AKI GENE PANEL DG 2.17 (81 genes)

Releasedate: 06-12-2019

Gene	*Median Coverage*	*% covered > 10x*	*% covered > 20x*	*Associated Phenotype description and OMIM disease ID*
ACTA1	106.8	99.9%	98.5%	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	180.3	100.0%	100.0%	?Lethal congenital contracture syndrome 8, 616287
ADGRG6	134.6	99.8%	98.8%	Lethal congenital contracture syndrome 9, 616503
ALG3	117.9	100.0%	100.0%	Congenital disorder of glycosylation, type Id, 601110
ASCC1	122.4	95.7%	92.1%	Barrett esophagus/esophageal adenocarcinoma, 614266 ?Spinal muscular atrophy with congenital bone fractures 2, 616867
BIN1	125.8	100.0%	99.5%	Centronuclear myopathy 2, 255200
CHAT	125.5	97.1%	89.5%	Myasthenic syndrome, congenital, 6, presynaptic, 254210
CHRNA1	96.7	94.5%	93.4%	Myasthenic syndrome, congenital, 1A, slow-channel, 601462 Myasthenic syndrome, congenital, 1B, fast-channel, 608930 Multiple pterygium syndrome, lethal type, 253290
CHRND	154.8	99.9%	98.3%	?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	186.2	100.0%	100.0%	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	153.9	100.0%	100.0%	Escobar syndrome, 265000 Multiple pterygium syndrome, lethal type, 253290
CHST14	180.8	100.0%	99.5%	Ehlers-Danlos syndrome, musculocontractural type 1, 601776
CNTNAP1	170.0	100.0%	99.4%	Lethal congenital contracture syndrome 7, 616286 Hypomyelinating neuropathy, congenital, 3, 618186
COL6A1	178.1	100.0%	100.0%	Ullrich congenital muscular dystrophy 1, 254090 Bethlem myopathy 1, 158810
COL6A2	195.3	100.0%	99.9%	Bethlem myopathy 1, 158810

				?Myosclerosis, congenital, 255600 Ullrich congenital muscular dystrophy 1, 254090
COL6A3	163.9	100.0%	99.8%	Bethlem myopathy 1, 158810 Dystonia 27, 616411 Ullrich congenital muscular dystrophy 1, 254090
COX15	90.4	99.9%	98.7%	Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency 2, 615119 Leigh syndrome due to cytochrome c oxidase deficiency, 256000
DHCR24	170.7	100.0%	99.9%	Desmosterolosis, 602398
DHCR7	158.7	100.0%	100.0%	Smith-Lemli-Opitz syndrome, 270400
DNM2	134.2	99.8%	97.7%	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	156.2	94.4%	93.5%	Myasthenic syndrome, congenital, 10, 254300 ?Fetal akinesia deformation sequence 3, 618389
ECEL1	121.5	100.0%	99.0%	Arthrogryposis, distal, type 5D, 615065
EGR2	140.2	100.0%	100.0%	Dejerine-Sottas disease, 145900 Hypomyelinating neuropathy, congenital, 1, 605253 Charcot-Marie-Tooth disease, type 1D, 607678
ERBB3	120.0	100.0%	99.3%	?Lethal congenital contractural syndrome 2, 607598
ERCC5	130.9	100.0%	99.4%	Xeroderma pigmentosum, group G/Cockayne syndrome, 278780 Xeroderma pigmentosum, group G, 278780 Cerebrooculofacioskeletal syndrome 3, 616570
ERCC6	161.8	100.0%	100.0%	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	144.5	100.0%	99.9%	Contractural arachnodactyly, congenital, 121050 Macular degeneration, early-onset, 616118
FKRP	178.0	100.0%	100.0%	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	131.7	100.0%	100.0%	Proliferative vasculopathy and hydranencephaly-hydrocephaly syndrome, 225790
GBA	180.2	100.0%	100.0%	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	152.5	100.0%	99.5%	Polyglucosan body disease, adult form, 263570 Glycogen storage disease IV, 232500
GLDN	112.6	99.8%	97.6%	Lethal congenital contracture syndrome 11, 617194
GLE1	100.8	100.0%	99.9%	Lethal congenital contracture syndrome 1, 253310 Congenital arthrogryposis with anterior horn cell disease, 611890
GMPPB	233.1	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	117.7	99.9%	98.2%	Neuronopathy, distal hereditary motor, type VI, 604320 Charcot-Marie-Tooth disease, axonal, type 2S, 616155
ISPD	112.0	99.7%	97.8%	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 7, 616052 Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 7, 614643
KIF5C	113.8	100.0%	99.0%	Cortical dysplasia, complex, with other brain malformations 2, 615282
KLHL40	144.2	100.0%	100.0%	Nemaline myopathy 8, autosomal recessive, 615348
KLHL41	174.8	100.0%	99.6%	Nemaline myopathy 9, 615731
LGI4	114.5	99.5%	97.2%	Arthrogryposis multiplex congenita, neurogenic, with myelin defect, 617468
LMNA	118.2	98.3%	93.2%	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	134.2	100.0%	99.9%	Nemaline myopathy 10, 616165
MEGF10	129.9	100.0%	99.9%	Myopathy, areflexia, respiratory distress, and dysphagia, early-onset, mild variant, 614399 Myopathy, areflexia, respiratory distress, and dysphagia, early-onset, 614399
MPZ	135.9	100.0%	99.0%	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	78.4	98.4%	91.8%	Myotubular myopathy, X-linked, 310400
MUSK	135.2	100.0%	100.0%	Fetal akinesia deformation sequence 1, 208150 Myasthenic syndrome, congenital, 9, associated with acetylcholine receptor deficiency, 616325
MYBPC1	129.2	100.0%	99.4%	Arthrogryposis, distal, type 1B, 614335 Myopathy, congenital, with tremor, 618524 Lethal congenital contracture syndrome 4, 614915
MYCN	200.6	100.0%	100.0%	Feingold syndrome 1, 164280
MYH3	100.2	100.0%	99.1%	Contractures, pterygia, and variable skeletal fusions syndrome 1B, 618469 Contractures, pterygia, and variable skeletal fusions syndrome 1A, 178110 Arthrogryposis, distal, type 2B3 (Sheldon-Hall), 618436 Arthrogryposis, distal, type 2A (Freeman-Sheldon), 193700
MYH8	119.4	100.0%	99.6%	Carney complex variant, 608837 Trismus-pseudocamptodactyly syndrome, 158300
NEB	101.5	83.0%	82.5%	Nemaline myopathy 2, autosomal recessive, 256030
NEK9	123.4	99.9%	98.9%	Lethal congenital contracture syndrome 10, 617022 Nevus comedonicus, somatic, 617025 ?Arthrogryposis, Perthes disease, and upward gaze palsy, 614262
PHGDH	116.2	100.0%	99.6%	Neu-Laxova syndrome 1, 256520 Phosphoglycerate dehydrogenase deficiency, 601815
PIEZO2	106.9	99.9%	99.4%	Arthrogryposis, distal, with impaired proprioception and touch, 617146 Arthrogryposis, distal, type 5, 108145 ?Marden-Walker syndrome, 248700 Arthrogryposis, distal, type 3, 114300
PIP5K1C	151.5	99.9%	98.4%	Lethal congenital contractural syndrome 3, 611369
PLOD1	141.5	99.9%	97.9%	Ehlers-Danlos syndrome, kyphoscoliotic type, 1, 225400
PSAT1	46.2	91.6%	74.2%	Neu-Laxova syndrome 2, 616038 ?Phosphoserine aminotransferase deficiency, 610992
RAPSN	162.2	99.9%	99.2%	Fetal akinesia deformation sequence 2, 618388 Myasthenic syndrome, congenital, 11, associated with acetylcholine receptor deficiency, 616326
RIPK4	189.1	100.0%	100.0%	Popliteal pterygium syndrome, Bartsocas-Papas type, 263650 CHAND syndrome, 214350
RYR1	128.2	99.3%	96.8%	Central core disease, 117000 King-Denborough syndrome, 145600 Minicore myopathy with external ophthalmoplegia, 255320 Neuromuscular disease, congenital, with uniform type 1 fiber, 117000
SCN4A	182.1	99.9%	99.4%	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	143.9	85.3%	84.0%	Muscular dystrophy, rigid spine, 1, 602771 Myopathy, congenital, with fiber-type disproportion, 255310
SLC5A7	102.5	100.0%	99.9%	Neuronopathy, distal hereditary motor, type VIIA, 158580 Myasthenic syndrome, congenital, 20, presynaptic, 617143
SLC6A9	164.3	100.0%	100.0%	Glycine encephalopathy with normal serum glycine, 617301
SMN1	89.1	99.7%	97.7%	Spinal muscular atrophy-2, 253550 Spinal muscular atrophy-3, 253400 Spinal muscular atrophy-1, 253300 Spinal muscular atrophy-4, 271150
TBCD	145.8	98.8%	95.5%	Encephalopathy, progressive, early-onset, with brain atrophy and thin corpus callosum, 617193
TNNI2	169.0	100.0%	100.0%	Arthrogryposis, distal, type 2B1, 601680
TNNT3	160.3	100.0%	99.8%	Arthrogryposis, distal, type 2B2, 618435
TPM2	114.5	100.0%	99.8%	Nemaline myopathy 4, autosomal dominant, 609285 Arthrogryposis, distal, type 2B4, 108120 Arthrogryposis, distal, type 1A, 108120 CAP myopathy 2, 609285
TPM3	76.2	89.4%	88.3%	CAP myopathy 1, 609284 Nemaline myopathy 1, autosomal dominant or recessive, 609284 Myopathy, congenital, with fiber-type disproportion, 255310
TRIP4	105.3	100.0%	98.9%	Spinal muscular atrophy with congenital bone fractures 1, 616866 ?Muscular dystrophy, congenital, Davignon-Chauveau type, 617066
TRPV4	150.4	100.0%	100.0%	Spinal muscular atrophy, distal, congenital nonprogressive, 600175 Spondylometaphyseal dysplasia, Kozlowski type, 184252 Parastremmatic dwarfism, 168400 SED, Maroteaux type, 184095 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	165.0	98.6%	98.1%	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	82.5	99.9%	97.8%	Lissencephaly 3, 611603
TUBB2B	88.3	100.0%	99.9%	Cortical dysplasia, complex, with other brain malformations 7, 610031
UBA1	139.8	99.7%	98.4%	Spinal muscular atrophy, X-linked 2, infantile, 301830
VIPAS39	114.4	100.0%	99.9%	Arthrogryposis, renal dysfunction, and cholestasis 2, 613404
VPS33B	111.7	100.0%	100.0%	Arthrogryposis, renal dysfunction, and cholestasis 1, 208085
WDR62	166.4	100.0%	100.0%	Microcephaly 2, primary, autosomal recessive, with or without cortical malformations, 604317
ZC4H2	74.3	99.6%	95.6%	Wieacker-Wolff syndrome, 314580
ZMPSTE24	126.2	100.0%	99.7%	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.
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 : December 11 , 2019.
This list is accurate for panel version DG 2.17
Ad 1. "No OMIM phenotype" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors