ERCC6	100,00%	100,00%	100,00%	100,00%	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	100,00%	99,90%	100,00%	100,00%	Contractural arachnodactyly, congenital, 121050 Macular degeneration, early-onset, 616118
FKRP	100,00%	100,00%	100,00%	99,90%	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
FLVCR2	100,00%	100,00%	100,00%	100,00%	Proliferative vasculopathy and hydranencephaly-hydrocephaly syndrome, 225790

GBA	100,00%	100,00%	100,00%	100,00%	Gaucher disease, type III, 231000 Gaucher disease, type IIIC, 231005 Gaucher disease, type I, 230800 Gaucher disease, perinatal lethal, 608013 Gaucher disease, type II, 230900
GBE1	100,00%	99,60%	100,00%	100,00%	Polyglucosan body disease, adult form, 263570 Glycogen storage disease IV, 232500
GLDN	94,60%	91,00%	100,00%	100,00%	Lethal congenital contracture syndrome 11, 617194
GLE1	100,00%	100,00%	100,00%	100,00%	Lethal congenital contracture syndrome 1, 253310 Congenital arthrogryposis with anterior horn cell disease, 611890
GMPPB	100,00%	100,00%	100,00%	100,00%	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	98,80%	95,10%	100,00%	100,00%	Neuronopathy, distal hereditary motor, type VI, 604320 Charcot-Marie-Tooth disease, axonal, type 2S, 616155
ISPD	98,50%	94,80%	100,00%	99,40%	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 7, 616052 Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 7, 614643
KIAA1109	99,80%	99,20%	100,00%	100,00%	Alkuraya-Kucinskas syndrome, 617822
KIF5C	99,90%	98,80%	99,80%	99,80%	Cortical dysplasia, complex, with other brain malformations 2, 615282
KLHL40	100,00%	100,00%	100,00%	100,00%	Nemaline myopathy 8, autosomal recessive, 615348
KLHL41	100,00%	99,90%	100,00%	100,00%	Nemaline myopathy 9, 615731
LGI4	99,90%	97,90%	100,00%	100,00%	Arthrogryposis multiplex congenita, neurogenic, with myelin defect, 617468
LMNA	97,40%	91,90%	100,00%	100,00%	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	100,00%	99,70%	100,00%	100,00%	Nemaline myopathy 10, 616165
MEGF10	100,00%	100,00%	100,00%	100,00%	Myopathy, areflexia, respiratory distress, and dysphagia, early-onset, mild variant, 614399 Myopathy, areflexia, respiratory distress, and dysphagia, early-onset, 614399
MPZ	100,00%	100,00%	100,00%	100,00%	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	99,00%	93,30%	100,00%	100,00%	Myotubular myopathy, X-linked, 310400
MUSK	100,00%	99,90%	100,00%	100,00%	Fetal akinesia deformation sequence 1, 208150 Myasthenic syndrome, congenital, 9, associated with acetylcholine receptor deficiency, 616325
MYBPC1	99,90%	99,50%	100,00%	100,00%	Arthrogryposis, distal, type 1B, 614335 Myopathy, congenital, with tremor, 618524 Lethal congenital contracture syndrome 4, 614915
MYCN	100,00%	99,90%	99,30%	96,70%	Feingold syndrome 1, 164280
MYH3	99,90%	99,00%	100,00%	100,00%	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	100,00%	99,60%	100,00%	100,00%	Carney complex variant, 608837 Trismus-pseudocamptodactyly syndrome, 158300
NEB	83,00%	82,60%	99,90%	99,90%	Nemaline myopathy 2, autosomal recessive, 256030
NEK9	100,00%	99,60%	100,00%	100,00%	Lethal congenital contracture syndrome 10, 617022 Nevus comedonicus, somatic, 617025 ?Arthrogryposis, Perthes disease, and upward gaze palsy, 614262
NUP88	100,00%	100,00%	100,00%	100,00%	Fetal akinesia deformation sequence 4, 618393
PHGDH	99,90%	98,80%	100,00%	100,00%	Neu-Laxova syndrome 1, 256520 Phosphoglycerate dehydrogenase deficiency, 601815
PIEZO2	100,00%	99,50%	100,00%	100,00%	Arthrogryposis, distal, with impaired proprioception and touch, 617146 Arthrogryposis, distal, type 5, 108145 ?Marden-Walker syndrome, 248700 Arthrogryposis, distal, type 3, 114300
PIP5K1C	98,00%	95,80%	99,90%	99,80%	Lethal congenital contractural syndrome 3, 611369
PLOD1	100,00%	98,40%	100,00%	100,00%	Ehlers-Danlos syndrome, kyphoscoliotic type, 1, 225400
PSAT1	95,30%	81,60%	100,00%	100,00%	Neu-Laxova syndrome 2, 616038 ?Phosphoserine aminotransferase deficiency, 610992
RAPSN	100,00%	99,70%	100,00%	100,00%	Fetal akinesia deformation sequence 2, 618388 Myasthenic syndrome, congenital, 11, associated with acetylcholine receptor deficiency, 616326
RIPK4	100,00%	99,90%	100,00%	100,00%	Popliteal pterygium syndrome, Bartsocas-Papas type, 263650 CHAND syndrome, 214350

RYR1	96,90%	93,90%	99,40%	99,00%	Central core disease, 117000 King-Denborough syndrome, 145600 Minicore myopathy with external ophthalmoplegia, 255320 Neuromuscular disease, congenital, with uniform type 1 fiber, 117000
SCN4A	100,00%	99,60%	100,00%	100,00%	Paramyotonia congenita, 168300 Hyperkalemic periodic paralysis, type 2, 170500 Myotonia congenita, atypical, acetazolamide-responsive, 608390 Myasthenic syndrome, congenital, 16, 614198 Hypokalemic periodic paralysis, type 2, 613345
SELENON	84,50%	84,00%	87,70%	85,10%	Muscular dystrophy, rigid spine, 1, 602771 Myopathy, congenital, with fiber-type disproportion, 255310
SLC5A7	100,00%	99,90%	100,00%	100,00%	Neuronopathy, distal hereditary motor, type VIIA, 158580 Myasthenic syndrome, congenital, 20, presynaptic, 617143
SLC6A9	100,00%	100,00%	100,00%	100,00%	Glycine encephalopathy with normal serum glycine, 617301
SMN1	99,50%	94,70%	94,60%	94,60%	Spinal muscular atrophy-2, 253550 Spinal muscular atrophy-3, 253400 Spinal muscular atrophy-1, 253300 Spinal muscular atrophy-4, 271150
SMPD4	99,40%	94,20%	100,00%	100,00%	Neurodevelopmental disorder with microcephaly, arthrogryposis, and structural brain anomalies, 618622
SYNE1	98,30%	98,00%	98,80%	98,80%	Spinocerebellar ataxia, autosomal recessive 8, 610743 Arthrogryposis multiplex congenita, myogenic type, 618484 Emery-Dreifuss muscular dystrophy 4, autosomal dominant, 612998
TBCD	96,20%	94,40%	100,00%	100,00%	Encephalopathy, progressive, early-onset, with brain atrophy and thin corpus callosum, 617193
TNNI2	100,00%	99,70%	100,00%	100,00%	Arthrogryposis, distal, type 2B1, 601680
TNNT3	100,00%	99,70%	100,00%	100,00%	Arthrogryposis, distal, type 2B2, 618435
TPM2	100,00%	100,00%	84,00%	83,70%	Nemaline myopathy 4, autosomal dominant, 609285 Arthrogryposis, distal, type 2B4, 108120 Arthrogryposis, distal, type 1A, 108120 CAP myopathy 2, 609285
TPM3	89,20%	87,20%	100,00%	100,00%	CAP myopathy 1, 609284 Nemaline myopathy 1, autosomal dominant or recessive, 609284 Myopathy, congenital, with fiber-type disproportion, 255310
TRIP4	100,00%	99,10%	100,00%	100,00%	Spinal muscular atrophy with congenital bone fractures 1, 616866 ?Muscular dystrophy, congenital, Davignon-Chauveau type, 617066
TRPV4	100,00%	99,90%	100,00%	100,00%	Spondylometaphyseal dysplasia, Kozlowski type, 184252 Parastremmatic dwarfism, 168400 SED, Maroteaux type, 184095 Neuronopathy, distal hereditary motor, type VIII, 600175

					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	98,60%	98,10%	100,00%	100,00%	Cardiomyopathy, dilated, 1G, 604145 Tibial muscular dystrophy, tardive, 600334 Salih myopathy, 611705 Cardiomyopathy, familial hypertrophic, 9, 613765 Myopathy, myofibrillar, 9, with early respiratory failure, 603689 Muscular dystrophy, limb-girdle, autosomal recessive 10, 608807
TUBA1A	99,90%	97,00%	100,00%	100,00%	Lissencephaly 3, 611603
TUBB2B	100,00%	99,50%	100,00%	100,00%	Cortical dysplasia, complex, with other brain malformations 7, 610031
UBA1	99,40%	98,20%	99,80%	99,00%	Spinal muscular atrophy, X-linked 2, infantile, 301830
VIPAS39	100,00%	100,00%	100,00%	100,00%	Arthrogryposis, renal dysfunction, and cholestasis 2, 613404
VPS33B	100,00%	100,00%	100,00%	100,00%	Arthrogryposis, renal dysfunction, and cholestasis 1, 208085
WDR62	100,00%	99,50%	100,00%	100,00%	Microcephaly 2, primary, autosomal recessive, with or without cortical malformations, 604317
ZC4H2	100,00%	99,00%	100,00%	100,00%	Wieacker-Wolff syndrome, 314580 Wieacker-Wolff syndrome, female-restricted, 301041
ZMPSTE24	100,00%	99,90%	100,00%	100,00%	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-DNA coding genes.
non-DNA coding genes are covered, but as coverage statistics are based on DNA coding regions, statistics could not be generated.
OMIM release used for OMIM disease identifiers and descriptions : April 20th , 2020.
This list is accurate for panel version DG 2.18
Ad 1. "No OMIM Disease ID" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors