AKI GENE PANEL DG 2.16 (79 genes)

Releasedate: 07-06-2019

Gene	*Median coverage*	*% covered > 10x*	*% covered > 20x*	*Associated Phenotype description and OMIM disease ID*
ACTA1	95,3	99.8%	97.9%	?Myopathy, scapulohumeroperoneal, 616852 Myopathy, actin, congenital, with cores, 161800 Myopathy, actin, congenital, with excess of thin myofilaments, 161800 Myopathy, congenital, with fiber-type disproportion 1, 255310 Nemaline myopathy 3, autosomal dominant or recessive, 161800
ADCY6	162,8	100.0%	100.0%	?Lethal congenital contracture syndrome 8, 616287
ADGRG6	135,7	99.8%	98.7%	Lethal congenital contracture syndrome 9, 616503
ALG3	106,5	100.0%	99.9%	Congenital disorder of glycosylation, type Id, 601110
ASCC1	125,4	95.7%	92.0%	?Spinal muscular atrophy with congenital bone fractures 2, 616867 Barrett esophagus/esophageal adenocarcinoma, 614266
BIN1	113,4	99.9%	98.4%	Centronuclear myopathy 2, 255200
CHAT	117,1	95.4%	86.9%	Myasthenic syndrome, congenital, 6, presynaptic, 254210
CHRNA1	92,6	94.6%	93.3%	Multiple pterygium syndrome, lethal type, 253290 Myasthenic syndrome, congenital, 1A, slow-channel, 601462 Myasthenic syndrome, congenital, 1B, fast-channel, 608930
CHRND	140,4	99.8%	98.0%	?Myasthenic syndrome, congenital, 3A, slow-channel, 616321 ?Myasthenic syndrome, congenital, 3C, associated with acetylcholine receptor deficiency, 616323 Multiple pterygium syndrome, lethal type, 253290 Myasthenic syndrome, congenital, 3B, fast-channel, 616322
CHRNE	167,8	100.0%	100.0%	Myasthenic syndrome, congenital, 4A, slow-channel, 605809 Myasthenic syndrome, congenital, 4B, fast-channel, 616324 Myasthenic syndrome, congenital, 4C, associated with acetylcholine receptor deficiency, 608931
CHRNG	141,3	100.0%	100.0%	Escobar syndrome, 265000 Multiple pterygium syndrome, lethal type, 253290
CHST14	160,6	99.9%	98.9%	Ehlers-Danlos syndrome, musculocontractural type 1, 601776
CNTNAP1	157,2	99.9%	99.1%	Hypomyelinating neuropathy, congenital, 3, 618186 Lethal congenital contracture syndrome 7, 616286
COL6A1	158,8	100.0%	99.8%	Bethlem myopathy 1, 158810 Ullrich congenital muscular dystrophy 1, 254090
COL6A2	175,3	100.0%	99.8%	?Myosclerosis, congenital, 255600

				Bethlem myopathy 1, 158810 Ullrich congenital muscular dystrophy 1, 254090
COL6A3	154	100.0%	99.8%	Bethlem myopathy 1, 158810 Dystonia 27, 616411 Ullrich congenital muscular dystrophy 1, 254090
COX15	87,7	99.9%	98.3%	Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency 2, 615119 Leigh syndrome due to cytochrome c oxidase deficiency, 256000
DHCR24	155,8	100.0%	99.9%	Desmosterolosis, 602398
DHCR7	144,9	100.0%	100.0%	Smith-Lemli-Opitz syndrome, 270400
DNM2	123,9	99.7%	96.7%	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	135,1	94.0%	93.3%	?Fetal akinesia deformation sequence 3, 618389 Myasthenic syndrome, congenital, 10, 254300
ECEL1	107,1	100.0%	97.4%	Arthrogryposis, distal, type 5D, 615065
EGR2	130	100.0%	100.0%	Charcot-Marie-Tooth disease, type 1D, 607678 Dejerine-Sottas disease, 145900 Hypomyelinating neuropathy, congenital, 1, 605253
ERBB3	113,3	99.9%	99.2%	?Lethal congenital contractural syndrome 2, 607598 {?Erythroleukemia, familial, susceptibility to}, 133180
ERCC5	126,3	99.9%	99.5%	Cerebrooculofacioskeletal syndrome 3, 616570 Xeroderma pigmentosum, group G, 278780 Xeroderma pigmentosum, group G/Cockayne syndrome, 278780
ERCC6	158,2	100.0%	99.9%	Cerebrooculofacioskeletal syndrome 1, 214150 Cockayne syndrome, type B, 133540 De Sanctis-Cacchione syndrome, 278800 Premature ovarian failure 11, 616946 UV-sensitive syndrome 1, 600630 {Lung cancer, susceptibility to}, 211980 {Macular degeneration, age-related, susceptibility to, 5}, 613761
FBN2	142,2	100.0%	99.8%	Contractural arachnodactyly, congenital, 121050 Macular degeneration, early-onset, 616118
FKRP	153,3	100.0%	100.0%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 5, 613153 Muscular dystrophy-dystroglycanopathy (congenital with or without mental retardation), type B, 5, 606612 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 5, 607155
FLVCR2	124,8	100.0%	100.0%	Proliferative vasculopathy and hydranencephaly-hydrocephaly syndrome, 225790
GBA	169,8	100.0%	100.0%	Gaucher disease, perinatal lethal, 608013

				Gaucher disease, type I, 230800 Gaucher disease, type II, 230900 Gaucher disease, type III, 231000 Gaucher disease, type IIIC, 231005 {Lewy body dementia, susceptibility to}, 127750 {Parkinson disease, late-onset, susceptibility to}, 168600
GBE1	157,4	99.9%	99.7%	Glycogen storage disease IV, 232500 Polyglucosan body disease, adult form, 263570
GLDN	106,6	99.8%	96.9%	Lethal congenital contracture syndrome 11, 617194
GLE1	97,1	100.0%	99.9%	Congenital arthrogryposis with anterior horn cell disease, 611890 Lethal congenital contracture syndrome 1, 253310
GMPPB	211,8	100.0%	100.0%	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	108,3	99.6%	97.4%	Charcot-Marie-Tooth disease, axonal, type 2S, 616155 Neuronopathy, distal hereditary motor, type VI, 604320
ISPD	NC	NC	NC	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 7, 614643 Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 7, 616052
KIF5C	109,9	99.9%	99.0%	Cortical dysplasia, complex, with other brain malformations 2, 615282
KLHL40	130,6	100.0%	100.0%	Nemaline myopathy 8, autosomal recessive, 615348
KLHL41	172,8	100.0%	99.8%	Nemaline myopathy 9, 615731
LGI4	99,9	99.4%	96.7%	Arthrogryposis multiplex congenita, neurogenic, with myelin defect, 617468
LMNA	104,7	97.7%	91.9%	Cardiomyopathy, dilated, 1A, 115200 Charcot-Marie-Tooth disease, type 2B1, 605588 Emery-Dreifuss muscular dystrophy 2, autosomal dominant, 181350 Emery-Dreifuss muscular dystrophy 3, autosomal recessive, 616516 Heart-hand syndrome, Slovenian type, 610140 Hutchinson-Gilford progeria, 176670 Lipodystrophy, familial partial, type 2, 151660 Malouf syndrome, 212112 Mandibuloacral dysplasia, 248370 Muscular dystrophy, congenital, 613205 Restrictive dermopathy, lethal, 275210
LMOD3	128,6	100.0%	99.8%	Nemaline myopathy 10, 616165
MEGF10	125,9	100.0%	99.8%	Myopathy, areflexia, respiratory distress, and dysphagia, early-onset, 614399 Myopathy, areflexia, respiratory distress, and dysphagia, early-onset, mild variant, 614399
MPZ	125	100.0%	98.9%	Charcot-Marie-Tooth disease, dominant intermediate D, 607791 Charcot-Marie-Tooth disease, type 1B, 118200

				Charcot-Marie-Tooth disease, type 2I, 607677 Charcot-Marie-Tooth disease, type 2J, 607736 Dejerine-Sottas disease, 145900 Hypomyelinating neuropathy, congenital, 2, 618184 Roussy-Levy syndrome, 180800
MTM1	79,1	98.7%	91.9%	Myotubular myopathy, X-linked, 310400
MUSK	131,5	100.0%	100.0%	Fetal akinesia deformation sequence 1, 208150 Myasthenic syndrome, congenital, 9, associated with acetylcholine receptor deficiency, 616325
MYBPC1	127,8	99.9%	99.2%	Arthrogryposis, distal, type 1B, 614335 Lethal congenital contracture syndrome 4, 614915
MYCN	173,8	100.0%	100.0%	Feingold syndrome 1, 164280
MYH3	94,1	99.9%	98.3%	Arthrogryposis, distal, type 2A (Freeman-Sheldon), 193700 Arthrogryposis, distal, type 2B (Sheldon-Hall), 601680 Arthrogryposis, distal, type 8, 178110
MYH8	115,4	100.0%	99.4%	Carney complex variant, 608837 Trismus-pseudocamptodactyly syndrome, 158300
NEB	100,1	83.0%	82.4%	Nemaline myopathy 2, autosomal recessive, 256030
NEK9	118,9	99.8%	98.2%	?Arthrogryposis, Perthes disease, and upward gaze palsy, 614262 Lethal congenital contracture syndrome 10, 617022 Nevus comedonicus, somatic, 617025
PHGDH	106,6	100.0%	99.3%	Neu-Laxova syndrome 1, 256520 Phosphoglycerate dehydrogenase deficiency, 601815
PIEZO2	104,2	99.9%	99.2%	?Marden-Walker syndrome, 248700 Arthrogryposis, distal, type 3, 114300 Arthrogryposis, distal, type 5, 108145 Arthrogryposis, distal, with impaired proprioception and touch, 617146
PIP5K1C	136,6	99.8%	97.6%	Lethal congenital contractural syndrome 3, 611369
PLOD1	131,9	99.8%	97.3%	Ehlers-Danlos syndrome, kyphoscoliotic type, 1, 225400
PSAT1	42,8	90.3%	72.5%	?Phosphoserine aminotransferase deficiency, 610992 Neu-Laxova syndrome 2, 616038
RAPSN	149	99.8%	97.7%	Fetal akinesia deformation sequence 2, 618388 Myasthenic syndrome, congenital, 11, associated with acetylcholine receptor deficiency, 616326
RIPK4	167,5	100.0%	100.0%	CHAND syndrome, 214350 Popliteal pterygium syndrome, Bartsocas-Papas type, 263650
RYR1	117,1	98.7%	95.7%	Central core disease, 117000 King-Denborough syndrome, 145600 Minicore myopathy with external ophthalmoplegia, 255320 Neuromuscular disease, congenital, with uniform type 1 fiber, 117000

				{Malignant hyperthermia susceptibility 1}, 145600
SCN4A	167,9	99.8%	99.3%	Hyperkalemic periodic paralysis, type 2, 170500 Hypokalemic periodic paralysis, type 2, 613345 Myasthenic syndrome, congenital, 16, 614198 Myotonia congenita, atypical, acetazolamide-responsive, 608390 Paramyotonia congenita, 168300
SELENON	131	84.9%	83.9%	Muscular dystrophy, rigid spine, 1, 602771 Myopathy, congenital, with fiber-type disproportion, 255310
SLC5A7	100,2	100.0%	99.9%	Myasthenic syndrome, congenital, 20, presynaptic, 617143 Neuronopathy, distal hereditary motor, type VIIA, 158580
SLC6A9	148,8	100.0%	100.0%	Glycine encephalopathy with normal serum glycine, 617301
SMN1	97,6	99.8%	96.9%	Spinal muscular atrophy-1, 253300 Spinal muscular atrophy-2, 253550 Spinal muscular atrophy-3, 253400 Spinal muscular atrophy-4, 271150
TBCD	136,2	98.2%	94.3%	Encephalopathy, progressive, early-onset, with brain atrophy and thin corpus callosum, 617193
TNNI2	150,5	100.0%	100.0%	Arthrogryposis multiplex congenita, distal, type 2B, 601680
TNNT3	146,3	100.0%	99.8%	Arthrogryposis, distal, type 2B, 601680
TPM2	105,2	100.0%	99.7%	Arthrogryposis multiplex congenita, distal, type 1, 108120 Arthrogryposis, distal, type 2B, 601680 CAP myopathy 2, 609285 Nemaline myopathy 4, autosomal dominant, 609285
TPM3	74,3	89.5%	88.0%	CAP myopathy 1, 609284 Myopathy, congenital, with fiber-type disproportion, 255310 Nemaline myopathy 1, autosomal dominant or recessive, 609284
TRIP4	103,3	99.8%	98.5%	?Muscular dystrophy, congenital, Davignon-Chauveau type, 617066 Spinal muscular atrophy with congenital bone fractures 1, 616866
TRPV4	138,4	100.0%	99.8%	?Avascular necrosis of femoral head, primary, 2, 617383 Brachyolmia type 3, 113500 Digital arthropathy-brachydactyly, familial, 606835 Hereditary motor and sensory neuropathy, type IIc, 606071 Metatropic dysplasia, 156530 Parastremmatic dwarfism, 168400 Scapuloperoneal spinal muscular atrophy, 181405 SED, Maroteaux type, 184095 Spinal muscular atrophy, distal, congenital nonprogressive, 600175 Spondylometaphyseal dysplasia, Kozlowski type, 184252 [Sodium serum level QTL 1], 613508

TTN	163	98.6%	98.1%	Cardiomyopathy, dilated, 1G, 604145 Cardiomyopathy, familial hypertrophic, 9, 613765 Muscular dystrophy, limb-girdle, autosomal recessive 10, 608807 Myopathy, myofibrillar, 9, with early respiratory failure, 603689 Salih myopathy, 611705 Tibial muscular dystrophy, tardive, 600334
UBA1	130,5	99.6%	98.1%	Spinal muscular atrophy, X-linked 2, infantile, 301830
VIPAS39	114,7	100.0%	99.9%	Arthrogryposis, renal dysfunction, and cholestasis 2, 613404
VPS33B	107,2	100.0%	99.9%	Arthrogryposis, renal dysfunction, and cholestasis 1, 208085
WDR62	152,6	100.0%	99.8%	Microcephaly 2, primary, autosomal recessive, with or without cortical malformations, 604317
ZC4H2	72,4	99.8%	95.9%	Wieacker-Wolff syndrome, 314580
ZMPSTE24	128,7	100.0%	99.6%	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.
Median Coverage describes the average number of reads seen across 50 exomes.
% 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 Median Coverage and % Covered 10x/20x denoting NC are non-coding genes for which coverage statistics could not be generated.
OMIM release used for OMIM disease identifiers and descriptions : May 8th , 2019.
This list is accurate for panel version DG 2.16
Ad 1. "No OMIM phenotype" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors