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Ion AmpliSeq™ Inherited Disease Panel target gene list

Developed to match genes targeted by clinical molecular geneticists studying inherited diseases, including genes listed in the NIH Genetic Testing Registry, the Ion AmpliSeq™ Inherited Disease Panel employs over 10,000 primer pairs in just 3 tubes to amplify the exons of 328 genes. These genes are associated with over 700 inherited diseases according to NCBI ClinVar database.

Symbol	Disease	Symbol	Disease
ABCA4	Retinitis Pigmentosa	***BMPR1A***	Juvenile Polyposis Syndrome
ABCC9	Dilated Cardiomyopathy 10	***BTD***	Biotinidase Deficiency
ABCD1	X-Linked Adrenoleukodystrophy	***BTK***	Agammaglobulinemia, X-Linked, Type 1
ACADVL	Very Long Chain Acyl-Coenzyme A Dehydrogenase Deficiency	***CA4***	Retinitis Pigmentosa
ACTA2	Thoracic Aortic Aneurysms and Aortic Dissections	***CACNA1C***	Brugada Syndrome
ACTC1	Familial Hypertrophic Cardiomyopathy	***CACNB2***	Brugada Syndrome
ACTN2	Dilated Cardiomyopathy 1AA	***CALR3***	Familial Hypertrophic Cardiomyopathy
ADA	Severe Combined Immunodeficiency	***CAPN3***	Limb-Girdle Muscular Dystrophy Type 2A - Calpainopathy
AIPL1	Leber Congenital Amaurosis	***CASQ2***	Catecholaminergic Polymorphic Ventricular Tachycardia (CPVT)
AIRE	Autoimmune Polyendocrine Syndrome	***CAV3***	Familial Hypertrophic Cardiomyopathy
AKAP9	Long QT Syndrome, Autosomal Dominant	***CCDC39***	Primary Ciliary Dyskinesia
AKR1B1	Androgen Insensitivity Syndrome	***CCDC40***	Primary Ciliary Dyskinesia
ALPL	Hypophosphatasia	***CDH23***	Usher Syndrome Type 1
AMT	Glycine Encephalopathy	***CEP290***	Leber Congenital Amaurosis
ANK2	Long/Short QT Syndrome, Autosomal Dominant	***CERKL***	Retinitis Pigmentosa
APC	APC-Associated Polyposis Conditions	***CFTR***	Cystic Fibrosis
APP	Early-Onset Familial Alzheimer Disease	***CHAT***	Congenital Myasthenic Syndromes
APTX	Ataxia with Oculomotor Apraxia Type 2	***CHD7***	Charge Syndrome
ARL6	Retinitis Pigmentosa	***CHEK2***	Li-Fraumeni Syndrome
ARSA	Arylsulfatase A Deficiency	***CHM***	Choroideremia
ASL	Argininosuccinate Lyase Deficiency	***CHRNA1***	Congenital Myasthenic Syndromes
ASPA	Canavan	***CHRNB1***	Congenital Myasthenic Syndromes
ATL1	Spastic Paraplegia-3A	***CHRND***	Congenital Myasthenic Syndromes
ATM	Ataxia-Telangiectasia	***CHRNE***	Congenital Myasthenic Syndromes
ATP2A2	Darier Disease	***CLCN1***	Myotonia Congenita
ATP7A	Menkes/ATP7A-Related Copper Transport Disease	***CNGB1***	Retinitis Pigmentosa
ATP7B	Wilson Disease	***COL11A1***	Stickler Syndrome, AD
ATXN1	Spinocerebellar Ataxia 1	***COL11A2***	Inherited Deafness
ATXN2	Spinocerebellar Ataxia 2	***COL1A1***	Osteogenesis Imperfecta
ATXN7	Spinocerebellar Ataxia 7	***COL1A2***	Osteogenesis Imperfecta
BAG3	Dilated Cardiomyopathy 1HH	***COL2A1***	Stickler Syndrome, AD
BCKDHA	Maple Syrup Urine Disease	***COL3A1***	Ehlers-Danlos Syndrome
BCKDHB	Maple Syrup Urine Disease	***COL4A1***	Thoracic Aortic Aneurysms and Aortic Dissections
BEST1	Retinitis Pigmentosa	***COL4A5***	Alport Syndrome

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COL5A1	Ehlers-Danlos Syndrome, Classic Type
COL5A2	Ehlers-Danlos Syndrome, Classic Type
COL7A1	Epidermolysis Bullosa Simplex
COL9A1	Stickler Syndrome
CRB1	Leber Congenital Amaurosis
CRX	Retinitis Pigmentosa
CTDP1	Congenital Cataracts, Facial Dysmorphism, and Neuropathy
CTNS	Cystinosis
CYP27A1	Cerebrotendinous Xanthomatosis
DBT	Maple Syrup Urine Disease
DCX	Double Cortex Syndrome
DES	Dilated Cardiomyopathy
DHCR7	Smith-Lemli-Opitz Syndrome
DKC1	Dyskeratosis Congenita
DLD	Maple Syrup Urine Disease
DMD	Duchenne/Becker Muscular Dystrophy
DNAH5	Primary Ciliary Dyskinesia
DNAH9	Primary Ciliary Dyskinesia
DNAH11	Primary Ciliary Dyskinesia
DNAI1	Primary Ciliary Dyskinesia
DNAI2	Primary Ciliary Dyskinesia
DNM2	Charcot-Marie-Tooth Disease Type 2B
DOK7	Congenital Myasthenic Syndromes
DSC2	Arrhythmogenic Right Ventricular Dysplasia/Cardiomyopathy
DSG2	Arrhythmogenic Right Ventricular Dysplasia/Cardiomyopathy
DSP	Arrhythmogenic Right Ventricular Dysplasia/Cardiomyopathy
DYSF	Dysferlinopathy
ELN	Supravalvular Aortic Stenosis
EMD	Emery-Dreifuss Muscular Dystrophy, X-Linked
ENG	Hereditary Hemorrhagic Telangiectasia
EXT1	Exostoses, Multiple, Type 1
EYA1	Branchiootorenal Spectrum Disorders
EYS	Retinitis Pigmentosa
F8	Hemophilia A
F9	Hemophilia B
FANCA	Fanconi Anemia
FANCC	Fanconi Anemia
FANCF	Fanconi Anemia
FANCG	Fanconi Anemia
FBN1	Marfan Syndrome
FBXO7	Parkinson Disease
FGFR1	FGFR-Related Craniosynostosis Syndromes
FGFR3	Hypochondroplasia
FMO3	Trimethylaminuria
FOXL2	Blepharophimosis-Ptosis-Epicanthus Inversus
FRG1	Facioscapulohumeral Muscular Dystrophy
FRMD7	FRMD7-Related Infantile Nystagmus
FSCN2	Retinitis Pigmentosa
FXN	Friedreich Ataxia
GAA	Pompe Disease -GSD II
GALT	Galactosemia
GATA4	Atrial Septal Defect
GBA	Gaucher Disease
GBE1	Glycogen Storage Disease Type VI
GCSH	Glycine Encephalopathy
GDF5	Brachydactyly
GJB2	Inherited Deafness, Top Genes
GJB3	Inherited Deafness, Top Genes
GJB6	Inherited Deafness, Top Genes
GLA	Fabry Disease
GLDC	Glycine Encephalopathy
GNE	Inclusion Body Myopathy 2
GNPTAB	Mucolipidosis II
GPC3	Wilms Tumor, Classical
GPD1L	Brugada Syndrome
GPR143	Ocular Albinism, X-Linked
GUCY2D	Leber Congenital Amaurosis
HBA2	Alpha-Thalassemia - Southeast Asia
HBB	Sickle Cell Disease Beta-Thalassemia
HCN4	Brugada Syndrome
HEXA	Hexosaminidase A Deficiency
HFE	HFE-Associated Hereditary Hemochromatosis
HIBCH	Beta-Hydroxyisobutyryl CoA Deacylase Deficiency (HIBCH Deficiency)
HMBS	Hydroxymethylbilane Synthase (HMBS) Deficiency
HR	Alopecia Universalis Congenita (ALUNC)
IDS	Hunter Syndrome (MPSII)
IDUA	Hurler Syndrome (MPSI)
IKBKAP	Familial Dysautonomia (HSAN III)
IL2RG	X-Linked SCIDS
IMPDH1	Leber Congenital Amaurosis
ITGB4	Epidermolysis Bullosa Simplex
JAG1	Alagille Syndrome
JUP	Arrhythmogenic Right Ventricular Dysplasia/Cardiomyopathy
KCNE1	Long QT Syndrome, Autosomal Dominant
KCNE2	Long QT Syndrome, Autosomal Dominant
KCNE3	Brugada Syndrome
KCNH2	Long QT Syndrome, Autosomal Dominant
KCNJ2	Short QT Syndrome

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KCNQ1	Long QT Syndrome, Autosomal Dominant	***NPC2***	Niemann-Pick Disease Type C2
KCNQ4	Inherited Deafness	***NR2E3***	Retinitis Pigmentosa
KIAA0196	Spastic Paraplegia 8	***NRAS***	Noonan Syndrome
KLHL7	Retinitis Pigmentosa	***NSD1***	Sotos Syndrome
KRAS	Noonan Syndrome	***NUDT19***	Retinitis Pigmentosa
KRT5	Epidermolysis Bullosa Simplex	***OCA2***	Oculocutaneous Albinism Type 2
KRT14	Epidermolysis Bullosa Simplex	***OCRL***	Lowe Syndrome
L1CAM	Spastic Paraplegia Type 1 - L1 Syndrome	***OTC***	Ornithine Transcarbamylase Deficiency
LAMB3	Epidermolysis Bullosa Simplex	***PABPN1***	Oculopharyngeal Muscular Dystrophy
LAMP2	Dilated Cardiomyopathy	***PAFAH1B1***	Lissencephaly 1
LDB3	Dilated Cardiomyopathy	***PAH***	Phenylketonuria (PKU)
LMNA	Limb-Girdle Muscular Dystrophy, Type 1B	***PAX3***	Waardenburg Syndrome, Type 1
LRAT	Retinitis Pigmentosa	***PAX6***	Aniridia
LRRK2	Parkinson Disease	***PCDH15***	Usher Syndrome Type 1
MAPRE2	Retinitis Pigmentosa	***PEX1***	Zellweger Syndrome
MAPT	Parkinson-Dementia Syndrome	***PEX3***	Peroxisome Biogenesis, Zellweger
MC1R	Oculocutaneous Albinism Type 2	***PEX5***	Neonatal Adrenoleucodystrophy
MECP2	MECP2-Rett Syndrome	***PEX10***	Peroxisome Biogenesis, Zellweger
MED12	Fryns Syndrome	***PEX13***	Peroxisome Biogenesis, Zellweger
MEN1	Multiple Endocrine Neoplasia Type 1	***PEX14***	Peroxisome Biogenesis, Zellweger
MERTK	Retinitis Pigmentosa	***PEX19***	Peroxisome Biogenesis, Zellweger
MFN2	Charcot-Marie-Tooth Neuropathy Type 2A	***PEX26***	Peroxisome Biogenesis, Zellweger
MLH1	Turcot Syndrome	***PINK1***	Parkinson Disease
MMAA	Methylmalonic Acidemia	***PKD1***	Polycystic Kidney Disease, Autosomal Dominant
MMAB	Methylmalonic Acidemia	***PKD2***	Polycystic Kidney Disease, Autosomal Recessive
MMACHC	Methylmalonic Acidemia	***PKHD1***	Polycystic Kidney Disease, Autosomal Recessive
MPZ	Charcot-Marie-Tooth Neuropathy Type 1B	***PKP2***	Arrhythmogenic Right Ventricular Dysplasia/Cardiomyopathy
MSH2	Turcot Syndrome	***PLEC***	Epidermolysis Bullosa Simplex
MTM1	X-Linked Myotubular Myopathy	***PLN***	Dilated Cardiomyopathy 1P
MUT	Methylmalonic Acidemia	***PLOD1***	Ehlers-Danlos Syndrome, Kyphoscoliotic Form
MYBPC3	Familial Hypertrophic Cardiomyopathy	***PMM2***	Congenital Disorder of Glycosylation Type 1a
MYH11	Thoracic Aortic Aneurysms and Aortic Dissections	***PMP22***	Charcot-Marie-Tooth Neuropathy Type 1A
MYH6	Familial Hypertrophic Cardiomyopathy	***POLG***	Alpers Syndrome
MYH7	Familial Hypertrophic Cardiomyopathy	***PPT1***	Ceroid Lipofuscinoses (Batten Disease)
MYL2	Familial Hypertrophic Cardiomyopathy	***PRCD***	Retinitis Pigmentosa
MYL3	Familial Hypertrophic Cardiomyopathy	***PRKAG2***	Familial Hypertrophic Cardiomyopathy
MYLK	Familial Hypertrophic Cardiomyopathy	***PROM1***	Retinitis Pigmentosa
MYO7A	Usher Syndrome Type 1	***PRPF8***	Retinitis Pigmentosa
MYOZ2	Familial Hypertrophic Cardiomyopathy	***PRPF31***	Retinitis Pigmentosa
NF1	Neurofibromatosis Type 1	***PRPH2***	Retinitis Pigmentosa
NF2	Neurofibromatosis Type 2	***PSEN1***	Early-Onset Familial Alzheimer Disease
NIPBL	Cornelia de Lange Syndrome	***PSEN2***	Early-Onset Familial Alzheimer Disease
NKX2-5	Tetralogy of Fallot	***PTCH1***	Holoprosencephaly-7 & Basal Cell Nevus Syndrome
NPC1	Niemann-Pick Disease Type C1	***PTPN11***	Noonan Syndrome

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RAF1	Noonan Syndrome	***SLC26A4***	Pendred Syndrome/Syndromic Deafness
RAG1	Severe Combined Immunodeficiency	***SMAD3***	Thoracic Aortic Aneurysms and Aortic Dissections
RAG2	Severe Combined Immunodeficiency	***SMAD4***	Juvenile Polyposis Syndrome
RAI1	Smith-Magenis Syndrome	***SNCA***	Parkinson Disease
RAPSN	Congenital Myasthenic Syndromes	***SNRNP200***	Retinitis Pigmentosa
RB1	Retinoblastoma	***SNTA1***	Long QT Syndrome, Autosomal Dominant
RDH12	Leber Congenital Amaurosis	***SOD1***	Amyotrophic Lateral Sclerosis (Lou Gehrig's Disease)
RET	Multiple Endocrine Neoplasia Type 2	***SOS1***	Noonan Syndrome
RHO	Retinitis Pigmentosa	***SOX9***	Campomelic Dysplasia
ROR2	Brachydactyly, Type B1	***SPATA7***	Retinitis Pigmentosa
RP9	Retinitis Pigmentosa	***SPG7***	Spastic Paraplegia 7
RPE65	Leber Congenital Amaurosis	***STARD3***	Cardiomyopathy (Dilated)
RPGR	Retinitis Pigmentosa	***TAF1***	X-Linked Dystonia-Parkinsonism
RPGRIP1	Leber Congenital Amaurosis	***TAZ***	Cardiomyopathy (Dilated)
RPL11	Diamond-Blackfan Anemia	***TBX5***	Holt-Oram Syndrome
RPL35A	Diamond-Blackfan Anemia	***TCOF1***	Treacher Collins Syndrome
RPS6KA3	Coffin-Lowry Syndrome	***TGFBR1***	Thoracic Aortic Aneurysms and Aortic Dissections
RPS7	Familial Hypertrophic Cardiomyopathy	***TGFBR2***	Thoracic Aortic Aneurysms and Aortic Dissections
RPS10	Diamond-Blackfan Anemia	***TMEM43***	Arrhythmogenic Right Ventricular Dysplasia/Cardiomyopathy
RPS19	Diamond-Blackfan Anemia	***TNNC1***	Dilated Cardiomyopathy
RPS24	Diamond-Blackfan Anemia	***TNNI3***	Dilated Cardiomyopathy
RPS26	Diamond-Blackfan Anemia	***TNNT1***	Nemaline Myopathy
RS1	X-Linked Juvenile Retinoschisis	***TNNT2***	Familial Hypertrophic Cardiomyopathy
RSPH4A	Primary Ciliary Dyskinesia	***TNXB***	Ehlers-Danlos Syndrome, Hypermobility Type
RSPH9	Primary Ciliary Dyskinesia	***TOPORS***	Retinitis Pigmentosa
RYR1	Malignant Hyperthermia Susceptibility	***TP53***	Li-Fraumeni Syndrome
RYR2	Arrhythmogenic Right Ventricular Dysplasia/Cardiomyopathy	***TPM1***	Familial Hypertrophic Cardiomyopathy
SALL4	Duane Syndrome - Autosomal Dominant	***TSC1***	Tuberous Sclerosis Complex
SCN1B	Brugada Syndrome	***TSC2***	Tuberous Sclerosis Complex
SCN3B	Brugada Syndrome	***TTPA***	Ataxia with Vitamin E Deficiency
SCN4B	Long QT Syndrome, Autosomal Dominant	***TTR***	Familial Transthyretin Amyloidosis
SCN5A	Brugada Syndrome	***TULP1***	Retinitis Pigmentosa
SCN9A	SCN9A-Related Inherited Erythromelalgia	***TWIST1***	Saethre-Chotzen Syndrome
SEMA4A	Retinitis Pigmentosa	***TXNDC3***	Primary Ciliary Dyskinesia
SERPINA1	Alpha-1-Antitrypsin Deficiency	***TYR***	Oculocutaneous Albinism Type 1
SERPING1	Angioedema, Hereditary, Types I and II	***USH1C***	Usher Syndrome Type 1
SGCD	Dilated Cardiomyopathy	***USH2A***	Usher Syndrome Type 2
SH3BP2	Cherubism	***VCL***	Familial Hypertrophic Cardiomyopathy
SIX1	Branchiootorenal Spectrum Disorders	***VHL***	von Hippel-Lindau Syndrome
SIX5	Branchiootorenal Spectrum Disorders	***WAS***	Wiskott-Aldrich Syndrome
SLC25A4	Familial Hypertrophic Cardiomyopathy	***WRN***	Werner Syndrome
SLC25A13	Citrin Deficiency	***WT1***	Wilms Tumor, Classical

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