



GENÉTICA MOLECULAR DE COLOMBIA

ESTUDIOS DE ADN ORDENADOS POR GENE

Gene	Enfermedad	Comentario
AAAS (ALADIN, ADRACALIN)	ACHALASIA-ADDISONIANISM-ALACRIMA SYNDROME, AAA » TRIPLE-A SYNDROME » ALACRIMA-ACHALASIA-ADRENAL INSUFFICIENCY NEUROLOGIC DISORDER » GLUCOCORTICOID DEFICIENCY AND ACHALASIA » ALLGROVE SYNDROME » ADDISONIAN-ACHALASIA SYNDROME » HYPOADRENALISM WITH ACHALASIA » ALACRIMA-ACHALASIA-ADDISONIANISM » ACTH-RESISTANT ADRENAL INSUFFICIENCY, ACHALASIA AND ALACRIMA » ACHALASIA-ALACRIMA SYNDROME	
ABCA4 (ATP-BINDING CASSETTE, SUBFAMILY A, MEMBER 4, ABCR)	STARGARDT DISEASE, TYPE 1 » MACULAR DEGENERATION, JUVENILE » FUNDUS FLAVIMACULATUS » MACULAR DYSTROPHY WITH FLECKS, TYPE 1	5 Exons: 28-32

MACULAR DEGENERATION, AGE-RELATED, TYPE 2 »
MACULOPATHY, AGE-RELATED, TYPE 2 » MACULAR
DEGENERATION, SENILE
RETINITIS PIGMENTOSA 19, RP19

CONE-ROD DYSTROPHY, TYPE 3, CORD3

ABCA12 (ATP-BINDING CASSETTE,
SUBFAMILY A, MEMBER 12)

ICHTHYOSIS, LAMELLAR, 2, LI2 » LAMELLAR
ICHTHYOSIS, TYPE 2 » ICHTHYOSIS CONGENITA 2B

5 Exons: 28-32

ICHTHYOSIS CONGENITA, HARLEQUIN FETUS TYPE »
HARLEQUIN ICHTHYOSIS

Whole Gene

ABCC8 (ATP-BINDING CASSETTE,
SUBFAMILY C, MEMBER 8, SUR1)

HYPERINSULINEMIC HYPOGLYCEMIA, FAMILIAL, TYPE
1 » PERSISTENT HYPERINSULINEMIC HYPOGLYCEMIA
OF INFANCY » HYPOGLYCEMIA, HYPERINSULINEMIC,
OF INFANCY » HYPERINSULINEMIC HYPOGLYCEMIA
DUE TO FOCAL ADENOMATOUS HYPERPLASIA »
NESIDIOBLASTOSIS OF PANCREAS »
HYPERINSULINISM, FAMILIAL, WITH PANCREATIC
NESIDIOBLASTOSIS » HYPERINSULINISM,
CONGENITAL ONEMIA SYNDROME

DIABETES MELLITUS, PERMANENT NEONATAL

ABCB7 (ATP-BINDING CASSETTE,
SUBFAMILY B, MEMBER 7, ABC
TRANSPORTER 7)

ANEMIA, SIDEROBLASTIC, AND SPINOCEREBELLAR
ATAXIA

ABCB11 (ATP-BINDING CASSETTE,
SUBFAMILY B, MEMBER 11)

CHOLESTASIS, BENIGN RECURRENT INTRAHEPATIC,
BRIC » SUMMERSKILL SYNDROME

CHOLESTASIS, PROGRESSIVE FAMILIAL INTRAHEPATIC
1, PFIC1 » BYLER DISEASE

ABCC6 (MULTIDRUG RESISTANCE-ASSOCIATED PROTEIN 6, MRP6)	PSEUDOXANTHOMA ELASTICUM (AUTOSOMAL RECESSIVE), PXE » GRONBLAD - STRANDBERG SYNDROME (AUTOSOMAL RECESSIVE)	Whole Gene or R1141X representing 30% of mutations or Deletion Exons 23-29 representing 10% of mutations
	PSEUDOXANTHOMA ELASTICUM (AUTOSOMAL DOMINANT), PXE » GRONBLAD - STRANDBERG SYNDROME (AUTOSOMAL DOMINANT)	Whole Gene or R1141X representing 30% of mutations or Deletion Exons 23-29 representing 10% of mutations
ABCD1	ADRENOLEUKODYSTROPHY, ALD » ADRENOMYELONEUROPATHY, AMN	
ACAD8 (ACYL-CoA DEHYDROGENASE FAMILY, MEMBER 8)	ISOBUTYRYL GLYCINURIA » ISOBUTYRYL-CoA DEHYDROGENASE DEFICIENCY » ACAD8 DEFICIENCY	
ACADM	MEDIUM CHAIN ACYL-COA DEHYDROGENASE DEFICIENCY, MCAD	Whole Gene or 1 Mutation: K329E
ACADS (ACYL-CoA DEHYDROGENASE, SHORT-CHAIN)	SCAD DEFICIENCY » SHORT-CHAIN ACYL-CoA DEHYDROGENASE DEFICIENCY	Preferentially on skin fibroblast culture for mutation analysis in proband, eventually blood in PAX RNA tubes
ACADSB (ACYL-CoA DEHYDROGENASE, SHORT/BRANCHED CHAIN)	2-ALPHA-METHYLBUTYRYLGLYCINURIA » 2-ALPHA-METHYLBUTYRYL-CoA DEHYDROGENASE DEFICIENCY, MBD	
ACADVL (ACYL-CoA DEHYDROGENASE, VERY LONG-CHAIN, VLCAD)	ACYL-CoA DEHYDROGENASE, VERY LONG-CHAIN, DEFICIENCY OF, VLCAD DEFICIENCY	
ACTA1 (ACTIN)	ACTIN MYOPATHY NEMALINE MYOPATHY (AUTOSOMAL DOMINANT), NEM1 NEMALINE MYOPATHY (AUTOSOMAL RECESSIVE), NEM2	

	MYOPATHY, CONGENITAL, WITH FIBER-TYPE DISPROPORTION	
ACTC (ACTIN, ALPHA, CARDIAC MUSCLE, SMOOTH MUSCLE ACTIN)	DILATED CARDIOMYOPATHY HYPERTROPHIC CARDIOMYOPATHY, FAMILIAL, 1, CMH1 » VENTRICULAR HYPERTROPHY, HEREDITARY » ASYMMETRIC SEPTAL HYPERTROPHY » HYPERTROPHIC SUBAORTIC STENOSIS, IDIOPATHIC	See ACTC, MYL2 and MYL3 See ACTC, MYL2 and MYL3
ACTC MYL2 MYL3 ACTN4 (ACTININ, ALPHA-4) ADA (ADENOSINE DEAMINASE, ADENOSINE AMINOHYDROLASE)	FOCAL SEGMENTAL GLOMERULOSCLEROSIS, TYPE 1 FOCAL SEGMENTAL GLOMERULOSCLEROSIS, TYPE 1 SEVERE COMBINED IMMUNODEFICIENCY, (AUTOSOMAL RECESSIVE), T CELL-NEGATIVE, B CELL-NEGATIVE, NK CELL-NEGATIVE, DUE TO ADENOSINE DEAMINASE DEFICIENCY, SCID	19 Exons
ADAMTS13 (VON WILLEBRAND FACTOR-CLEAVING PROTEASE)	THROMBOTIC THROMBOCYTOPENIC PURPURA, CONGENITAL, TTP » SCHULMAN-UPSHAW SYNDROME HEMOLYTIC-UREMIC SYNDROME, HUS » COMBINED DEFICIENCY OF FACTOR H AND FACTOR H-LIKE 1	
ADSL (ADENYLOSUCCINATE LYASE)	ADENYLOSUCCINASE DEFICIENCY » SUCCINYLPURINEMIC AUTISM	Preferentially on skin fibroblast culture for mutation analysis in proband, eventually blood in PAX RNA tubes
AGL (AMYLO-1,6-GLUCOSIDASE, 4-ALPHA-GLUCANOTRANSFER, GLYCOGEN DEBRANCHER ENZYME)	GLYCOGEN STORAGE DISEASE, TYPE 3 » GSD TYPE 3 » FORBES DISEASE » CORI DISEASE » AMYLO-1,6-GLUCOSIDASE DEFICIENCY » GLYCOGEN DEBRANCHER DEFICIENCY	
AGXT (ALANINE-GLYOXYLATE AMINOTRANSFERASE, AGT, SERINE-PYRUVATE AMINOTRANSFERASE, SPT)	HYPEROXALURIA, PRIMARY, TYPE 1 » OXALOSIS 1 » GLYCOLIC ACIDURIA » ALANINE-GLYOXYLATE AMINOTRANSFERASE DEFICIENCY » HEPATIC AGT DEFICIENCY » SERINE:PYRUVATE AMINOTRANSFERASE DEFICIENCY	Whole Gene or 3 Exons: 1, 4 and 7 (Including 33-34insC, 508A, 731C Mutations)

AHI1 (ABELSON HELPER INTEGRATION SITE 1, JOUBERIN)	JOUBERT SYNDROME, TYPE 3	
AICDA (ACTIVATION-INDUCED CYTIDINE DEAMINASE, AID)	IMMUNODEFICIENCY WITH HYPER-IgM, TYPE 2 » HYPER-IgM SYNDROME 2	
AICDA, CD40, CD40LG, UNG	IMMUNODEFICIENCY WITH HYPER-IgM » HYPER-IgM SYNDROME	
AIRE (AUTOIMMUNE REGULATOR)	AUTOIMMUNE POLYENDOCRINOPATHY SYNDROME, TYPE 1 » AUTOIMMUNE POLYENDOCRINOPATHY-CANDIDIASIS-ECTODERMAL DYSTROPHY, APECED » AUTOIMMUNE POLYGLANDULAR SYNDROME, TYPE 1 » HYPOADRENOCORTICISM WITH HYPOPARATHYROIDISM AND SUPERFICIAL MONILIASIS » POLYGLANDULAR DEFICIENCY SYNDROME, PERSIAN-JEWISH TYPE	Whole Gene
ALDOB (ALDOLASE B)	FRUCTOSE INTOLERANCE » FRUCTOSEMIA » FRUCTOSE-1-PHOSPHATE » ALDOLASE B DEFICIENCY	Whole Gene or 3 Common Mutations: A149P, A174D, N334K
ALK1 (ACTIVIN A RECEPTOR, TYPE II-LIKE 1, ACVRL1)	ELANGIECTASIA, HEREDITARY HEMORRHAGIC, OF RENDU, OSLER, AND WEBER, TYPE 2 » OSLER-RENDU-WEBER DISEASE, TYPE 2 » ORW DISEASE, TYPE 2	
ALMS1	ALSTROM SYNDROME, ALMS	Exons 10, 16, and part of Exon 8
ALOX12B (ARACHIDONATE 12-LIPOXYGENASE, R TYPE, 12R-@LIPOXYGENASE)	ICHTHYOSIFORM ERYTHRODERMA, NONBULLOUS CONGENITAL	
ALOXE3 (ARACHIDONATE LIPOXYGENASE 3, LIPOXYGENASE TYPE 3)	ICHTHYOSIFORM ERYTHRODERMA, NONBULLOUS CONGENITAL	
ALX4 (ARISTALESS-LIKE 4, MOUSE, HOMOLOG OF)	PARIETAL FORAMINA, TYPE 2 » FORAMINA PARIETALIA PERMAGNA » CATLIN MARKS	
AMPD1 (AMP DEAMINASE)	MYOADENYLATE DEAMINASE DEFICIENCY	2 Mutations: Q12X, P48L

AMT (AMINOMETHYLTRANSFERASE, GLYCINE CLEAVAGE SYSTEM T PROTEIN)	NONKETOTIC HYPERGLYCINEMIA » GLYCINE ENCEPHALOPATHY	
ANG (ANGIOGENIN, RNASE5)	AMYOTROPHIC LATERAL SCLEROSIS, FAMILIAL, FALS, ALS1	
ANGELMAN	AMYOTROPHIC LATERAL SCLEROSIS, FAMILIAL, FALS, ALS1	
ANTITRYPSINE	See PI	
APC	POLYPOSIS COLI, ADENOMATOUS » FAMILIAL ADENOMATOUS POLYPOSIS, FAP » ATTENUATED POLYPOSIS COLI	Whole Gene or MLPA for Deletions-Duplications
APOA1 (APOLIPOPROTEIN A-1)	HYPO-ALPHALIPOPROTEINEMIA	L178P Mutation
APOB (APOLIPOPROTEIN B, APOB100, APOB48)	HYPERCHOLESTEROLEMIA (AUTOSOMAL DOMINANT), TYPE B » APOLIPOPROTEIN B-100, FAMILIAL LIGAND - DEFECTIVE	2 Mutations: R3500Q and R3531C
	HYPERCHOLESTEROLEMIA	3 mutations: R3500Q, R3500W, H3543Y
	HYPOBETALIPOPROTEINEMIA, FAMILIAL » ABETALIPOPROTEINEMIA, NORMOTRIGLYCERIDEMIC, STEINBERG TYPE » FAMILIAL HYPOBETALIPOPROTEINEMIA » ACANTHOCYTOSIS WITH HYPOBETALIPOPROTEINEMIA	Whole Gene
APOC2 (APOLIPOPROTEIN C2)	HYPERCHYLOMICRONEMIA	
APOE (APOLIPOPROTEIN E)	DYSBETALIPOPROTEINEMIA DUE TO DEFECT IN APOLIPOPROTEIN E » APOLIPOPROTEIN E DEFICIENCY » HYPERLIPOPROTEINEMIA, TYPE 3 » FAMILIAL HYPERBETA- AND PREBETALIPOPROTEINEMIA » FAMILIAL HYPERCHOLESTEROLEMIA WITH HYPERLIPEMIA	
APP	ALZHEIMER DEMENTIA, EARLY-ONSET, TYPE 1, AD1	Whole Gene or Exons 16 and 17

		harbouring the majority of mutations Whole Gene or Exons 16 and 17 harbouring the majority of mutations
APRT (ADENINE PHOSPHORIBOSYLTRANSFERASE)	CEREBRAL AMYLOID ANGIOPATHY » AMYLOIDOSIS, CEREBROARTERIAL » AMYLOIDOSIS 6 » HEREDITARY CEREBRAL HEMORRHAGE WITH AMYLOIDOSIS, HCHWA » AMYLOIDOSIS, CEREBRAL AMYLOID ANGIOPATHY APRT DEFICIENCY	
APTAX (APRATAXIN)	ATAXIA, EARLY-ONSET, WITH OCULOMOTOR APRAXIA AND HYPOALBUMINEMIA » ATAXIA-OCULOMOTOR APRAXIA SYNDROME » ATAXIA-OCULOMOTOR APRAXIA 1 » ATAXIA-TELANGIECTASIA-LIKE SYNDROME » CEREBELLAR ATAXIA, EARLY-ONSET, WITH HYPOALBUMINEMIA » ATAXIA, ADULT-ONSET, WITH OCULOMOTOR APRAXIA	
AQP2 (AQUAPORIN 2)	DIABETES INSIPIDUS, NEPHROGENIC (AUTOSOMAL RECESSIVE) DIABETES INSIPIDUS, NEPHROGENIC (AUTOSOMAL DOMINANT)	
AR (ANDROGEN RECEPTOR)	ANDROGEN INSENSITIVITY SYNDROME, AIS REIFENSTEIN SYNDROME HYPOSPADIAS (X-LINKED) KENNEDY DISEASE » SPINAL AND BULBAR MUSCULAR ATROPHY, SBMA	Repeat
ARH	HYPERCHOLESTEROLEMIA (AUTOSOMAL RECESSIVE), ARH	
ARIX (ARISTALESS HOMEBOX, DROSOPHILA, HOMOLOG , PHOX2A)	FIBROSIS OF EXTRAOCULAR MUSCLES, CONGENITAL, TYPE2, FEOM2, CFEOM2	
ARSA (ARYLSULFATASE A, CEREBROSIDE-SULFATASE)	METACHROMATIC LEUKODYSTROPHY » SULFATIDE LIPIDOSIS » ARYLSULFATASE A DEFICIENCY PSEUDOARYLSULFATASE A DEFICIENCY	

ARSB (ARYLSULFATASE B, N-ACETYL GALACTOSAMINE-4-SULFATASE)	MUCOPOLYSACCHARIDOSIS TYPE 6, MPS6 » MAROTEAUX-LAMY SYNDROME » ARYLSULFATASE B DEFICIENCY » N-ACETYL GALACTOSAMINE-4-SULFATASE DEFICIENCY	
ARSC1 (STEROID SULFATASE, STS)	ICHTHYOSIS (X-LINKED) » STEROID SULFATASE DEFICIENCY	Deletion
ARX	INFANTILE SPASMS (X-LINKED), ISS X » WEST SYNDROME » EPILEPSY, INFANTILE SPASMS (X-LINKED) PARTINGTON SYNDROME, MRXS1 » MENTAL RETARDATION, PARTINGTON SYNDROME, MRXS1 LISSENCEPHALY WITH AMBIGUOUS GENITALIA (X-LINKED) MYOCLONIC EPILEPSY WITH MENTAL RETARDATION AND SPASTICITY (X-LINKED) MENTAL RETARDATION, NONSPECIFIC (X-LINKED), TYPE54, MRX54	See also Mental Retardation Panel
ASPA (ASPARTOACYLASE)	CANAVAN DISEASE » (See also Molecular Screening Tests)	3 Mutations: E285A, A305E, Y231X
ASS (ARGININOSUCCINATE SYNTHETASE)	CITRULLINEMIA, CLASSIC » CITRULLINEMIA, TYPE 1 » CITRULLINURIA » ARGININOSUCCINATE SYNTHETASE DEFICIENCY	
AT3 (SERPINC1)	ANTITHROMBIN 3 DEFICIENCY » THROMBOPHILIA, HEREDITARY, DUE TO DEFICIENCY OF AT3	
ATM	ATAXIA-TELANGIECTASIA, AT » LOUIS-BAR SYNDROME	
ATP1A2	FAMILIAL PARAPLEGIC MIGRAINE TYPE 2	Whole Gene or 2 Mutations: L764P and W887R
ATP2A2 (ATP2B, SERCA2)	DARIER-WHITE DISEASE » KERATOSIS FOLLICULARIS » DARIER DISEASE ACROKERATOSIS VERRUCIFORMIS » HOPF DISEASE	
ATP2C1	HAILEY-HAILEY DISEASE » PEMPHIGUS, BENIGN FAMILIAL	

ATP6V1B1 (ATP6B1 VACUOLAR PROTON PUMP, SUBUNIT 3, VPP3)	RENAL TUBULAR ACIDOSIS, DISTAL, WITH PROGRESSIVE NERVE DEAFNESS	
ATP7A	MENKES DISEASE » KINKY HAIR DISEASE NEONATAL CUTIS LAXA » OCCIPITAL HORN SYNDROME	
ATP7B	WILSON DISEASE » HEPATOLENTICULAR DEGENERATION	Whole Gene or 1 Mutation: HIS1069GLN
ATP8B1 (FIC1)	CHOLESTASIS, BENIGN RECURRENT INTRAHEPATIC, BRIC » SUMMERSKILL SYNDROME CHOLESTASIS, PROGRESSIVE FAMILIAL INTRAHEPATIC 1, PFIC1 » BYLER DISEASE	
ATP6V0A4 (ATP6N2 VACUOLAR PROTEIN PUMP, SUBUNIT 2, VPP)	RENAL TUBULAR ACIDOSIS, DISTAL (AUTOSOMAL RECESSIVE) » RENAL TUBULAR ACIDOSIS (AUTOSOMAL RECESSIVE) WITH PRESERVED HEARING » RENAL TUBULAR ACIDOSIS, DISTAL, (AUTOSOMAL RECESSIVE) WITH LATE-ONSET SENSORINEURAL HEARING LOSS	
ATROPHIN1	See DRPLA	
ATRX (XNP)	ALPHA-THALASSEMIA / MENTAL RETARDATION SYNDROME, ATRX (X-LINKED) » ATR-X SYNDROME » XLMR-HYPOTONIC FACE SYNDROME » MENTAL RETARDATION, XLMR-HYPOTONIC FACE SYNDROME JUBERG-MARSIDI SYNDROME » MENTAL RETARDATION, WITH GROWTH RETARDATION, DEAFNESS, AND MICROGENITALISM (X-LINKED) SMITH-FINEMAN-MYERS MENTAL RETARDATION SYNDROME » MENTAL RETARDATION, SMITH-FINEMAN-MYERS SYNDROME	Blood in RNA PAX tubes Blood in RNA PAX tubes Blood in RNA PAX tubes
ATX1 (ATAXIN 1, SCA1, OPCA1)	SPINOCEREBELLAR ATAXIA 1, SCA1	See SCA1, SCA2, SCA3, SCA6, SCA7
ATX2 (ATAXIN 2, SCA2, OPCA2)	SPINOCEREBELLAR ATAXIA 2, SCA2	See SCA1, SCA2, SCA3, SCA6, SCA7
ATX3 (ATAXIN 3, SCA3)	SPINOCEREBELLAR ATAXIA 3, SCA3 » MACHADO-JOSEPH DISEASE	See SCA1, SCA2, SCA3, SCA6, SCA7
ATXN8OS (ATAXIN 8 OPPOSITE STRAND, SCA8)	SPINOCEREBELLAR ATAXIA 8, SCA8	Repeat

ATXN10 (SCA10)	SPINOCEREBELLAR ATAXIA 10, SCA10	Repeat
AUH (AU-SPECIFIC RNA-BINDING PROTEIN, 3-ALPHA-METHYLGLUTACONYL -CoA HYDRATASE)	3-ALPHA-METHYLGLUTACONICACIDURIA, TYPE I » 3-ALPHA-METHYLGLUTACONYL-CoA HYDRATASE DEFICIENCY » 3-ALPHA-MG-CoA-HYDRATASE DEFICIENCY » MGA, TYPE 1	
AVPR2 (VASOPRESSIN RECEPTOR 2, ANTIDIURETIC HORMONE RECEPTOR)	DIABETES INSIPIDUS, NEPHROGENIC (X-LINKED)	
AZFa, AZFb and AZFc (including DAZ)	AZOSPERMIA-OLIGOSPERMIA » SERTOLI-CELL-ONLY SYNDROME » MALE INFERTILITY	Deletions
BBS1	BARDET-BIEDL SYNDROME TYPE 1, BBS1	
BBS2	BARDET-BIEDL SYNDROME TYPE 2, BBS2	
BBS10	BARDET-BIEDL SYNDROME TYPE 10, BBS10	
BBS12	BARDET-BIEDL SYNDROME TYPE 12, BBS12	
BCKDHA (BRANCHED-CHAIN KETO ACID DEHYDROGENASE E1, ALPHA POLYPEPTIDE)	MAPLE SYRUP URINE DISEASE » BRANCHED-CHAIN KETOACIDURIA » BRANCHED-CHAIN ALPHA-KETO ACID DEHYDROGENASE DEFICIENCY » KETO ACID DECARBOXYLASE DEFICIENCY » LIPOAMIDE DEHYDROGENASE DEFICIENCY, LACTIC ACIDOSIS DUE TO	
BCS1L (BCS1, S. CEREVISIAE, HOMOLOG-LIKE)	CYTOCHROME c OXIDASE DEFICIENCY » COX DEFICIENCY » COMPLEX 3, MITOCHONDRIAL RESPIRATORY CHAIN, DEFICIENCY OF GRACILE SYNDROME » GROWTH RETARDATION, AMINO ACIDURIA, CHOLESTASIS, IRON OVERLOAD, LACTIC ACIDOSIS, AND EARLY DEATH » FINNISH LETHAL NEONATAL METABOLIC SYNDROME » LACTIC ACIDOSIS, FINNISH, WITH HEPATIC HEMOSIDEROSIS » FELLMAN SYNDROME LEIGH SYNDROME	
BMPR1A (BONE MORPHOGENETIC PROTEIN RECEPTOR, TYPE 1A, ACTIVIN A RECEPTOR, TYPE II-LIKE KINASE 3, ACVRLK3)	JUVENILE POLYPOSIS SYNDROME POLYPOSIS SYNDROME, HEREDITARY MIXED, TYPE 2 COWDEN DISEASE » LHERMITTE-DUCLOS DISEASE	

BMPR1B (BONE MORPHOGENETIC PROTEIN RECEPTOR, TYPE IB, ACTIVIN RECEPTOR-LIKE KINASE 6)	BRACHYDACTYLY TYPE A2, BDA2 » BRACHYMESOPHALANGY 2 » MOHR - WRIEDT TYPE BRACHYDACTYLY	
BMPR2	PRIMARY PULMONARY HYPERTENSION, PPH1	
BRAF (V-RAF MURINE SARCOMA VIRAL ONCOGENE HOMOLOG B1, RAFB1)	CARDIOFACIOCUTANEOUS SYNDROME, CFC LYMPHOMA, NON-HODGKIN NONSMALL CELL LUNG CANCER, SOMATIC ADENOCARCINOMA OF LUNG, SOMATIC COLORECTAL CANCER, SOMATIC THYROID CARCINOMA, PAPILLARY, SOMATIC MELANOMA, MALIGNANT, SOMATIC	
BRCA1	BREAST AND OVARIAN CANCER PROSTATE CANCER	Whole Gene (sequencing and MLPA) Whole Gene (sequencing and MLPA)
BRCA1 and BRCA2	BREAST AND OVARIAN CANCER	2 Whole Genes (sequencing and MLPA) or MLPA Discounts are available for more than 100 tests per year
	PROSTATE CANCER	Whole Gene (sequencing and MLPA) or 3 Common Ashkenazi Jewish Mutations: 185delAG and 5382insC (BRCA1) and 6174delT (BRCA2)
BRCA2	BREAST AND OVARIAN CANCER	Whole Gene (sequencing and MLPA) or 3 Common Ashkenazi Jewish Mutations: 185delAG and 5382insC (BRCA1) and 6174delT (BRCA2)
	PROSTATE CANCER	Whole Gene (sequencing and MLPA) or 3 Common Ashkenazi Jewish Mutations: 185delAG and 5382insC (BRCA1) and 6174delT (BRCA2)

BSCL2 (SEIPIN)	FAMILIAL SPASTIC PARAPLEGIA 17, SPG17 » SPASTIC PARAPLEGIA WITH AMYOTROPHY OF HANDS AND FEET » SILVER SYNDROME » SILVER SPASTIC PARAPLEGIA SYNDROME » SPINAL MUSCULAR ATROPHY, DISTAL, TYPE 5, DSMA5	
BSND (BARTTIN)	BARTTER SYNDROME, TYPE 4 » BARTTER SYNDROME, INFANTILE, WITH SENSORINEURAL DEAFNESS	
BTD (BIOTINIDASE)	BIOTINIDASE DEFICIENCY » MULTIPLE CARBOXYLASE DEFICIENCY, LATE-ONSET » MULTIPLE CARBOXYLASE DEFICIENCY, JUVENILE-ONSET » BTD DEFICIENCY	8 Mutations: D444H, A171T, F403V, G98, 7-BP DEL/3-BP INS, Q456H, R157H, R538C, D252G, Detecting 60% of all BTD Mutations
BTK (BRUTON TYROSINE KINASE, ATK, BPK)	AGAMMAGLOBULINEMIA (X-LINKED), XLA » BRUTON AGAMMAGLOBULINEMIA » HYPOGAMMAGLOBULINEMIA (X-LINKED)	
C10ORF2 (CHROMOSOME 10 OPEN READING FRAME 2, T7 GENE 4-LIKE PROTEIN WITH INTRAMITOCHONDRIAL NUCLEOID LOCALIZATION, TWINKLE)	PROGRESSIVE EXTERNAL OPHTHALMOPLEGIA WITH MITOCHONDRIAL DNA DELETIONS (AUTOSOMAL DOMINANT), 3	
C1NH (C1 ESTERASE INHIBITOR, SERPING 1)	PROGRESSIVE EXTERNAL OPHTHALMOPLEGIA WITH MITOCHONDRIAL DNA DELETIONS, DIGENIC SENSORY ATAXIC NEUROPATHY, DYSARTHRIA, AND OPHTHALMOPARESIS, SANDO » SPINOCEREBELLAR ATAXIA WITH EPILEPSY	
CA2 (CARBONIC ANHYDRASE 2)	ANGIOEDEMA » ANGIONEUROTIC EDEMA » QUINCKE EDEMA OSTEOPETROSIS WITH RENAL TUBULAR ACIDOSIS » GUIBAUD - VAINSEL SYNDROME » CARBONIC ANHYDRASE 2 DEFICIENCY » MARBLE BRAIN DISEASE	
CACNA1A (CALCIUM CHANNEL,	SPINOCEREBELLAR ATAXIA 6, SCA6	See SCA1, SCA2, SCA3, SCA6, SCA7

VOLTAGE-DEPENDENT, P/Q TYPE, ALPHA-1A SUBUNIT, CALCIUM CHANNEL, L TYPE, ALPHA-1 POLYPEPTIDE, ISOFORM 4, CACNL1A4)	MIGRAINE, FAMILIAL HEMIPLEGIC, 1 » MIGRAINE, SPORADIC HEMIPLEGIC EPISODIC ATAXIA, TYPE 2, EA2 » ATAXIA, EPISODIC, WITH NYSTAGMUS » EPISODIC ATAXIA, NYSTAGMUS-ASSOCIATED » CEREBELLOPATHY, HEREDITARY PAROXYSMAL ATAXIA » FAMILIAL PAROXYSMAL ACETAZOLAMIDE-RESPONSIVE » HEREDITARY PAROXYSMAL CEREBELLAR ATAXIA » CEREBELLAR ATAXIA, PAROXYSMAL, ACETAZOLAMIDE-RESPONSIVE	
CACNA1S (CACNL1A3)	HYPOKALEMIC PERIODIC PARALYSIS, HOKPP	Exons 11, 26 and 30, containing all described mutations or 3 Mutations: R528H, R1239H, R1239G
	MALIGNANT HYPERTHERMIA SUSCEPTIBILITY 5, MHS5	Exons 11, 26 and 30, containing all described mutations or 3 Mutations: R528H, R1239H, R1239G
	THYROTOXIC PERIODIC PARALYSIS » HASHITOXIC PERIODIC PARALYSIS	Exons 11, 26 and 30, containing all described mutations or 3 Mutations: R528H, R1239H, R1239G
CACNA1S (CACNL1A3) and SCN4A	HYPOKALEMIC PERIODIC PARALYSIS, HOKPP	4 Mutations in CACNA1S: R528G, R528H, R1239H, R1239G and 5 Mutations in SCN4A: R672S, R672H, R672G, R672C, R669H
CAPN3 (CALPAIN 3)	LIMB GIRDLE MUSCULAR DYSTROPHY 2A, LGMD2A » MUSCULAR DYSTROPHY, LIMB GIRDLE MUSCULAR DYSTROPHY 2A, LGMD2A	
CASP8 (CASPASE 8, FLICE, MCH5)	CASPASE 8 DEFICIENCY AUTOIMMUNE LYMPHOPROLIFERATIVE SYNDROME, TYPE 1, ALPS, ALPS1A, ALPS1B	
CASP10 (CASPASE 10, MCH4, CASP10B, FLICE2)	AUTOIMMUNE LYMPHOPROLIFERATIVE SYNDROME, TYPE 2, ALPS2	
CASQ2 (CALSEQUESTRIN 2)	VENTRICULAR TACHYCARDIA, CATECHOLAMINERGIC POLYMORPHIC » VENTRICULAR TACHYCARDIA, STRESS-INDUCED POLYMORPHIC	

CASR (CALCIUM-SENSING RECEPTOR, PCAR1)	HYPOCALCIURIC HYPERCALCEMIA, FAMILIAL, TYPE 1 » FAMILIAL BENIGN HYPERCALCEMIA 1 HYPOCALCEMIA (AUTOSOMAL DOMINANT)	
	NEONATAL SEVERE PRIMARY HYPERPARATHYROIDISM HYPOPARATHYROIDISM, FAMILIAL ISOLATED	
CAT (CATALASE)	ACATALASEMIA » ACATALASIA » CATALASE DEFICIENCY	
CAV3 (CAVEOLIN 3)	MUSCULAR DYSTROPHY, LIMB-GIRDLE, TYPE 1C, LGMD -1C RIPPLING MUSCLE DISEASE 2, RMD	
CCM1 (KREV INTERACTION TRAPPED 1, KRIT1)	CEREBRAL CAVERNOUS MALFORMATIONS, CCM1 » CAVERNOUS ANGIOMA, FAMILIAL » HYPERKERATOTIC CUTANEOUS CAPILLARY-VENOUS MALFORMATIONS ASSOCIATED WITH CEREBRAL CAPILLARY MALFORMATIONS	See also CCM1, CCM2 and PDCD10
CCM2 (CHROMOSOME 7 OPEN READING FRAME 22, C7ORF22, MALCAVERNIN, MGC4067)	CEREBRAL CAVERNOUS MALFORMATIONS 2, CCM2	See also CCM1, CCM2 and PDCD10
CCM1, CCM2 and PDCD10 CCR5	CEREBRAL CAVERNOUS MALFORMATIONS, CCM RESISTANCE TO HIV (HUMAN IMMUNODEFICIENCY VIRUS TYPE 1)	Analysis of 3 Genes 1 Mutation: DEL32 BP
CBS (CYSTATHIONINE BETA-SYNTHASE)	HOMOCYSTINURIA » CYSTATHIONINE BETA-SYNTHASE DEFICIENCY	Whole Gene or Exons 4 and 8 (Including GLY307SER and ILE278THR)
CD2AP (CD2-ASSOCIATED PROTEIN)	FOCAL SEGMENTAL GLOMERULOSCLEROSIS, TYPE 3	
CD3D (CD3 ANTIGEN, DELTA SUBUNIT, T-CELL ANTIGEN RECEPTOR COMPLEX, DELTA SUBUNIT OF T3)	SEVERE COMBINED IMMUNODEFICIENCY (AUTOSOMAL RECESSIVE), T CELL-NEGATIVE, B CELL-POSITIVE, NK CELL-POSITIVE, SCID	

CD3E (CD3 ANTIGEN, EPSILON SUBUNIT, T-CELL ANTIGEN RECEPTOR COMPLEX, EPSILON SUBUNIT OF T3)	SEVERE COMBINED IMMUNODEFICIENCY (AUTOSOMAL RECESSIVE), T CELL-NEGATIVE, B CELL-POSITIVE, NK CELL-POSITIVE, SCID IMMUNODEFICIENCY DUE TO DEFECT IN CD3-EPSILON	
CD40 (CD40 ANTIGEN, B CELL-ASSOCIATED MOLECULE CD40 TUMOR NECROSIS FACTOR RECEPTOR SUPERFAMILY, MEMBER 5, TNFRSF5)	IMMUNODEFICIENCY WITH HYPER-IgM, TYPE 3 » HYPER-IgM SYNDROME 3	
CDAN1 (CODANIN 1, DISCS LOST, DROSOPHILA, HOMOLOG OF)	ANEMIA, DYSERYTHROPOIETIC CONGENITAL, TYPE 1 » DYSERYTHROPOIETIC ANEMIA, CONGENITAL, TYPE 1	
CDH1 (CADHERIN 1, UVOMORULIN)	GASTRIC CANCER, FAMILIAL DIFFUSE CLEFT LIP WITH OR WITHOUT CLEFT PALATE, WITH GASTRIC CANCER, FAMILIAL DIFFUSE CANCER (SOMATIC MUTATIONS)	
CDK4	MALIGNANT MELANOMA, CMM3	Exon 2
CDKL5 (CYCLIN-DEPENDENT KINASE-LIKE 5, STK9)	RETT SYNDROME, ATYPICAL INFANTILE SPASMS (X-LINKED), ISS X » WEST SYNDROME » EPILEPSY, INFANTILE SPASMS (X-LINKED) ANGELMAN SYNDROME, ATYPICAL	
CDKN1C (CYCLIN-DEPENDENT KINASE INHIBITOR 1C, p57-KIP2)	BECKWITH-WIEDEMANN SYNDROME, BWS » EXOMPHALOS-MACROGLOSSIA-GIGANTISM SYNDROME	
CEP290 (CENTROSOMAL PROTEIN, 290-KD, NEPHROCYSTIN 6; NPHP6)	JOUBERT SYNDROME, TYPE 5 LEBER CONGENITAL AMAUROSIS, TYPE 1, LCA1 » RETINAL BLINDNESS, CONGENITAL MECKEL SYNDROME, TYPE 4 SENIOR-LOKEN SYNDROME TYPE 6, SLSN6	
CETP (CHOLESTERYL ESTER TRANSFER PROTEIN, LIPID TRANSFER PROTEIN 1)	CHOLESTERYL ESTER TRANSFER PROTEIN DEFICIENCY » CETP DEFICIENCY	

CFI (I FACTOR, COMPLEMENT COMPONENT 3 INACTIVATOR)	COMPLEMENT FACTOR 1 DEFICIENCY	
CFH (HF1, COMPLEMENT FACTOR H)	HEMOLYTIC-UREMIC SYNDROME, HUS » COMBINED DEFICIENCY OF FACTOR H AND FACTOR H-LIKE 1	
CFTR	CYSTIC FIBROSIS, CF	Whole Gene or Mutations Kit or MLPA
	CONGENITAL BILATERAL ABSENCE OF VAS DEFERENS, CBAVD	Whole Gene or Mutations Kit or MLPA
CGI58 (COMPARATIVE GENE IDENTIFICATION 58, ABHD5)	CHANARIN-DORFMAN DISEASE » ICHTHYOTIC NEUTRAL LIPID STORAGE DISEASE » NEUTRAL LIPID STORAGE DISEASE » ICHTHYOSIFORM ERYTHRODERMA WITH LEUKOCYTE VACUOLATION » TRIGLYCERIDE STORAGE DISEASE WITH IMPAIRED LONG-CHAIN FATTY ACID OXIDATION	
CHD7 (CHROMODOMAIN HELICASE DNA-BINDING PROTEIN 7)	CHARGE SYNDROME	
CHEK2 (CHECKPOINT KINASE 2)	LI-FRAUMENI SYNDROME 2 BREAST CANCER, FAMILIAL	Whole Gene or *1100delC
CHM (REP1, RAB ESCORT PROTEIN 1, RAB GERANYLGERANYL TRANSFERASE)	CHOROIDEREMIA » TAPETOCHOROIDAL DYSTROPHY, PROGRESSIVE » CHOROIDAL SCLEROSIS	
CHRNA1 (CHOLINERGIC RECEPTOR, NICOTINIC, ALPHA POLYPEPTIDE 1; ACETYLCHOLINE RECEPTOR, MUSCLE, ALPHA SUBUNIT)	MYASTHENIC SYNDROME, CONGENITAL, SLOW-CHANNEL » MYASTHENIC SYNDROME, CONGENITAL, TYPE 2a MYASTHENIC SYNDROME, CONGENITAL, FAST-CHANNEL MULTIPLE PTERYGIUM SYNDROME, LETHAL TYPE » PTERYGIUM SYNDROME, MULTIPLE, LETHAL TYPE	
CHRNA4 (CHOLINERGIC RECEPTOR, NEURONAL NICOTINIC, ALPHA POLYPEPTIDE 4, ACETYLCHOLINE RECEPTOR, NEURONAL NICOTINIC, ALPHA-4 SUBUNIT)	EPILEPSY, NOCTURNAL FRONTAL LOBE, TYPE 1 TOBACCO ADDICTION, SUSCEPTIBILITY TO » NICOTINE DEPENDENCE, SUSCEPTIBILITY TO	
CHRN1 (CHOLINERGIC RECEPTOR,	MYASTHENIC SYNDROME, CONGENITAL, SLOW-	

NICOTINIC, BETA POLYPEPTIDE
1;ACETYLCHOLINE RECEPTOR,
MUSCLE, BETA SUBUNIT)

CHANNEL » MYASTHENIC SYNDROME, CONGENITAL,
TYPE 2a
MYASTHENIC SYNDROME, CONGENITAL,
ASSOCIATED WITH ACETYLCHOLINE RECEPTOR
DEFICIENCY » MYASTHENIC SYNDROME,
CONGENITAL, TYPE 1d » MYASTHENIC SYNDROME,
CONGENITAL, ASSOCIATED WITH FACIAL
DYSMORPHISM

CHRNE (CHOLINERGIC RECEPTOR,
NICOTINIC, EPSILON POLYPEPTIDE;
ACETYLCHOLINE RECEPTOR,
MUSCLE, EPSILON SUBUNIT)

MYASTHENIC SYNDROME, CONGENITAL, FAST-
CHANNEL
MYASTHENIC SYNDROME, CONGENITAL,
ASSOCIATED WITH ACETYLCHOLINE RECEPTOR
DEFICIENCY » MYASTHENIC SYNDROME,
CONGENITAL, TYPE 1d » MYASTHENIC SYNDROME,
CONGENITAL, ASSOCIATED WITH FACIAL
DYSMORPHISM
MYASTHENIC SYNDROME, CONGENITAL, SLOW-
CHANNEL » MYASTHENIC SYNDROME, CONGENITAL,
TYPE 2a

CHRND (CHOLINERGIC RECEPTOR,
NICOTINIC, DELTA POLYPEPTIDE;
ACETYLCHOLINE RECEPTOR,
MUSCLE, DELTA SUBUNIT)

MYASTHENIC SYNDROME, CONGENITAL, FAST-
CHANNEL
MYASTHENIC SYNDROME, CONGENITAL, SLOW-
CHANNEL » MYASTHENIC SYNDROME, CONGENITAL,
TYPE 2a

CHRNA (CHOLINERGIC RECEPTOR,
NICOTINIC, ALPHA POLYPEPTIDE)

ESCOBAR SYNDROME » MULTIPLE PTERYGIUM
SYNDROME, ESCOBAR VARIANT » MULTIPLE
PTERYGIUM SYNDROME, NONLETHAL TYPE »
PTERYGIUM COLLI SYNDROME » PTERYGIUM
UNIVERSALE
MULTIPLE PTERYGIUM SYNDROME, LETHAL TYPE »
PTERYGIUM SYNDROME, MULTIPLE, LETHAL TYPE
COLD URTICARIA, FCU » FAMILIAL COLD
AUTOINFLAMMATORY SYNDROME
CINCA SYNDROME » MULTISYSTEM INFLAMMATORY
DISEASE, NEONATAL ONSET, NOMID
MUCKLE-WELLS SYNDROME

CIAS1 (CRYOPYRIN)

CKN1 (CSA)	COCKAYNE SYNDROME, TYPE 1 » COCKAYNE SYNDROME, TYPE A	
CLCN1	MYOTONIA CONGENITA (AUTOSOMAL DOMINANT) » THOMSEN DISEASE MYOTONIA CONGENITA (AUTOSOMAL RECESSIVE) » BECKER MYOTONIA MYOTONIA LEVIOR	
CLCN5	DENT NEPHROCALCINOSIS NEPHROLITHIASIS (X-LINKED)	
CLCNKA (CLCK1, CHLORIDE CHANNEL, KIDNEY, A)	BARTTER SYNDROME, TYPE 4 » BARTTER SYNDROME, INFANTILE, WITH SENSORINEURAL DEAFNESS	
CLCNKB (CLCNB, CHLORIDE CHANNEL, KIDNEY, B)	BARTTER SYNDROME, TYPE 3 » BARTTER SYNDROME, CLASSIC » BARTTER SYNDROME, TYPE 3, WITH HYPOCALCIURIA BARTTER SYNDROME, TYPE 4 » BARTTER SYNDROME, INFANTILE, WITH SENSORINEURAL DEAFNESS	
CLDN16 (CLAUDIN 16, PARACELLIN 1)	HYPOMAGNESEMIA, PRIMARY » MAGNESIUM, DEFECT IN RENAL TUBULAR TRANSPORT OF	
CLDN19 (CLAUDIN 19)	HYPOMAGNESEMIA, RENAL, WITH OCULAR INVOLVEMENT » MACULAR COLOBOMA, BILATERAL, WITH HYPERCALCIURIA	
CLN1 (PALMITOYL-PROTEIN ESTERASE, PPT)	CEROID LIPOFUCSINOSIS, CLN1 » SANTAVUORI DISEASE	
CLN2	CEROID LIPOFUCSINOSIS, CLN2 » JANSKY-BIELSCHOWSKY DISEASE	Whole Gene or 2 Common Mutations: R208X and IVS5-1G>C
CLN3	CEROID LIPOFUCSINOSIS, CLN3 » VOGT-SPIELMEYER DISEASE » BATTEN DISEASE	Whole Gene or Common 1kb Deletion
CLN6	CEROID LIPOFUCSINOSIS, CLN6 » CEROID LIPOFUCSINOSIS, NEURONAL, LATE-INFANTILE	
COCH (COCHLIN)	DEAFNESS, DFNA9 » DEAFNESS, (AUTOSOMAL DOMINANT), NONSYNDROMIC SENSORINEURAL 9	Whole Gene or 1 Mutation: P51S

COH1	COHEN SYNDROME, COH1	Whole Gene or Exon 23, including the "Finnish mutation" (c.3348-3349delCT)
COL1A1 COL1A1 and COL1A2	See COL1A1 and COL1A2 EHLERS-DANLOS TYPE 1, EDS1 EHLERS-DANLOS TYPE 7 , DOMINANT , EDS7 OSTEOGENESIS IMPERFECTA TYPE 1, OI1 OSTEOGENESIS IMPERFECTA TYPE 2, OI2 OSTEOGENESIS IMPERFECTA TYPE 3, OI3 OSTEOGENESIS IMPERFECTA TYPE 4, OI4	Only on skin fibroblast cultures for mutation analysis in proband (See note) Splice site mutations exons 5, 6 and 7 Only on skin fibroblast cultures for mutation analysis in proband (See note) Only on skin fibroblast cultures for mutation analysis in proband (See note) Only on skin fibroblast cultures for mutation analysis in proband (See note) Only on skin fibroblast cultures for mutation analysis in proband (See note)
COL1A2 COL2A1	See COL1A1 and COL1A2 ACHONDROGENESIS TYPE 2 HYPOCHONDROGENESIS KNIEST DYSPLASIA » METATROFIC DWARFISM STICKLER SYNDROME TYPE 1 SPONDYLOEPIPHYSEAL DYSPLASIA (SED CONGENITA)	
COL3A1	EHLERS -DANLOS TYPE 4 EDS 4 FIBROMUSCULAR DYSPLASIA	Only on skin fibroblast cultures for mutation analysis in proband (See note) Only on skin fibroblast cultures for mutation analysis in proband (See note)
COL4A3	ALPORT SYNDROME (AUTOSOMAL RECESSIVE) HEMATURIA, BENIGN FAMILIAL » THIN-BASEMENT-MEMBRANE NEPHROPATHY	

COL4A4	ALPORT SYNDROME (AUTOSOMAL RECESSIVE) HEMATURIA, BENIGN FAMILIAL » THIN-BASEMENT-MEMBRANE NEPHROPATHY	
COL4A5	ALPORT SYNDROME (X-LINKED) » ALPORT SYNDROME-LIKE HEREDITARY NEPHRITIS	
COL5A1 and COL5A2	EHLERS - DANLOS TYPE 1, EDS 1 EHLERS - DANLOS TYPE 2, EDS 2	Only on skin fibroblast cultures for mutation analysis in proband (See note) Only on skin fibroblast cultures for mutation analysis in proband (See note)
COL7A1 (COLLAGEN, TYPE 7, ALPHA-1)	EPIDERMOLYSIS BULLOSA DYSTROPHICA (AUTOSOMAL RECESSIVE) » EPIDERMOLYSIS BULLOSA DYSTROPHICA, TYPE HALLOPEAU-SIEMENS » EPIDERMOLYSIS BULLOSA DYSTROPHICA, LOCALISATA VARIANT (AUTOSOMAL RECESSIVE) EPIDERMOLYSIS BULLOSA DYSTROPHICA (AUTOSOMAL DOMINANT) » EPIDERMOLYSIS BULLOSA DYSTROPHICA, PASINI TYPE » ALBOPAPULOID DOMINANT DYSTROPHIC EPIDERMOLYSIS BULLOSA » EPIDERMOLYSIS BULLOSA, PRETIBIAL WITH LICHENOID FEATURES EPIDERMOLYSIS BULLOSA WITH CONGENITAL LOCALIZED ABSENCE OF SKIN AND DEFORMITY OF NAILS » EPIDERMOLYSIS BULLOSA DYSTROPHICA, BART TYPE TRANSIENT BULLOUS DERMOLYSIS OF THE NEWBORN » EPIDERMOLYSIS BULLOSA DYSTROPHICA, NEONATAL (AUTOSOMAL DOMINANT) » TOENAIL DYSTROPHY, ISOLATED EPIDERMOLYSIS BULLOSA PRURIGINOSA (AUTOSOMAL DOMINANT AND RECESSIVE) EPIDERMOLYSIS BULLOSA DYSTROPHICA WITH SUBCORNEAL CLEAVAGE » EPIDERMOLYSIS BULLOSA SIMPLEX SUPERFICIALIS » EPIDERMOLYSIS BULLOSA DYSTROPHICA, COCKAYNE-TOURAINÉ TYPE	

COL10A1 (COLLAGEN, TYPE X, ALPHA1)	METAPHYSEAL CHONDRODYSPLASIA, SCHMID TYPE	Whole Gene
COL11A1 (COLLAGEN, TYPE 11, ALPHA-1)	MARSHALL SYNDROME	Turn-around-time: 30 Weeks
COL11A2 (COLLAGEN, TYPE 11, ALPHA-2)	STICKLER SYNDROME, TYPE 2	Turn-around-time: 30 Weeks
	STICKLER SYNDROME, TYPE 3	Turn-around-time: 30 Weeks
	DEAFNESS, DFNA53 »DEAFNESS, (AUTOSOMAL DOMINANT), NONSYNDROMIC SENSORINEURAL 53	Turn-around-time: 30 Weeks
	WEISSENBACHER-ZWEYMULLER SYNDROME » PIERRE ROBIN SYNDROME WITH FETAL CHONDRODYSPLASIA	Turn-around-time: 30 Weeks
	DEAFNESS, DFNA13 »DEAFNESS, (AUTOSOMAL DOMINANT), NONSYNDROMIC SENSORINEURAL 13	Turn-around-time: 30 Weeks
	OTOSPONDYLOMEGAEPIPHYSEAL DYSPLASIA, OSMED	Turn-around-time: 30 Weeks
COL17A1 (COLLAGEN, TYPE 17, ALPHA-1)	EPIDERMOLYSIS BULLOSA, GENERALIZED ATROPHIC BENIGN » EPIDERMOLYSIS BULLOSA JUNCTIONALIS, PROGRESSIVE » EPIDERMOLYSIS BULLOSA JUNCTIONALIS, SEVERE NONLETHAL » EPIDERMOLYSIS BULLOSA JUNCTIONALIS, DISSENTIS TYPE » EPIDERMOLYSIS BULLOSA, JUNCTIONAL, LOCALISATA VARIANT	
COMP	MULTIPLE EPIPHYSEAL DYSPLASIA 1, EDM1	Whole Gene or Exons 13, 14 and 16 harbouring 70% of COMP mutations
	PSEUDOACHONDROPLASIA	Whole Gene or Exons 13, 14 and 16 harbouring 70% of COMP mutations
COX10 (CYTOCHROME c OXIDASE ASSEMBLY PROTEIN COX10, HEME A:FARNESYLTRANSFERASE)	CYTOCHROME c OXIDASE DEFICIENCY » COX DEFICIENCY » COMPLEX 4, MITOCHONDRIAL RESPIRATORY CHAIN, DEFICIENCY OF LEIGH SYNDROME	
COX15 (CYTOCHROME c OXIDASE ASSEMBLY PROTEIN COX15)	CYTOCHROME c OXIDASE DEFICIENCY » COX DEFICIENCY » COMPLEX 4, MITOCHONDRIAL RESPIRATORY CHAIN, DEFICIENCY OF LEIGH SYNDROME	

CPT1A (CARNITINE PALMITOYLTRANSFERASE 1, LIVER, CPT1)

CARNITINE PALMITOYLTRANSFERASE 1A DEFICIENCY » CPT1 DEFICIENCY

CPT2 (CARNITINE PALMITOYLTRANSFERASE 2, LIVER, CPT2)

CARNITINE PALMITOYLTRANSFERASE 2 DEFICIENCY, INFANTILE » CARNITINE PALMITOYLTRANSFERASE 2 DEFICIENCY WITH HYPOKETOTIC HYPOGLYCEMIA » CARNITINE PALMITOYLTRANSFERASE 2 DEFICIENCY, HEPATOCARDIOMUSCULAR » CPT2 DEFICIENCY, HEPATIC
CARNITINE PALMITOYLTRANSFERASE 2 DEFICIENCY, LATE-ONSET » CARNITINE PALMITOYLTRANSFERASE 2 (CPT2) DEFICIENCY, MYOPATHIC » CARNITINE PALMITOYLTRANSFERASE 2 (CPT2) DEFICIENCY, ADULT-ONSET » CPT2 DEFICIENCY, MYOPATHIC » CPT2 DEFICIENCY, LATE-ONSET
CARNITINE PALMITOYLTRANSFERASE 2 DEFICIENCY, LETHAL NEONATAL » CARNITINE PALMITOYLTRANSFERASE 2 DEFICIENCY, NEONATAL » CPT2 DEFICIENCY, LETHAL NEONATAL
MYOPATHY, VARIABLE

CRB1 (CRUMBS, DROSOPHILA, HOMOLOG OF, 1)

LEBER CONGENITAL AMAUROSIS DUE TO DEFECT IN CRB1
RETINITIS PIGMENTOSA 12, RP12 » RP12 WITH COATS-LIKE EXUDATIVE VASCULOPATHY » RETINITIS PIGMENTOSA WITH PARAARTERIAL PRESERVATION OF RETINAL PIGMENT EPITHELIUM PIGMENTED PARAVENOUS CHORIORETINAL ATROPHY

CREBBP (CREB-BINDING PROTEIN, CBP)

RUBINSTEIN-TAYBI SYNDROME » BROAD THUMBS AND GREAT TOES, CHARACTERISTIC FACIES, AND MENTAL RETARDATION » BROAD THUMB-HALLUX SYNDROME

Whole Gene

CRTAP (CARTILAGE-ASSOCIATED PROTEIN)	OSTEOGENESIS IMPERFECTA, TYPE 7, OI7	
CRYGD (CRYSTALLIN, GAMMA-D)	CATARACT, PUNCTATE CATARACT, CRYSTALLINE ACULEIFORM OR FROSTED » CACA	
CSTB (CYSTATIN B, STEFIN B)	EPILEPSY, PROGRESSIVE MYOCLONUS » UNVERRICH-LUNDBORG DISEASE	Repeat
CTNS (CYSTINOSIN)	CYSTINOSIS, NEPHROPATHIC CYSTINOSIS, LATE-ONSET JUVENILE OR ADOLESCENT, NEPHROPATHIC TYPE » CYSTINOSIS, INTERMEDIATE CYSTINOSIS, ADULT NONNEPHROPATHIC PYCNODYSTOSIS	
CTSK (CATHEPSIN K)		
CX26 (CONNEXIN 26)	See GJB2	
CX30 (CONNEXIN 30)	See GJB6	
CX30.3 (CONNEXIN 30.3)	See GJB4	
CX31 (CONNEXIN 31)	See GJB3	
CYBA (p22 PHOX)	CHRONIC GRANULOMATOUS DISEASE (AUTOSOMAL RECESSIVE) CYTOCHROME-b-NEGATIVE FORM	
CYBB (p91 PHOX)	CHRONIC GRANULOMATOUS DISEASE (X-LINKED)	
CYP1B1, OPTN and MYOC	GLAUCOMA	Analysis of 3 Genes
CYP1B1 (CYTOCHROME P450, SUBFAMILY 1, POLYPEPTIDE 1)	GLAUCOMA 3, PRIMARY INFANTILE A, GLC3A » BUPHTHALMOS PETERS ANOMALY	See also CYP1B1, OPTN and MYOC
CYP11B1 (CYTOCHROME P450, SUBFAMILY 11B, POLYPEPTIDE 1, STEROID 11-BETA-HYDROXYLASE, P450C11)	ADRENAL HYPERPLASIA, CONGENITAL, DUE TO STEROID 11-BETA-HYDROXYLASE DEFICIENCY » ADRENAL HYPERPLASIA 4 STEROID 11-BETA-HYDROXYLASE DEFICIENCY » 11-@BETA-HYDROXYLASE DEFICIENCY » ADRENAL HYPERPLASIA, HYPERTENSIVE FORM » P450C11B1 DEFICIENCY	See also CYP1B1, OPTN and MYOC Whole Gene or CYP11B1/ CYP11B2 fusion

CYP11B2 (CYTOCHROME P450, SUBFAMILY XIB, POLYPEPTIDE 2, STEROID 11/18-BETA-HYDROXYLASE, STEROID 18-OXIDASE, ALDOSTERONE SYNTHASE, CORTICOSTERONE METHYLOXIDASE)	CORTICOSTERONE METHYLOXIDASE TYPE 1 DEFICIENCY » ALDOSTERONE DEFICIENCY 1 » HYPERRENINEMIC HYPOALDOSTERONISM, FAMILIAL, 1 » ALDOSTERONE DEFICIENCY DUE TO DEFECT IN STEROID 18-HYDROXYLASE » 18-@HYDROXYLASE DEFICIENCY » STEROID 18-@HYDROXYLASE DEFICIENCY	
CYP19A1 (AROMATASE)	AROMATASE DEFICIENCY » PSEUDOHERMAPHRODITISM, FEMALE, DUE TO PLACENTAL AROMATASE DEFICIENCY	Sequence analysis of exons 9 and 10, including the following common mutations: Arg365Gln Val370Met Arg375Cys Arg435Cys Cys437Tyr Arg457Term
CYP21A2	ADRENAL HYPERPLASIA, CONGENITAL DUE TO 21-HYDROXYLASE DEFICIENCY, CAH1 » 21-ALPHA-HYDROXYLASE DEFICIENCY » CYP21 DEFICIENCY HYPERANDROGENISM	Whole Gene and MLPA or 6 most common mutations: Pro30Leu, A/C 655->G, Ile172Asn, Val281Leu, Gln318X, Trp356Arg Whole Gene and MLPA or 6 most common mutations: Pro30Leu, A/C 655->G, Ile172Asn, Val281Leu, Gln318X, Trp356Arg
CYP27A1	CEREBROTENDINOUS XANTHOMATOSIS, CTX » CEREBRAL CHOLESTERINOSIS	
CYP27B1	PSEUDO-VITAMIN D DEFICIENCY RICKETS (AUTOSOMAL RECESSIVE), PDDR » RICKETS, PSEUDO-VITAMIN D DEFICIENCY RICKETS (AUTOSOMAL RECESSIVE)	Sequence analysis of exons 9 and 10, including the following common mutations: Arg365Gln Val370Met Arg375Cys Arg435Cys Cys437Tyr Arg457Term
DAX1 (NROB1)	ADDISON DISEASE (X-LINKED) » ADRENAL HYPOPLASIA, CONGENITAL » CONGENITAL ADRENOCORTICAL HYPOPLASIA WITH HYPOGONADOTROPIC HYPOGONADISM	
DAZ (DELETED IN AZOOSPERMIA)	See AZFa, AZFb and AZFc	

DCLRE1C (DNA CROSS-LINK REPAIR PROTEIN 1C, ARTEMIS)	SEVERE COMBINED IMMUNODEFICIENCY WITH SENSITIVITY TO IONIZING RADIATION, SCID » SEVERE COMBINED IMMUNODEFICIENCY (AUTOSOMAL RECESSIVE), T CELL-NEGATIVE, B CELL-NEGATIVE, NK CELL-POSITIVE, WITH SENSITIVITY TO IONIZING RADIATION » SEVERE COMBINED IMMUNODEFICIENCY, ATHABASKAN-TYPE OMENN SYNDROME » RETICULOENDOTHELIOSIS, FAMILIAL, WITH EOSINOPHILIA » SEVERE COMBINED IMMUNODEFICIENCY WITH HYPEREOSINOPHILIA	
DCX (DOUBLECORTIN)	DOUBLE CORTEX SYNDROME » LISSENCEPHALY (X-LINKED) » SUBCORTICAL LAMINAR HETEROTOPIA	Whole Gene Sequencing or Deletions - Duplications
DES (DESMIN)	MYOPATHY, MYOFIBRILLAR, DESMIN-RELATED » DESMINOPATHY, PRIMARY	
DHCR7 (DEHYDROCHOLESTEROL REDUCTASE)	DILATED CARDIOMYOPATHY, 1I, CMD1I SMITH-LEMLI-OPITZ SYNDROME, SLO	
DIA1 (CYTOCHROME b5 REDUCTASE, CYB5R)	METHEMOGLOBINEMIA DUE TO DEFICIENCY OF METHEMOGLOBIN REDUCTASE	
DJ1	PARKINSON DISEASE, TYPE 7, PARK7 (AUTOSOMAL RECESSIVE)	Whole Gene Sequencing or Deletions - Duplications
DKC1 (DYSKERIN)	DYSKERATOSIS CONGENITA (X-LINKED), DKC » ZINSSER-COLE-ENGMAN SYNDROME HOYERAAL-HREIDARSSON SYNDROME	
DLG3	MENTAL RETARDATION, NONSPECIFIC (X-LINKED)	See also Mental Retardation Panel
DLL3	SPONDYLOCOSTAL DYSOSTOSIS, AUTOSOMAL RECESSIVE 1, SCDO1	
DMD (DYSTROPHIN)	DUCHENNE MUSCULAR DYSTROPHY, DMD » MUSCULAR DYSTROPHY, DUCHENNE MUSCULAR DYSTROPHY	Whole Gene or Deletions-Duplications (MLPA)
	BECKER MUSCULAR DYSTROPHY, BMD » MUSCULAR DYSTROPHY, BECKER MUSCULAR DYSTROPHY	Whole Gene or Deletions-Duplications (MLPA)
	DILATED CARDIOMYOPATHY (X-LINKED)	Whole Gene or Deletions-Duplications (MLPA)
DMPK (DM KINASE, MYOTONIN)	MYOTONIC DYSTROPHY, TYPE 1 » STEINERT DISEASE	Repeat

DRD2 (DOPAMINE RECEPTOR D2)	MYOCLONIC DYSTONIA » DYSTONIA, ALCOHOL-RESPONSIVE » MYOCLONUS, HEREDITARY ESSENTIAL » DYSTONIA 11, DYT11	
DRPLA (ATROPHIN 1)	DENTATORUBRAL-PALLIDOLUYSIAN ATROPHY, DRPLA HAW RIVER SYNDROME, HRS	Repeat Repeat
DYSF (DYSFERLIN)	MUSCULAR DYSTROPHY, LIMB-GIRDLE, TYPE 2B, LGMD2B MIYOSHI MYOPATHY » MUSCULAR DYSTROPHY, DISTAL, LATE-ONSET (AUTOSOMAL RECESSIVE) MYOPATHY, DISTAL, WITH ANTERIOR TIBIAL ONSET	
DYT1 (TORSIN A)	DYSTONIA MUSCULORUM DEFORMANS » TORSION DYSTONIA, EARLY ONSET, DYT1	GAG Deletion or Whole Gene
EBP (EMOPAMIL-BINDING PROTEIN, 3-BETA-HYDROXYSTEROID-DELTA- 8, DELTA-7 ISOMERASE)	CHONDRODYSPLASIA PUNCTATA 2 (X-LINKED DOMINANT), CDPX2 » CONRADI-HUNERMANN SYNDROME » HAPPLE SYNDROME	
ECGF1 (ENDOTHELIAL CELL GROWTH FACTOR, PLATELET-DERIVED, THYMIDINE PHOSPHORYLASE, GLIOSTATIN)	MITOCHONDRIAL NEUROGASTROINTESTINAL ENCEPHALOPATHY SYNDROME, MNGIE » MYONEUROGASTROINTESTINAL ENCEPHALOPATHY SYNDROME » POLYNEUROPATHY, OPHTHALMOPLÉGIA, LEUKOENCEPHALOPATHY, AND INTESTINAL PSEUDOObSTRUCTION » POLIP SYNDROME » MNGIE WITHOUT LEUKOENCEPHALOPATHY	
ED1 (ECTODYSPLASIN A, EDA)	ECTODERMAL DYSPLASIA, HYPO-ANHIDROTIC (X- LINKED), ED1	
EDAR (ECTODYSPLASIN 1)	ANHIDROTIC ECTODERMAL DYSPLASIA 3, ED3 » HYPOHIDROTIC ECTODERMAL DYSPLASIA (AUTOSOMAL DOMINANT), EDA3 » ECTODERMAL DYSPLASIA, ANHIDROTIC, TYPE 3 HYPOHIDROTIC ECTODERMAL DYSPLASIA (AUTOSOMAL RECESSIVE) » ECTODERMAL DYSPLASIA, HYPOHIDROTIC (AUTOSOMAL RECESSIVE)	

EFNB1 (EPHRIN B1, EPLG2, LERK2, EFL3)	CRANIOFRONTONASAL SYNDROME, CFNS » CRANIOFRONTONASAL DYSOSTOSIS	
EGR2 (EARLY GROWTH RESPONSE 2)	CHARCOT-MARIE-TOOTH DISEASE, AXONAL, TYPE 1D, CMT1D CHARCOT-MARIE-TOOTH DISEASE, DEMYELINATING, TYPE 4E, CMT4E » CONGENITAL HYPOMYELINATING NEUROPATHY CHARCOT-MARIE-TOOTH DISEASE, DEMYELINATING, TYPE 4F, CMT4F » DEJERINE-SOTTAS HYPERTROPHIC NEUROPATHY » HEREDITARY MOTOR AND SENSORY NEUROPATHY TYPE 3, HMSN3	
EIF2AK3 (EUKARYOTIC TRANSLATION INITIATION FACTOR 2-ALPHA KINASE 3)	EPIPHYSEAL DYSPLASIA, MULTIPLE, WITH EARLY-ONSET DIABETES MELLITUS » MED-IDDM SYNDROME » IDDM-MED SYNDROME » WOLCOTT-RALLISON SYNDROME	
ELA2 (ELASTASE 2)	CYCLIC HEMATOPOIESIS » CYCLIC NEUTROPENIA NEUTROPENIA, SEVERE CONGENITAL, SCN » CONGENITAL NEUTROPENIA » INFANTILE AGRANULOCYTOSIS » KOSTMANN DISEASE	
ELN (ELASTIN)	SUPRAVALVAR AORTIC STENOSIS, EISENBERG TYPE CUTIS LAXA (AUTOSOMAL DOMINANT)	Sequencing, MLPA or Both Sequencing, MLPA or Both
EMERIN (EMD)	EMERY-DREYFUSS MUSCULAR DYSTROPHY (X-LINKED), EDMD » MUSCULAR DYSTROPHY, EMERY-DREYFUSS MUSCULAR DYSTROPHY (X-LINKED), EDMD	
EMX2 (EMPTY SPIRACLES, DROSOPHILA, 2, HOMOLOG OF)	SCHIZENCEPHALY	
ENG (ENDOGLIN)	TELANGIECTASIA, HEREDITARY HEMORRHAGIC, OF RENDU, OSLER, AND WEBER, TYPE 1 » OSLER-RENDU-WEBER DISEASE, TYPE 1 » ORW DISEASE, TYPE 1	
ENG, ALK1 and SMAD4	TELANGIECTASIA, HEREDITARY HEMORRHAGIC, OF RENDU, OSLER, AND WEBER » OSLER-RENDU-WEBER DISEASE » ORW DISEASE	

EPM2A (LAFORIN)	MYOCLONIC EPILEPSY OF LAFORA » LAFORA DISEASE » EPILEPSY, PROGRESSIVE MYOCLONIC TYPE 2	
ERCC6 (CSB)	COCKAYNE SYNDROME, TYPE 2 » COCKAYNE SYNDROME, TYPE B	
EVC	ELLIS-VAN CREVELD SYNDROME	
EVC2	ELLIS-VAN CREVELD SYNDROME	
EXT1 (EXOSTOSIN 1)	MULTIPLE EXOSTOSES, TYPE 1, EXT1, HME	
EXT2 (EXOSTOSIN 2)	MULTIPLE EXOSTOSES, TYPE 2, EXT2, HME	
EYA1	BRANCHIO-OTO-RENAL SYNDROME, BOR	
F7 (FACTOR 7, COAGULATION FACTOR 7)	FACTOR 7 DEFICIENCY » HYPOPROCONVERTINEMIA	
F8 (FACTOR 8)	HAEMOPHILIA A	Whole Gene or Intron 22 Inversion
F9 (FACTOR 9)	HAEMOPHILIA B	
F11 (FACTOR 11, COAGULATION FACTOR 11)	PTA DEFICIENCY » COAGULATION FACTOR 11 DEFICIENCY » ROSENTHAL SYNDROME	
FACL4 (FATTY ACID COA LIGASE, LONG CHAIN 4)	MENTAL RETARDATION, NONSPECIFIC (X-LINKED), TYPE 63, MRX63 MENTAL RETARDATION, NONSPECIFIC (X-LINKED), TYPE 68, MRX68	See also Mental Retardation Panel
FAH (FUMARYLACETOACETATE HYDROLASE, FUMARYLACETOACETASE)	TYROSINEMIA, TYPE 1 » HEPATORENAL TYROSINEMIA » FUMARYLACETOACETASE DEFICIENCY » FAH DEFICIENCY	5 Exons (Most Common Mutations)
FALDH	SJOGREN-LARSSON SYNDROME » FATTY ALDEHYDE DEHYDROGENASE DEFICIENCY	
FANCA	FANCONI ANEMIA » FANCONI PANCYTOPENIA	Whole Gene, 14 Exons: Exons 1, 6, 7, 8, 11, 15, 16, 17, 19, 20, 32, 39, 42, 43, representing 25% of reported mutations or 8 Exons: Exons 13, 27, 29, 34-38, representing 67% of reported mutations

FANCC	FANCONI ANEMIA, COMPLEMENTATION GROUP C » FANCONI PANCYTOPENIA, TYPE 3 » (See also Molecular Screening Tests)	1 Mutation: IVS4+4A-G
FBLN4 (FIBULIN 4, EGF-CONTAINING FIBULIN-LIKE EXTRACELLULAR MATRIX PROTEIN 2, EFEMP2)	CUTIS LAXA (AUTOSOMAL RECESSIVE)	
FBLN5 (FIBULIN 5)	CUTIS LAXA (AUTOSOMAL RECESSIVE) CUTIS LAXA (AUTOSOMAL DOMINANT)	
FBN1 (FIBRILLIN1)	MARFAN SYNDROME, TYPE 1, MFS MASS PHENOTYPE » MASS SYNDROME ECTOPIA LENTIS FAMILIAL AORTIC ANEURYSM SHPHRINTZEN–GOLDBERG CRANIOSYNOSTOSIS SYNDROME	At least 20mg DNA is needed
FBN2 (FIBRILLIN 2)	CONTRACTURAL CONGENITAL ARACHNODACTYLY, CCA » BEALS SYNDROME	15 exons (exons 15, 22 -33 , 35-36)
FGA (FIBRINOGEN ALPHA) FGB (FIBRINOGEN BETA) FGD1	AFIBRINOGENEMIA » DYSFIBRINOGENEMIA AFIBRINOGENEMIA » DYSFIBRINOGENEMIA AARSKOG SYNDROME » FACIODIGITOGENITAL SYNDROME	
FGF14 (FIBROBLAST GROWTH FACTOR 14) FGF23 (FIBROBLAST GROWTH FACTOR 23)	SPINOCEREBELLAR ATAXIA 27, SCA27 » CEREBELLAR ATAXIA (AUTOSOMAL DOMINANT), FGF14-RELATED HYPOPHOSPHATEMIC RICKETS (AUTOSOMAL DOMINANT) » RICKETS, HYPOPHOSPHATEMIC RICKETS (AUTOSOMAL DOMINANT)	
FGFR1	KALLMANN SYNDROME TYPE 2, KAL2 » HYPOGONADOTROPIC HYPOGONADISM AND ANOSMIA JACKSON-WEISS SYNDROME » CRANIOSYNOSTOSIS, JACKSON-WEISS SYNDROME PFEIFFER SYNDROME » CRANIOSYNOSTOSIS, PFEIFFER SYNDROME	Whole Gene Selected exons. Also includes testing of selected exons of FGFR2
FGFR2	APERT SYNDROME » CRANIOSYNOSTOSIS, APERT	2 Mutations: S252W, P253R

	<p>SYNDROME BEARE-STEVENSON CUTIS GYRATA » CRANIOSYNOSTOSIS, BEARE-STEVENSON CUTIS GYRATA CROUZON SYNDROME » CRANIOSYNOSTOSIS, CROUZON SYNDROME JACKSON-WEISS SYNDROME » CRANIOSYNOSTOSIS, JACKSON-WEISS SYNDROME SAETHRE-CHOTZEN SYNDROME » CRANIOSYNOSTOSIS, SAETHRE-CHOTZEN SYNDROME PFEIFFER SYNDROME » CRANIOSYNOSTOSIS, PFEIFFER SYNDROME ACHONDROPLASIA HYPOCHONDROPLASIA</p>	<p>Selected exons. Also includes testing of selected exons of FGFR2 2 Mutations: G380R, G375C 7 Mutations: I538V, N540T, N540S, N540K, K650N, K650M, K650Q</p>
FGFR3	<p>NONSYNDROMIC CORONAL CRANIOSYNOSTOSIS (MUENKE) » CRANIOSYNOSTOSIS, NONSYNDROMIC CORONAL CRANIOSYNOSTOSIS SAETHRE-CHOTZEN SYNDROME » CRANIOSYNOSTOSIS, SAETHRE-CHOTZEN SYNDROME THANATOPHORIC DYSPLASIA, TYPE 1 SADDAN DYSPLASIA » CRANIOSYNOSTOSIS, SADDAN DYSPLASIA CROUZON CRANIOSYNOSTOSIS WITH ACANTHOSIS NIGRICANS » CROUZONO DERMO SKELETAL SYNDROME » CRANIOSYNOSTOSIS, CROUZON CRANIOSYNOSTOSIS WITH ACANTHOSIS NIGRICANS AFIBRINOGENEMIA » DYSFIBRINOGENEMIA FUMARASE DEFICIENCY » FUMARIC ACIDURIA</p>	
FGG (FIBRINOGEN GAMMA) FH (FUMARATE HYDRATASE, FUMARASE)	<p>MULTIPLE CUTANEOUS AND UTERINE LEIOMYOMATA 1, MCUL1 HEREDITARY LEIOMYOMATOSIS AND RENAL CELL CANCER, HLRCC</p>	<p>Preferentially on skin fibroblast culture for mutation analysis in proband, eventually blood in PAX RNA tubes</p>
FKRP (FUKUTIN-RELATED PROTEIN)	<p>MUSCULAR DYSTROPHY, CONGENITAL, TYPE 1C, MDC1C</p>	

FLCN (FOLLICULIN, FLCL)	<p>MUSCULAR DYSTROPHY, LIMB-GIRDLE, TYPE 2I, LGMD2I BIRT-HOGG-DUBE SYNDROME, BHD » FIBROFOLLICULOMAS WITH TRICHODISCOMAS AND ACROCHORDONS</p>	
FLG (FILAGGRIN, PROFILAGGRIN)	<p>PNEUMOTHORAX, PRIMARY SPONTANEOUS ICHTHYOSIS VULGARIS » ICHTHYOSIS SIMPLEX</p> <p>ATOPIC DERMATITIS</p> <p>ASTHMA</p>	<p>2 Common Mutations: c.2282delCAGT and c.1501C>T (p.Arg501X) 2 Common Mutations: c.2282delCAGT and c.1501C>T (p.Arg501X) 2 Common Mutations: c.2282delCAGT and c.1501C>T (p.Arg501X)</p>
FLNA (FILAMIN A)	<p>HETEROTOPIA, PERIVENTRICULAR (X-LINKED DOMINANT) » PERIVENTRICULAR NODULAR HETEROTOPIA 1 » NODULAR HETEROTOPIA » BILATERAL PERIVENTRICULAR HETEROTOPIA » PERIVENTRICULAR NODULAR, WITH FRONTOMETAPHYSEAL DYSPLASIA OTOPALATODIGITAL SYNDROME, TYPE 1, OPD1 OTOPALATODIGITAL SYNDROME, TYPE 2, OPD2 » CRANIOORODIGITAL SYNDROME » FACIOPALATOOSSEOUS SYNDROME FRONTOMETAPHYSEAL DYSPLASIA MELNICK-NEEDLES SYNDROME » OSTEODYSPLASTY OF MELNICK AND NEEDLES HETEROTOPIA, PERIVENTRICULAR, EHLERS-DANLOS VARIANT » PERIVENTRICULAR NODULAR HETEROTOPIA 4</p>	<p>Exon 3 Exons 3, 5, 11, 22, 29, 45</p> <p>Exons 22, 29 Exon 22</p>
FLNB (FILAMIN B)	<p>SPONDYLOCARPOTARSAL SYNOSTOSIS SYNDROME » SYNSPONDYLISM, CONGENITAL LARSEN SYNDROME (AUTOSOMAL DOMINANT) ATELOSTEOGENESIS, TYPE 1 » GIANT CELL CHONDRODYSPLASIA » SPONDYLOHUMEROFEMORAL HYPOPLASIA ATELOSTEOGENESIS, TYPE 3 BOOMERANG DYSPLASIA</p>	

FLT4 (FMS-LIKE TYROSINE KINASE 4, VASCULAR ENDOTHELIAL GROWTH FACTOR RECEPTOR 3, VEGFR3)	LYMPHEDEMA, HEREDITARY, TYPE 1 » NONNE-MILROY LYMPHEDEMA » MILROY DISEASE » PRIMARY CONGENITAL LYMPHEDEMA	Whole Gene or 8 Exons: Exons 18-25, representing 90% of reported mutations
FMO3 (FLAVIN - CONTAINING MONOOXYGENASE 3)	FISH-ODOR SYNDROME » TRIMETHYLAMINURIA	
FMR1	FRAGILE X SYNDROME, FRAXA	Repeat - At least 20mg DNA with a concentration higher than 300 nanogram per microliter is needed
FMR2	FRAGILE E SYNDROME, FRAXE	Repeat
FOXC1 (FORKHEAD BOX C1, FORKHEAD, DROSOPHILA, HOMOLOG-LIKE 7, FKHL7)	RIEGER SYNDROME, TYPE 1, RIEG1	Whole Gene Sequencing or Deletion-Duplication
FORKHEAD-RELATED ACTIVATOR 3, FREAC3)	IRIDOGONIODYSGENESIS SYNDROME, TYPE 2, IRID2 » IRIS HYPOPLASIA WITH EARLY-ONSET GLAUCOMA (AUTOSOMAL DOMINANT), IHGA PETERS ANOMALY	Whole Gene Sequencing or Deletion-Duplication
	ANTERIOR SEGMENT MESENCHYMAL DYSGENESIS	Whole Gene Sequencing or Deletion-Duplication
FOXC2 (FORKHEAD BOX C2)	LYMPHEDEMA-DISTICHIASIS SYNDROME LYMPHEDEMA, HEREDITARY, TYPE 2 » MEIGE LYMPHEDEMA LYMPHEDEMA AND PTOSIS	
FOXE1 (FORKHEAD BOX E1, THYROID TRANSCRIPTION FACTOR 2, TTF2, TITF2)	BAMFORTH-LAZARUS SYNDROME » HYPOTHYROIDISM, ATHYROIDAL, WITH SPIKY HAIR AND CLEFT PALATE	
FOXL2	BLEPHAROPHIMOSIS, PTOSIS, AND EPICANTHUS INVERSUS, BPES	

FOXP3 (FORKHEAD BOX P3,
SCURFIN)

IMMUNODYSREGULATION, POLYENDOCRINOPATHY,
AND ENTEROPATHY (X-LINKED), IPEX » X-LINKED
AUTOIMMUNITY-ALLERGIC DYSREGULATION
SYNDROME » IDDM-SECRETORY DIARRHEA
SYNDROME » AUTOIMMUNITY-IMMUNODEFICIENCY
SYNDROME » DIARRHEA, POLYENDOCRINOPATHY,
FATAL INFECTION SYNDROME » ENTEROPATHY,
AUTOIMMUNE, WITH HEMOLYTIC ANEMIA AND
POLYENDOCRINOPATHY » POLYENDOCRINOPATHY,
IMMUNE DYSFUNCTION, AND DIARRHEA » DIABETES
MELLITUS, CONGENITAL INSULIN-DEPENDENT, WITH
FATAL SECRETORY DIARRHEA » IMMUNODEFICIENCY,
POLYENDOCRINOPATHY, AND ENTEROPATHY

FRAXF (FRAGILE SITE F)

FRAGILE F SYNDROME

Repeat

FRDA (FRATAXINE)

FRIEDREICH ATAXIA, FRDA

Repeat or 5 Exons: 1-5

FREM2 (FRAS1-RELATED
EXTRACELLULAR MATRIX PROTEIN 2)

FRASER SYNDROME » CRYPTOPHTHALMOS WITH
OTHER MALFORMATIONS » CRYPTOPHTHALMOS-
SYNDACTYLY SYNDROME

Exon 6

FSHD

FACIOSCAPULOHUMERAL MUSCULAR DYSTROPHY,
FSHD » LANDOUZY-DEJERINE MUSCULAR DYSTROPHY
» MUSCULAR DYSTROPHY, FACIOSCAPULOHUMERAL
MUSCULAR DYSTROPHY, FSHD » MUSCULAR
DYSTROPHY, LANDOUZY-DEJERINE MUSCULAR
DYSTROPHY

Repeat At least 10ml EDTA Blood

FTL (FERRITIN LIGHT CHAIN)

HYPERFERRITINEMIA-CATARACT SYNDROME
BASAL GANGLIA DISEASE, ADULT-ONSET »
NEUROFERRITINOPATHY

FTSJ1 (FTSJ HOMOLOG 1)

MENTAL RETARDATION, NONSPECIFIC (X-LINKED),
TYPE 44, MRX44
MENTAL RETARDATION, NONSPECIFIC (X-LINKED),
TYPE 9, MRX9

See also Mental Retardation Panel

FUCA1 (ALPHA-L-FUCOSIDASE)	FUCOSIDOSIS » ALPHA-L-FUCOSIDASE DEFICIENCY	
FXRD2 (FXRD DOMAIN-CONTAINING ION TRANSPORT REGULATOR 2, SODIUM-POTASSIUM-ATPase, GAMMA-1 POLYPEPTIDE)	HYPOMAGNESEMIA, TYPE 2, RENAL	
FY (DUFFY BLOOD GROUP SYSTEM)	HEMOLYTIC DISEASE, NEWBORN	1 Mutation: G44D
G6PC (GLUCOSE-6-PHOSPHATASE)	GLYCOGEN STORAGE DISEASE, TYPE 1A » GSD TYPE 1A » VON GIERKE SYNDROME	
G6PD (GLUCOSE-6-PHOSPHATE DEHYDROGENASE)	G6PD DEFICIENCY » GLUCOSE-6-PHOSPHATE DEHYDROGENASE DEFICIENCY	
G6PT1 (GLUCOSE-6-PHOSPHATE TRANSPORTER 1, GLUCOSE-6-PHOSPHATE TRANSLOCASE)	GLYCOGEN STORAGE DISEASE, TYPE 1B » GSD TYPE 1B	
GAA (GLUCOSIDASE, ALPHA, ACID, ACID MALTASE)	GLYCOGEN STORAGE DISEASE, TYPE 2 » GSD TYPE 2 » ACID ALPHA-GLUCOSIDASE DEFICIENCY » POMPE DISEASE » GLYCOGENOSIS, GENERALIZED, CARDIAC FORM » CARDIOMEGALIA GLYCOGENICA DIFFUSA » ACID MALTASE DEFICIENCY » ALPHA-1,4-GLUCOSIDASE DEFICIENCY	
GABRG2 (GAMMA-AMINO BUTYRIC ACID RECEPTOR, GAMMA-2)	GENERALIZED EPILEPSY WITH FEBRILE SEIZURES PLUS, GEFS+ » GENERALIZED EPILEPSY WITH FEBRILE SEIZURES PLUS, TYPE 2, GEFS+, TYPE 2 » FEBRILE SEIZURES ASSOCIATED WITH AFEBRILE SEIZURES EPILEPSY, CHILDHOOD ABSENCE, 2	
GALC (GALACTOSYL CERAMIDASE, GALACTOCEREBROSIDASE)	KRABBE DISEASE » GLOBOID CELL LEUKODYSTROPHY » GALACTOSYLCERAMIDE BETA-GALACTOSIDASE DEFICIENCY » GALACTOCEREBROSIDASE DEFICIENCY	

GALK1 (GALACTOKINASE 1)	GALACTOKINASE DEFICIENCY » GALK DEFICIENCY » GALACTOSEMIA TYPE 2 » GALACTOKINASE DEFICIENCY WITH CATARACT	
GALT (GALACTOSE-1-PHOSPHATE URIDYLTRANSFERASE)	GALACTOSEMIA TYPE 1 » GALACTOSE-1-PHOSPHATE URIDYLTRANSFERASE DEFICIENCY	Whole Gene or 1 Mutation: GLN188ARG
GARS (GLYCYL T RNA SYNTHETASE)	CHARCOT-MARIE-TOOTH DISEASE, AXONAL, TYPE 2D, CMT2D SPINAL MUSCULAR ATROPHY, DISTAL, TYPE 5, DSMA5	
GATA1 (GATA-BINDING PROTEIN 1; ERYTHROID TRANSCRIPTION FACTOR 1, GLOBIN TRANSCRIPTION FACTOR 1)	DYSERYTHROPOIETIC ANEMIA WITH THROMBOCYTOPENIA » MACROTHROMBOCYTOPENIA (X-LINKED) THROMBOCYTOPENIA, PLATELET DYSFUNCTION, HEMOLYSIS, AND IMBALANCED GLOBIN SYNTHESIS » THROMBOCYTOPENIA WITH BETA-THALASSEMIA (X-LINKED)	
GBA (ACID BETA- GLUCOSIDASE GLUCOCEROBROSIDASE)	GAUCHER DISEASE, TYPE 1 » (See also Molecular Screening Tests) GAUCHER DISEASE, TYPE 2 » (See also Molecular Screening Tests) GAUCHER DISEASE, TYPE 3 » (See also Molecular Screening Tests)	Whole Gene or 6 Mutations: 84GG, IVS2+1, N370S, 1297T, L444P, V394L Whole Gene or 6 Mutations: 84GG, IVS2+1, N370S, 1297T, L444P, V394L Whole Gene or 6 Mutations: 84GG, IVS2+1, N370S, 1297T, L444P, V394L
GBE1 (GLYCOGEN BRANCHING ENZYME)	GLYCOGEN STORAGE DISEASE, TYPE 4 » GSD TYPE 4 » GLYCOGEN BRANCHING ENZYME DEFICIENCY » GBE1 DEFICIENCY » ANDERSEN DISEASE » BRANCHER DEFICIENCY » GLYCOGENOSIS 4 » AMYLOPECTINOSIS POLYGLUCOSAN BODY DISEASE, ADULT FORM, APBD	
GCDH (GLUTARYL COA DEHYDROGENASE)	GLUTARICACIDURIA, TYPE 1	
GCH1 (GTP CYCLOHYDROLASE 1)	DYSTONIA, PROGRESSIVE, WITH DIURNAL VARIATION » DYSTONIA-PARKINSONISM WITH DIURNAL FLUCTUATION » DYSTONIA 5, DYT5 » SEGAWA SYNDROME (AUTOSOMAL DOMINANT) » DOPA-RESPONSIVE DYSTONIA, (AUTOSOMAL DOMINANT), DRD	

	GTP CYCLOHYDROLASE 1 DEFICIENCY » GCH DEFICIENCY » HYPERPHENYLALANINEMIA WITH NEOPTERIN DEFICIENCY » PHENYLKETONURIA, ATYPICAL SEVERE, DUE TO GTP CYCLOHYDROLASE 1 DEFICIENCY
	MYOCLONIC DYSTONIA » MYOCLONUS-DYSTONIA SYNDROME » DYSTONIA, ALCOHOL-RESPONSIVE » DYSTONIA 11, DYT11
GCK (GLUCOKINASE)	MATURITY-ONSET DIABETES OF THE YOUNG, TYPE 2, MODY2 HYPERINSULINEMIC HYPOGLYCEMIA, FAMILIAL, TYPE 3
GDAP1 (GANGLIOSIDE-INDUCED DIFFERENTIATION-ASSOCIATED PROTEIN 1)	CHARCOT-MARIE-TOOTH DISEASE, DEMYELINATING, TYPE 4A, CMT4A (AUTOSOMAL RECESSIVE) CHARCOT-MARIE-TOOTH DISEASE, AXONAL, WITH VOCAL CORD PARESIS (AUTOSOMAL RECESSIVE) CHARCOT-MARIE-TOOTH DISEASE, AXONAL, TYPE 2K, CMT2K CHARCOT-MARIE-TOOTH DISEASE, INTERMEDIATE A, CMTRIA (AUTOSOMAL RECESSIVE)
GDF5 (GROWTH / DIFFERENTIATION FACTOR 5, CDMP1, LAP4)	ACROMESOMELIC DYSPLASIA, HUNTER-THOMPSON TYPE BRACHYDACTYLY, TYPE C » BRACHYDACTYLY, HAWS TYPE CHONDRODYSPLASIA, GREBE TYPE » ACHONDROGENESIS, BRAZILIAN TYPE » ACROMESOMELIC DYSPLASIA, GREBE TYPE FIBULAR HYPOPLASIA AND COMPLEX BRACHYDACTYLY » DU PAN SYNDROME
GFAP (GLIAL FIBRILLARY ACIDIC PROTEIN)	ALEXANDER DISEASE
GGCX	VITAMIN K-DEPENDENT CLOTTING FACTORS, COMBINED DEFICIENCY OF, 1
GH1	PITUITARY DWARFISM 1 » PRIMORDIAL DWARFISM » SEXUAL ATELEIOTIC DWARFISM » GROWTH HORMONE DEFICIENCY, ISOLATED » AUTOSOMAL

	RECESSIVE ISOLATED GROWTH HORMONE DEFICIENCY, TYPE 1A / 1B » ILLIG-TYPE GROWTH HORMONE DEFICIENCY
	PITUITARY DWARFISM DUE TO ISOLATED GROWTH HORMONE DEFICIENCY (AUTOSOMAL DOMINANT) » GROWTH HORMONE DEFICIENCY, ISOLATED (AUTOSOMAL DOMINANT) » ISOLATED GROWTH HORMONE DEFICIENCY, TYPE 2
	PITUITARY DWARFISM 4 » PITUITARY DWARFISM WITH NORMAL IMMUNOREACTIVE GROWTH HORMONE AND LOW SOMATOMEDIN » BIODEFECTIVE GROWTH HORMONE » KOWARSKI SYNDROME
GHR (GROWTH HORMONE RECEPTOR, GROWTH HORMONE-BINDING PROTEIN, GHBP)	SHORT STATURE, IDIOPATHIC (AUTOSOMAL) PITUITARY DWARFISM 2 » GROWTH HORMONE INSENSITIVITY SYNDROME » LARON SYNDROME » GROWTH HORMONE RECEPTOR DEFICIENCY
GJA1 (GAP JUNCTION PROTEIN, ALPHA-1, CONNEXIN 43, CX43)	SHORT STATURE, IDIOPATHIC (AUTOSOMAL) OCULODENTODIGITAL DYSPLASIA » OCULODENTOOSSEOUS DYSPLASIA SYNDACTYLY, TYPE 3 » SYNDACTYLY OF FINGERS 4 AND 5
GJB1 (CONNEXIN 32, CX32)	HYPOPLASTIC LEFT HEART SYNDROME CHARCOT-MARIE-TOOTH PERONEAL MUSCULAR ATROPHY (X-LINKED), CMTX » HEREDITARY MOTOR AND SENSORY NEUROPATHY (X-LINKED)
GJB2 (CONNEXIN 26, CX26)	DEAFNESS, DFNA3 » DEAFNESS, (AUTOSOMAL DOMINANT), NONSYNDROMIC SENSORINEURAL 3 DEAFNESS, DFNB1 » DEAFNESS, (AUTOSOMAL RECESSIVE), NEUROSENSORY 1 PALMOPLANTAR KERATODERMA WITH DEAFNESS » HYPERKERATOSIS, PALMOPLANTAR KERATODERMA WITH DEAFNESS VOHWINKEL SYNDROME KERATITIS-ICHTHYOSIS-DEAFNESS

GJB3 (CONNEXIN 31, CX31)	ERYTHROKERATODERMIA VARIABILIS, EKV	Whole Gene
	DEAFNESS, DFNA2 » DEAFNESS, (AUTOSOMAL DOMINANT), NONSYNDROMIC SENSORINEURAL 2	Whole Gene
GJB4 (CONNEXIN 30.3, CX30.3)	ERYTHROKERATODERMIA VARIABILIS, EKV	
GJB6 (CONNEXIN 30, CX30)	DEAFNESS, DFNB1 » DEAFNESS, (AUTOSOMAL RECESSIVE), NEUROSENSORY 1	Whole Gene
	CLOUSTON SYNDROME » HYDROTIC ECTODERMAL DYSPLASIA, ED2 » ECTODERMAL DYSPLASIA, CLOUSTON SYNDROME	Whole Gene
	DEAFNESS, DFNA3 » DEAFNESS, (AUTOSOMAL DOMINANT), NONSYNDROMIC SENSORINEURAL 3	Whole Gene
GLA (GALACTOSIDASE)	FABRY DISEASE	
GLB1 (GALACTOSIDASE, BETA-1)	BETA-GALACTOSIDASE-1 DEFICIENCY, GLB1 DEFICIENCY » GANGLIOSIDOSIS, GENERALIZED GM1, TYPE 1 » MORQUIO DISEASE, TYPE B » MUCOPOLYSACCHARIDOSIS TYPE 4B, MPS 4B	
GLDC (GLYCINE DECARBOXYLASE, GLYCINE CLEAVAGE SYSTEM P PROTEIN)	NONKETOTIC HYPERGLYCINEMIA » GLYCINE ENCEPHALOPATHY	
GLI3 (GLI-KRUPPEL FAMILY MEMBER 3)	PALLISTER-HALL SYNDROME » HYPOTHALAMIC HAMARTOBLASTOMA, HYPOPITUITARISM, IMPERFORATE ANUS, AND POSTAXIAL POLYDACTYLY	Deletions
	GREIG CEPHALOPOLYSYNDACTYLY SYNDROME » POLYSYNDACTYLY WITH PECULIAR SKULL SHAPE	Deletions
	POLYDACTYLY, POSTAXIAL, TYPE A1	Deletions
	POLYDACTYLY, PREAXIAL 4 » POLYSYNDACTYLY, UNCOMPLICATED CROSSED » POLYDACTYLY, TYPE 1	Deletions
	ACROCALLOSAL SYNDROME » SCHINZEL ACROCALLOSAL SYNDROME	Deletions
GLOBIN	See HBB	
GLRA1 (GLYCIN RECEPTOR)	HYPEREKPLEXIA » STARTLE DISEASE » KOK DISEASE » STIFF BABY SYNDROME	

GLUD1 (GLUTAMATE DEHYDROGENASE 1)	HYPERINSULINEMIC HYPOGLYCEMIA, FAMILIAL, TYPE 6 » HYPERINSULINISM-HYPERAMMONEMIA SYNDROME » PERSISTENT HYPERINSULINEMIC HYPOGLYCEMIA OF INFANCY » HYPOGLYCEMIA, HYPERINSULINEMIC, OF INFANCY » HYPERINSULINEMIC HYPOGLYCEMIA DUE TO FOCAL ADENOMATOUS HYPERPLASIA » NESIDIOBLASTOSIS OF PANCREAS » HYPERINSULINISM, FAMILIAL, WITH PANCREATIC NESIDIOBLASTOSIS » HYPERINSULINISM, CONGENITAL ONEMIA SYNDROME606762Exons 6, 7, 10,	Exons 6, 7, 10, 11 and 12
GM2A (GM2-ACTIVATOR)	TAY-SACHS DISEASE, AB VARIANT » HEXOSAMINIDASE ACTIVATOR DEFICIENCY » GM2-GANGLIOSIDOSIS, TYPE AB » AB VARIANT GM2-GANGLIOSIDOSIS	
GNAS (GNAS1, ALPHA SUBUNIT OF Gs, ALPHA SUBUNIT OF ADENYLATE CYCLASE STIMULATORY PROTEIN)	ALBRIGHT HEREDITARY OSTEODYSTROPHY » PSEUDOHYPOPARATHYROIDISM, TYPE 1A » PSEUDOHYPOPARATHYROIDISM, TYPE 1C » PSEUDOPSEUDOHYPOPARATHYROIDISM MCCUNE-ALBRIGHT SYNDROME » ALBRIGHT SYNDROME » POLYOSTOTIC FIBROUS DYSPLASIA OSSEOUS HETEROPLASIA, PROGRESSIVE » ECTOPIC OSSIFICATION, FAMILIAL » OSTEOMA CUTIS PSEUDOHYPOPARATHYROIDISM, TYPE 1B PITUITARY TUMOR, SOMATOTROPHINOMA	Whole Gene or 2 Common Mutations: R201H and R201C Methylation
GNE (GLCNE, UDP-N-ACETYLGLUCOSAMINE 2-EPIMERASE/N-ACETYLMANNOSAMINE KINASE)	NONAKA DISTAL MYOPATHY » MYOPATHY, DISTAL, WITH RIMMED VACUOLES SIALURIA INCLUSION BODY MYOPATHY 2 (AUTOSOMAL RECESSIVE), IBM2	Whole Gene or 1 Mutation Whole Gene or 1 Mutation Whole Gene or 1 Mutation
GP1BA (GLYCOPROTEIN Ib, PLATELET, ALPHA POLYPEPTIDE, GLYCOCALICIN)	BERNARD-SOULIER SYNDROME » GIANT PLATELET SYNDROME » VON WILLEBRAND FACTOR RECEPTOR DEFICIENCY PSEUDO-VON WILLEBRAND DISEASE » VON	

GP1BB (GLYCOPROTEIN Ib, PLATELET, BETA POLYPEPTIDE)	WILLEBRAND DISEASE, PLATELET-TYPE BERNARD-SOULIER SYNDROME »GIANT PLATELET SYNDROME » PLATELET GLYCOPROTEIN Ib DEFICIENCY » GLYCOPROTEIN Ib, PLATELET, DEFICIENCY OF » VON WILLEBRAND FACTOR RECEPTOR DEFICIENCY » MACROTHROMBOCYTOPENIA, FAMILIAL, BERNARD-SOULIER TYPE	
GP9 (GLYCOPROTEIN IX, PLATELET)	BERNARD-SOULIER SYNDROME » GIANT PLATELET SYNDROME » VON WILLEBRAND FACTOR RECEPTOR DEFICIENCY	
GPC3 (GLYPICAN 3)	SIMPSON-GOLABI-BEHMEL SYNDROME, SGBS	Deletion Analysis or Deletion Analysis + Whole Gene Analysis
GRHPR (GLYOXYLATE REDUCTASE / HYDROXYPYRUVATE REDUCTASE, GLXR)	HYPEROXALURIA, PRIMARY, TYPE 2 » OXALOSIS 2 » GLYCERIC ACIDURIA » GLYOXYLATE REDUCTASE/HYDROXYPYRUVATE REDUCTASE DEFICIENCY » D-GLYCERATE DEHYDROGENASE DEFICIENCY	Whole Gene or 2 Mutations: 103delG and c.403_405+2delAAGT
GRN (GRANULIN, PROGRANULIN, EPITHELIN)	FRONTOTEMPORAL DEMENTIA, UBIQUITIN-POSITIVE » DEMENTIA, HEREDITARY DYSPHASIC DISINHIBITION	Whole Gene or Deletions
GYS2 (GLYCOGEN SYNTHASE 2)	GLYCOGEN STORAGE DISEASE, TYPE 0 » GSD TYPE 0 » LIVER GLYCOGEN SYNTHASE DEFICIENCY	
HADH (3-@HYDROXYACYL-CoA DEHYDROGENASE, HADSC, SCHAD)	3-@HYDROXYACYL-CoA DEHYDROGENASE DEFICIENCY » HADH DEFICIENCY » SCHAD DEFICIENCY	

HADH2 (HYDROXYACYL-CoA DEHYDROGENASE TYPE 2, AMYLOID BETA-BINDING POLYPEPTIDE, ERAB, 2-ALPHA-METHYL-3-HYDROXYBUTYRYL-CoA DEHYDROGENASE)	2-ALPHA-METHYL-3-HYDROXYBUTYRYL-CoA DEHYDROGENASE DEFICIENCY	
HADHA (HYDROXYACYL-CoA DEHYDROGENASE/3-KETOACYL-CoA THIOLASE/ENOYL-CoA HYDRATASE, ALPHA SUBUNIT, TRIFUNCTIONAL PROTEIN, ALPHA SUBUNIT MITOCHONDRIAL TRIFUNCTIONAL PROTEIN, ALPHA SUBUNIT, LONG-CHAIN HYDROXYACYL-CoA DEHYDROGENASE, LCHAD)	TRIFUNCTIONAL PROTEIN DEFICIENCY LONG-CHAIN 3-HYDROXYACYL-CoA DEHYDROGENASE DEFICIENCY, LCHAD DEFICIENCY NEUROPATHY-MYOPATHY, PROGRESSIVE, DUE TO TRIFUNCTIONAL PROTEIN DEFICIENCY	
HADHB (HYDROXYACYL-CoA DEHYDROGENASE/3-KETOACYL-CoA THIOLASE/ENOYL-CoA HYDRATASE, BETA SUBUNIT, TRIFUNCTIONAL PROTEIN, BETA SUBUNIT MITOCHONDRIAL TRIFUNCTIONAL PROTEIN, BETA SUBUNIT HYDROXYACYL-CoA DEHYDROGENASE, HADH)	TRIFUNCTIONAL PROTEIN DEFICIENCY	
HAMP (HEPCIDIN ANTIMICROBIAL PEPTIDE)	HEMOCHROMATOSIS, JUVENILE » HEMOCHROMATOSIS, TYPE 2, HFE2 » HEMOCHROMATOSIS DUE TO DEFECT IN HEMOJUVELIN	
HBA 1 and HBA 2 (ALPHA GLOBIN)	ALPHA HAEMOGLOBINOPATHIA » ALPHA THALASSEMIA	Whole Gene (both HBA1 and HBA2) or Deletion Analysis (MLPA)
HBB (BETA GLOBIN)	BETA HAEMOGLOBINOPATHIA » BETA THALASSEMIA SICKLE CELL ANEMIA	
HD (HUNTINGTIN)	HUNTINGTON CHOREA	Repeat

HERG	LONG QT SYNDROME 2, LQT2 » ROMANO-WARD SYNDROME	See also LONG QT PANEL 1 and LONG QT PANEL 2 and LONG QT PANEL 3
HESX1 (HOMEODOMAIN GENE EXPRESSED IN ES CELLS)	SEPTOOPTIC DYSPLASIA » DE MORSIER SYNDROME » PITUITARY HORMONE DEFICIENCY PITUITARY DWARFISM 3 » PANHYPOPITUITARISM » ATELIOTIC DWARFISM WITH HYPOGONADISM » HANHART DWARFISM	
HEXA (HEXOSAMINIDASE)	TAY-SACHS DISEASE »GANGLIOSIDOSIS, GENERALIZED GM2, TYPE 1 » HEXOSAMINIDASE A DEFICIENCY » (See also Molecular Screening Tests)	Whole Gene or 3 Mutations: 1277insTATC, IVS12+1G-C, G269S
HEXB (HEXOSAMINIDASE B)	SANDHOFF DISEASE » GM2-GANGLIOSIDOSIS, TYPE 2 » HEXOSAMINIDASES A AND B DEFICIENCY	
HFE	HEMOCHROMATOSIS, TYPE 1, HFE1	Whole Gene or 4 Mutations (C282Y, H63D, S65C, E168X)
HLXB9 (HOMEODOMAIN GENE HB9)	CURRARINO TRIAD » SACRAL AGENESIS SYNDROME » SACRAL AGENESIS, HEREDITARY, WITH PRESACRAL MASS, ANTERIOR MENINGOCELE, AND/OR TERATOMA, AND ANORECTAL MALFORMATION	
HMBS (HYDROXYMETHYLBILANE SYNTHASE)	PORPHYRIA, ACUTE INTERMITTENT » PORPHOBILINOGEN DEAMINASE DEFICIENCY » PBGD DEFICIENCY » UROPORPHYRINOGEN SYNTHASE DEFICIENCY	
HMGCL (HMG - COA SYNTHETASE)	HMG-COA LYASE DEFICIENCY » HMG-COA SYNTHETASE DEFICIENCY	
HNF1A (TCF1)	MATURITY-ONSET DIABETES OF THE YOUNG, TYPE 3, MODY3 HEPATIC ADENOMA	
HNF1B (HNF2, TCF2)	MATURITY-ONSET DIABETES OF THE YOUNG, TYPE 5, MODY5 HYPOPLASTIC GLOMERULOCYSTIC KIDNEY DISEASE	
HNF4A	MATURITY-ONSET DIABETES OF THE YOUNG, TYPE 1, MODY1	

HOXA 13	HAND-FOOT-UTERUS SYNDROME » HAND-FOOT-GENITAL SYNDROME GUTTMACHER SYNDROME	
HOXD 13 (HOMEODOMAIN D13)	BRACHYDACTYLY TYPE E, BDE BRACHYDACTYLY TYPE D, BDD SYNDACTYLY, TYPE 2 » SYNPOLYDACTYLY	
HPRT1 (HYPOXANTHINE GUANINE PHOSPHORIBOSYL TRANSFERASE 1, HGPRT)	LESCH - NYHAN SYNDROME, LNS » HYPOXANTHINE GUANINE PHOSPHORIBOSYLTRANSFERASE 1 DEFICIENCY » HPRT1 DEFICIENCY » HPRT DEFICIENCY GOUT, HPRT-RELATED » KELLEY-SEEGMILLER SYNDROME	
HPS1	HERMANSKY-PUDLAK SYNDROME	1 Mutation: 16bp Duplication
HPS3	HERMANSKY-PUDLAK SYNDROME	2 Mutations: 3.9kb Deletion and IVS5+1G>A Mutation
HPS1 and HPS3	HERMANSKY-PUDLAK SYNDROME	3 Mutations
HRAS (V-HA-RAS HARVEY RAT SARCOMA VIRAL ONCOGENE HOMOLOG)	COSTELLO SYNDROME » FACIOCUTANEOUS SKELETAL SYNDROME	1 Exon: 2 or 4 Exons: 2, 3, 4, 6
HRPT2 (PARAFIBROMIN)	HYPERPARATHYROIDISM 2, HRPT2 » HYPERPARATHYROIDISM, FAMILIAL PRIMARY, WITH MULTIPLE OSSIFYING JAW FIBROMAS » HYPERPARATHYROIDISM-JAW TUMOR SYNDROME, HEREDITARY, HPT-JT » PARATHYROID ADENOMATOSIS, FAMILIAL CYSTIC HYPERPARATHYROIDISM 1, HRPT1 » HYPERPARATHYROIDISM, FAMILIAL ISOLATED PRIMARY » PARATHYROID ADENOMA, FAMILIAL	Exons 1-7 (Majority of Mutations) or Exons 8-17
HSD11B2	CORTISOL 11-BETA-KETOREDUCTASE DEFICIENCY » APPARENT MINERALOCORTICOID EXCESS » 11-@BETA-HYDROXYSTEROID DEHYDROGENASE, TYPE 2	Exons 1-7 (Majority of Mutations) or Exons 8-17

HSD17B3 (17-@BETA HYDROXYSTEROID DEHYDROGENASE 3, ESTRADIOL 17-BETA-DEHYDROGENASE,EDH17B3)	17-@BETA HYDROXYSTEROID DEHYDROGENASE 3 DEFICIENCY » PSEUDOHERMAPHRODITISM, MALE, WITH GYNECOMASTIA » POLYCYSTIC OVARIAN DISEASE DUE TO 17-KETOSTEROID REDUCTASE DEFICIENCY » 17-@KETOSTEROID REDUCTASE DEFICIENCY OF TESTIS	
HSPD1 (HEAT-SHOCK 60-KD PROTEIN 1, SPG13)	SPASTIC PARAPLEGIA 13 (AUTOSOMAL DOMINANT)	
HSPG2 (PERLECAN, HEPARAN SULFATE PROTEOGLYCAN OF BASEMENT MEMBRANE)	DYSEGMENTAL DYSPLASIA, SILVERMAN-HANDMAKER TYPE » ANISOSPONDYLIC CAMPTOMICROMELIC DWARFISM, SILVERMAN-HANDMAKER TYPE SCHWARTZ-JAMPEL SYNDROME, TYPE 1 » SJA SYNDROME » MYOTONIC MYOPATHY, DWARFISM, CHONDRODYSTROPHY, AND OCULAR AND FACIAL ABNORMALITIES » CHONDRODYSTROPHIC MYOTONIA	
ICR1	SILVER-RUSSELL SYNDROME » RUSSELL-SILVER SYNDROME	Combined testing of epimutation at ICR1 on chromosome 11p15 and UPD of chromosome 7
IDS (IDURONATE SULFATASE)	MUCOPOLYSACCHARIDOSIS, TYPE 2 » HUNTER SYNDROME	
IFNGR1 (INTERFERON, GAMMA, RECEPTOR 1, ANTIVIRAL PROTEIN, TYPE 2)	ATYPICAL MYCOBACTERIOSIS, FAMILIAL » BCG AND SALMONELLA INFECTION, DISSEMINATED HELICOBACTER PYLORI INFECTION, SUSCEPTIBILITY TO	
IFNGR2 (INTERFERON, GAMMA, RECEPTOR 2)	ATYPICAL MYCOBACTERIOSIS, FAMILIAL » BCG AND SALMONELLA INFECTION, DISSEMINATED	
IFNGR1 and IFNGR2	ATYPICAL MYCOBACTERIOSIS, FAMILIAL » BCG AND SALMONELLA INFECTION, DISSEMINATED	
IGHM (IMMUNOGLOBULIN MU)	AGAMMAGLOBULINEMIA, NON-BRUTON TYPE (AUTOSOMAL RECESSIVE)	

IGHMBP2 (IMMUNOGLOBULIN MU BINDING PROTEIN 2, CARDIAC TRANSCRIPTION FACTOR 1, CATF1)	SPINAL MUSCULAR ATROPHY WITH RESPIRATORY DISTRESS 1, SMARD1 » NEURONOPATHY, DISTAL HEREDITARY MOTOR, TYPE 6 » SEVERE INFANTILE AXONAL NEUROPATHY WITH RESPIRATORY FAILURE	
IHH (INDIAN HEDGEHOG)	BRACHYDACTYLY TYPE A1, BDA1 » FARABEE TYPE BRACHYDACTYLY ACROCAPITOFEMORAL DYSPLASIA, ACFD	
IKBKAP (IKK COMPLEX-ASSOCIATED PROTEIN, IKAP)	DYSAUTONOMIA, FAMILIAL, DYS » RILEY-DAY SYNDROME » HEREDITARY SENSORY AND AUTONOMIC NEUROPATHY, TYPE 3, HSAN-3 » (See also Molecular Screening Tests)	2 Mutations: IVS2+6T-C, R696P
IKBKG (NEMO)	BLOCH-SULZBERGER DISEASE » INCONTINENTIA PIGMENTI HYPOHIDROTIC ECTODERMAL DYSPLASIA WITH IMMUNE DEFICIENCY » ECTODERMAL DYSPLASIA, HYPOHIDROTIC WITH IMMUNE DEFICIENCY	Whole Gene or 1 Mutation: Exon 4-10 Deletion Whole Gene or 1 Mutation: Exon 4-10 Deletion
IL2RG (INTERLEUKIN 2 RECEPTOR, GAMMA)	SEVERE COMBINED IMMUNODEFICIENCY (X-LINKED), T CELL-NEGATIVE, B CELL-POSITIVE, NK CELL-NEGATIVE, SCIDX1	
IL7R (INTERLEUKIN 7 RECEPTOR)	SEVERE COMBINED IMMUNODEFICIENCY (AUTOSOMAL RECESSIVE), T CELL-NEGATIVE, B CELL-POSITIVE, NK CELL-POSITIVE, SCID	
INSR (INSULIN RECEPTOR)	LEPRECHAUNISM » DONOHUE SYNDROME DIABETES MELLITUS, INSULIN-RESISTANT, WITH ACANTHOSIS NIGRICANS RABSON-MENDENHALL SYNDROME » PINEAL HYPERPLASIA, INSULIN-RESISTANT DIABETES MELLITUS, AND SOMATIC ABNORMALITIES INSULIN RESISTANCE HYPERINSULINEMIC HYPOGLYCEMIA, FAMILIAL, TYPE 5	
IPF1	MATURITY-ONSET DIABETES OF THE YOUNG, TYPE 4, MODY4	
IRF6	POPLITEAL PTERYGIUM SYNDROME	Selected Exons

JAK3 (JANUS KINASE 3)	HYPEREOSINOPHILIC SYNDROME THE MYELODYSPLASTIC SYNDROMES SEVERE COMBINED IMMUNODEFICIENCY (AUTOSOMAL RECESSIVE), T CELL-NEGATIVE, B CELL- POSITIVE, NK CELL-NEGATIVE, SCID	1Common Mutation: V617F 1Common Mutation: V617F
JK (KIDD BLOOD GROUP SYSTEM)	HEMOLYTIC DISEASE, NEWBORN	1 Mutation: N280D
JPH3 (JUNCTOPHILIN 3)	HUNTINGTON DISEASE-LIKE 2, HDL2	Repeat
KAL1 (ADHESION MOLECULE-LIKE, X- LINKED, ANOSMIN 1)	KALLMANN SYNDROME, TYPE 1, KAL1 » HYPOGONADOTROPIC HYPOGONADISM AND ANOSMIA » DYSPLASIA OLFACTOGENITALIS OF DE MORSIER » ANOSMIC HYPOGONADISM	
KCNA1 (POTASSIUM CHANNEL, VOLTAGE-GATED, SHAKER-RELATED SUBFAMILY, MEMBER 1)	EPISODIC ATAXIA, TYPE 1, EA1 » EPISODIC ATAXIA WITH MYOKYMIA » ATAXIA, EPISODIC, WITH MYOKYMIA » PAROXYSMAL ATAXIA WITH NEUROMYOTONIA, HEREDITARY » MYOKYMIA WITH PERIODIC ATAXIA » MYOKYMIA » CONTINUOUS MUSCLE FIBER ACTIVITY, HEREDITARY » ISAACS- MERTENS SYNDROME	
KCNE1 (MINK, ISK)	LONG QT SYNDROME 5, LQT5 » ROMANO-WARD SYNDROME JERVELL AND LANGE-NIELSEN SYNDROME, JLNS1	See also LONG QT PANEL 1 and LONG QT PANEL 2 and LONG QT PANEL 3
KCNE2	LONG QT SYNDROME 6, LQT6 » ROMANO-WARD SYNDROME	See also LONG QT PANEL 1 and LONG QT PANEL 2 and LONG QT PANEL 3
KCNJ1 (POTASSIUM CHANNEL, INWARDLY RECTIFYING, SUBFAMILY J, MEMBER 1, KIR1.1, ROMK1)	ANTENATAL BARTTER SYNDROME, TYPE 2 » HYPOKALEMIC ALKALOSIS WITH HYPERCALCIURIA, ANTENATAL, TYPE 2 » HYPERPROSTAGLANDIN E SYNDROME, TYPE 2	
KCNJ11	DIABETES MELLITUS, PERMANENT NEONATAL NESIDIOBLASTOSIS » HYPERINSULINISM (AUTOSOMAL RECESSIVE) » HYPERINSULINISM, FAMILIAL, WITH PANCREATIC NESIDIOBLASTOSIS » PERSISTENT HYPERINSULINEMIC HYPOGLYCEMIA OF	

KCNJ2 (POTASSIUM CHANNEL, INWARDLY RECTIFYING, SUBFAMILY J, MEMBER 2, KIR2.1)	INFANCY » PERSISTENT HYPERINSULINEMIC HYPOGLYCEMIA OF INFANCY DUE TO FOCAL ADENOMATOUS HYPERPLASIA ANDERSEN CARDIODYSRHYTHMIC PERIODIC PARALYSIS » ANDERSEN SYNDROME LONG QT SYNDROME 7, LQT7 » PERIODIC PARALYSIS, POTASSIUM-SENSITIVE CARDIODYSRHYTHMIC TYPE » ANDERSEN-TAWIL SYNDROME SHORT QT SYNDROME 3, SQT3	
KCNQ1 (KVLQT1)	LONG QT SYNDROME 1, LQT1 » ROMANO-WARD SYNDROME JERVELL AND LANGE-NIELSEN SYNDROME, JLNS1	See also LONG QT PANEL 1 and and LONG QT PANEL 3
KCNQ2 (POTASSIUM CHANNEL, VOLTAGE-GATED, KQT-LIKE SUBFAMILY, MEMBER 2)	EPILEPSY, BENIGN NEONATAL, TYPE 1 » CONVULSIONS, BENIGN FAMILIAL NEONATAL, TYPE 1 MYOKYMIA WITH NEONATAL EPILEPSY » EPILEPSY, BENIGN NEONATAL, WITH MYOKYMIA » CONVULSIONS, BENIGN FAMILIAL NEONATAL, WITH MYOKYMIA » BFNC/MYOKYMIA SYNDROME	
KCNQ1OT1 and H19	BECKWITH-WIEDEMANN SYNDROME, BWS » EXOMPHALOS-MACROGLOSSIA-GIGANTISM SYNDROME	KCNQ1OT1 and H19 Imprinting
KEL (KELL-CELLANO BLOOD GROUP SYSTEM, KELL NULL)	HEMOLYTIC DISEASE, NEWBORN	1 Mutation: T193M
KIF1B (KINESIN FAMILY MEMBER 1B)	CHARCOT-MARIE-TOOTH DISEASE, AXONAL, TYPE 2A, CMT2A	
KIF21A (KINESIN FAMILY MEMBER 21A)	FIBROSIS OF EXTRAOCULAR MUSCLES, CONGENITAL, TYPE 1, FEOM1, CFEOM3 » OPHTHALMOPLEGIA, CONGENITAL » BLEPHAROPTOSIS WITH ABSENT EYE MOVEMENTS	Hot Spots (Exons 8, 20, 21)
	FIBROSIS OF EXTRAOCULAR MUSCLES, CONGENITAL, TYPE 3, FEOM3, CFEOM3	Hot Spots (Exons 8, 20, 21)
KIF5A (KINESIN FAMILY MEMBER 5A)	FAMILIAL SPASTIC PARAPLEGIA 10 (AUTOSOMAL DOMINANT), SPG10	Whole Gene

KIND1 (KINDLIN 1)

KINDLER SYNDROME » POIKILODERMA, HEREDITARY ACROKERATOTIC » BULLOUS ACROKERATOTIC POIKILODERMA OF KINDLER AND WEARY » POIKILODERMA, CONGENITAL, WITH BULLAE, WEARY TYPE

KRAS (V-KI-RAS2 KIRSTEN RAT SARCOMA 2 VIRAL ONCOGENE HOMOLOG, KRAS2, KRAS1)

NOONAN SYNDROME, TYPE 3
CARDIOFACIOCUTANEOUS SINDROME, CFCY
LEUKEMIA, ACUTE MYELOGENOUS
STOMACH CANCER
PANCREATIC CARCINOMA

BLADDER CANCER, TRANSITIONAL CELL

BREAST CANCER, FAMILIAL

LUNG CANCER, SQUAMOUS CELL

KRT1 (KERATIN 1)

ICHTHYOSIS BULLOSA (SIEMENS)
ICHTHYOSIFORM ERYTHRODERMA, BULLOUS CONGENITAL » BROCC SYNDROME » EPIDERMOLYTIC HYPERKERATOSIS » HYPERKERATOSIS, ICHTHYOSIFORM ERYTHRODERMA, BULLOUS CONGENITAL » HYPERKERATOSIS, EPIDERMOLYTIC NONEPIDERMOLYTIC PALMOPLANTAR KERATODERMA » UNNA-THOST DISEASE » HYPERKERATOSIS, NONEPIDERMOLYTIC PALMOPLANTAR KERATODERMA
CYCLIC ICHTHYOSIS WITH EPIDERMOLYTIC HYPERKERATOSIS » HYPERKERATOSIS, CYCLIC

Whole Gene or Hotspots

Whole Gene or Hotspots

Whole Gene or Hotspots

Whole Gene or Hotspots

KRT1 and KRT10 (KERATIN 1 and KERATIN 10)	ICHTHYOSIS WITH EPIDERMOLYTIC HYPERKERATOSIS	
	ICHTHYOSIS BULLOSA (SIEMENS)	Hotspots
KRT2E (KERATIN 2E)	ICHTHYOSIFORM ERYTHRODERMA, BULLOUS CONGENITAL » BROcq SYNDROME » EPIDERMOLYTIC HYPERKERATOSIS » HYPERKERATOSIS, ICHTHYOSIFORM ERYTHRODERMA, BULLOUS CONGENITAL » HYPERKERATOSIS, EPIDERMOLYTIC HYPERKERATOSIS	Hotspots
	ICHTHYOSIS BULLOSA (SIEMENS)	
	ICHTHYOSIFORM ERYTHRODERMA, BULLOUS CONGENITAL » BROcq SYNDROME » EPIDERMOLYTIC HYPERKERATOSIS » HYPERKERATOSIS, ICHTHYOSIFORM ERYTHRODERMA, BULLOUS CONGENITAL » HYPERKERATOSIS, EPIDERMOLYTIC HYPERKERATOSIS	
	WHITE SPONGE NEVUS	
	KRT4 (KERATIN 4)	
	KRT5 (KERATIN 5)	
	EPIDERMOLYSIS BULLOSA SIMPLEX (DOWLING-MEARA TYPE)	Whole Gene
	EPIDERMOLYSIS BULLOSA SIMPLEX (WEBER-COCKAYNE TYPE)	Whole Gene
	EPIDERMOLYSIS BULLOSA SIMPLEX (KOEbNER TYPE)	Whole Gene
	KRT5 and KRT14 (KERATIN 5 and KERATIN 14)	EPIDERMOLYSIS BULLOSA SIMPLEX (DOWLING-MEARA TYPE)
	EPIDERMOLYSIS BULLOSA SIMPLEX (WEBER-COCKAYNE TYPE)	Hotspots
	EPIDERMOLYSIS BULLOSA SIMPLEX (KOEbNER TYPE)	Hotspots
KRT6A (KERATIN 6A)	JADASSOHN-LEWANDOWSKY SYNDROME » PACHYONYCHIA CONGENITA, TYPE 1	
KRT6B (KERATIN 6B)	JADASSOHN-LEWANDOWSKY SYNDROME » PACHYONYCHIA CONGENITA, TYPE 1	
KRT9 (KERATIN 9)	EPIDERMOLYTIC PALMOPLANTAR KERATODERMA » VORNER DISEASE » HYPERKERATOSIS, EPIDERMOLYTIC PALMOPLANTAR KERATODERMA	Whole Gene or Hotspots
KRT10 (KERATIN 10)	ICHTHYOSIS BULLOSA (SIEMENS)	Whole Gene or Hotspots
	ICHTHYOSIFORM ERYTHRODERMA, BULLOUS	Whole Gene or Hotspots

KRT13 (KERATIN 13)	CONGENITAL » BROCC SYNDROME » EPIDERMOLYTIC HYPERKERATOSIS » HYPERKERATOSIS, ICHTHYOSIFORM ERYTHRODERMA, BULLOUS CONGENITAL » HYPERKERATOSIS, EPIDERMOLYTIC HYPERKERATOSIS WHITE SPONGE NEVUS	
KRT14 (KERATIN 14)	EPIDERMOLYSIS BULLOSA SIMPLEX (DOWLING- MEARA TYPE)	Whole Gene
	EPIDERMOLYSIS BULLOSA SIMPLEX (WEBER- COCKAYNE TYPE)	Whole Gene
KRT16 (KERATIN 16)	EPIDERMOLYSIS BULLOSA SIMPLEX (KOEBSNER TYPE) JADASSOHN-LEWANDOWSKY SYNDROME » PACHYONYCHIA CONGENITA, TYPE 1 NONEPIDERMOLYTIC PALMOPLANTAR KERATODERMA » UNNA-THOST DISEASE » HYPERKERATOSIS, NONEPIDERMOLYTIC PALMOPLANTAR KERATODERMA	Whole Gene
KRT16 and KRT17 (KERATIN 16 and KERATIN 17)	NONEPIDERMOLYTIC PALMOPLANTAR KERATODERMA » UNNA-THOST DISEASE » HYPERKERATOSIS, NONEPIDERMOLYTIC PALMOPLANTAR KERATODERMA	Hotspots
KRT17 (KERATIN 17)	JACKSON-LAWLER DISEASE » PACHYONYCHIA CONGENITA, TYPE 2 STEATOCYSTOMA MULTIPLEX NONEPIDERMOLYTIC PALMOPLANTAR KERATODERMA » UNNA-THOST DISEASE » HYPERKERATOSIS, NONEPIDERMOLYTIC PALMOPLANTAR KERATODERMA	
L1 (L1CAM)	HYDROCEPHALUS DUE TO CONGENITAL STENOSIS OF AQUEDUCT OF SYLVIUS, HSAS » AQUEDUCTAL STENOSIS (X-LINKED) CORPUS CALLOSUM AGENESIS (X-LINKED), ACC CRASH SYNDROME » MENTAL RETARDATION, APHASIA, SHUFFLING GATE, AND ADDUCTED	

LAD (LIPOAMIDE DEHYDROGENASE)	THUMBS SYNDROME, MASA FAMILIAL SPASTIC PARAPLEGIA 1 (X-LINKED), SPG1 LIPOAMIDE DEHYDROGENASE DEFICIENCY	Preferentially on skin fibroblast culture for mutation analysis in proband, eventually blood in PAX RNA tubes
LAMA2 (ALPHA-2 LAMININ, HEAVY CHAIN LAMININ 2, MEROSIN)	MUSCULAR DYSTROPHY, CONGENITAL MEROSIN- DEFICIENT, TYPE 1A, MDC1A	
LAMA3 (LAMININ, ALPHA-3)	EPIDERMOLYSIS BULLOSA LETALIS » EPIDERMOLYSIS BULLOSA, JUNCTIONAL, HERLITZ TYPE » HERLITZ- PEARSON TYPE EPIDERMOLYSIS BULLOSA EPIDERMOLYSIS BULLOSA, GENERALIZED ATROPHIC BENIGN » EPIDERMOLYSIS BULLOSA JUNCTIONALIS, PROGRESSIVE » EPIDERMOLYSIS BULLOSA JUNCTIONALIS, SEVERE NONLETHAL » EPIDERMOLYSIS BULLOSA JUNCTIONALIS, DISENTIS TYPE » EPIDERMOLYSIS BULLOSA, JUNCTIONAL, LOCALISATA VARIANT	Whole Gene See also LAMA3, LAMB3 and LAMC2 Whole Gene See also LAMA3, LAMB3 and LAMC2
LAMB2 (LAMININ, BETA-2)	PIERSON SYNDROME » MICROCORIA-CONGENITAL NEPHROTIC SYNDROME » NEPHROTIC SYNDROME, CONGENITAL, WITH OR WITHOUT OCULAR ABNORMALITIES, INCLUDED	
LAMB3 (LAMININ, BETA-3)	EPIDERMOLYSIS BULLOSA LETALIS » EPIDERMOLYSIS BULLOSA, JUNCTIONAL, HERLITZ TYPE » HERLITZ- PEARSON TYPE EPIDERMOLYSIS BULLOSA EPIDERMOLYSIS BULLOSA, GENERALIZED ATROPHIC BENIGN » EPIDERMOLYSIS BULLOSA JUNCTIONALIS, PROGRESSIVE » EPIDERMOLYSIS BULLOSA JUNCTIONALIS, SEVERE NONLETHAL » EPIDERMOLYSIS BULLOSA JUNCTIONALIS, DISENTIS TYPE » EPIDERMOLYSIS BULLOSA, JUNCTIONAL, LOCALISATA VARIANT	Whole Gene See also LAMA3, LAMB3 and LAMC2 Whole Gene See also LAMA3, LAMB3 and LAMC2
LAMC2 (LAMININ, GAMMA-2)	EPIDERMOLYSIS BULLOSA LETALIS » EPIDERMOLYSIS BULLOSA, JUNCTIONAL, HERLITZ TYPE » HERLITZ- PEARSON TYPE EPIDERMOLYSIS BULLOSA	Whole Gene See also LAMA3, LAMB3 and LAMC2

LAMA3, LAMB3 and LAMC2	<p>EPIDERMOLYSIS BULLOSA, GENERALIZED ATROPHIC BENIGN » EPIDERMOLYSIS BULLOSA JUNCTIONALIS, PROGRESSIVE » EPIDERMOLYSIS BULLOSA JUNCTIONALIS, SEVERE NONLETHAL » EPIDERMOLYSIS BULLOSA JUNCTIONALIS, DISENTIS TYPE » EPIDERMOLYSIS BULLOSA, JUNCTIONAL, LOCALISATA VARIANT</p> <p>EPIDERMOLYSIS BULLOSA LETALIS » EPIDERMOLYSIS BULLOSA, JUNCTIONAL, HERLITZ TYPE » HERLITZ-PEARSON TYPE EPIDERMOLYSIS BULLOSA EPIDERMOLYSIS BULLOSA, GENERALIZED ATROPHIC BENIGN » EPIDERMOLYSIS BULLOSA JUNCTIONALIS, PROGRESSIVE » EPIDERMOLYSIS BULLOSA JUNCTIONALIS, SEVERE NONLETHAL » EPIDERMOLYSIS BULLOSA JUNCTIONALIS, DISENTIS TYPE » EPIDERMOLYSIS BULLOSA, JUNCTIONAL, LOCALISATA VARIANT</p>	<p>Whole Gene See also LAMA3, LAMB3 and LAMC2</p> <p>6 Common Mutations in 3 Genes: LAMA3: R650X LAMB3: R42X, Q243X, R635X, 77 BP Deletion LAMC2: R95X</p> <p>6 Common Mutations in 3 Genes: LAMA3: R650X LAMB3: R42X, Q243X, R635X, 77 BP Deletion LAMC2: R95X</p>
LAMP2 (LYSOSOME-ASSOCIATED MEMBRANE PROTEIN 2)	<p>GLYCOGEN STORAGE DISEASE, TYPE 2B » GSD TYPE 2B » VACUOLAR CARDIOMYOPATHY AND MYOPATHY (X-LINKED) » LYSOSOMAL GLYCOGEN STORAGE DISEASE WITHOUT ACID MALTASE DEFICIENCY » GLYCOGEN STORAGE CARDIOMYOPATHY » PSEUDOGLYCOGENOSIS 2 GLYCOGEN STORAGE DISEASE LIMITED TO THE HEART » ANTOPOL DISEASE » DANON DISEASE</p>	
LCAT (LECITHIN:CHOLESTEROL ACYLTRANSFERASE)	<p>LECITHIN:CHOLESTEROL ACYLTRANSFERASE DEFICIENCY » LCAT DEFICIENCY » NORUM DISEASE FISH-EYE DISEASE</p>	
LCT (LACTASE)	<p>LACTOSE INTOLERANCE » HYPOLACTASIA » DISACCHARIDE INTOLERANCE » ADULT LACTASE DEFICIENCY » LACTASE NONPERSISTENCE</p>	T13910C
LDLR (LDL RECEPTOR)	<p>HYPERCHOLESTEROLEMIA (AUTOSOMAL DOMINANT)</p>	Whole Gene LDLR + 3 Exons in APOB
LEMD3 (LEM DOMAIN-CONTAINING 3, MAN1)	<p>BUSCHKE-OLLENDORFF SYNDROME » DERMATOOSTEOPOIKILOSIS » DERMATOFIBROSIS,</p>	

LEPRE1 (LEUCINE- AND PROLINE- ENRICHED PROTEOGLYCAN 1, LEPRECAN, PROLYL 3-HYDROXYLASE 1)	DISSEMINATED, WITH OSTEOPOIKILOSIS » DERMATOFIBROSIS LENTICULARIS DISSEMINATA WITH OSTEOPOIKILOSIS » OSTEOPATHIA CONDENSANS DISSEMINATA » OSTEOPOIKILOSIS, ISOLATED, INCLUDED MELORHEOSTOSIS » MELORHEOSTOSIS WITH OSTEOPOIKILOSIS OSTEOGENESIS IMPERFECTA, TYPE 8, O18	
LFNG (LUNATIC FRINGE)	SPONDYLOCOSTAL DYSOSTOSIS, TYPE 3 (AUTOSOMAL RECESSIVE)	
LHCGR (LUTEINIZING HORMONE / CHORIOGONADOTROPIN RECEPTOR, LUTROPIN-CHORIOGONADOTROPIN RECEPTOR)	PRECOCIOUS PUBERTY, MALE-LIMITED » SEXUAL PRECOCITY, FAMILIAL, GONADOTROPIN-INDEPENDENT » TESTOTOXICOSIS, FAMILIAL LEYDIG CELL HYPOPLASIA WITH MALE PSEUDOHERMAPHRODITISM HYPERGONADOTROPIC HYPOGONADISM, FEMALE MICROPENIS	
LHX3 (LIM HOMEobox GENE 3, LIM3)	PITUITARY DWARFISM 3 » PANHYPOPITUITARISM » ATELIOTIC DWARFISM WITH HYPOGONADISM » HANHART DWARFISM » PITUITARY HORMONE DEFICIENCY, COMBINED » PITUITARY HORMONE DEFICIENCY, COMBINED WITH RIGID CERVICAL SPINE, INCLUDED	
LHX4 (LIM HOMEobox GENE 4, LHX4, LHX4/IGHG1 FUSION GENE)	SHORT STATURE, PITUITARY AND CEREBELLAR DEFECTS, AND SMALL SELLA TURCICA	
LIFR (LEUKEMIA INHIBITORY FACTOR RECEPTOR)	STUVE-WIEDEMANN SYNDROME » SCHWARTZ-JAMPEL SYNDROME, TYPE 2 » SCHWARTZ-JAMPEL SYNDROME, NEONATAL	Whole Gene or 653-654insT
LIPC (HEPATIC LIPASE, LIPH, HEPATIC TRIGLYCERIDE LIPASE, HTGL)	HEPATIC LIPASE DEFICIENCY	

LITAF (LIPOPOLYSACCHARIDE-INDUCED TUMOR NECROSIS FACTOR-ALPHA FACTOR)

LMNA (LAMIN A/C)

CHARCOT-MARIE-TOOTH DISEASE, DEMYELINATING, TYPE 1C, CMT1C, HMSN1C

MUSCULAR DYSTROPHY, LIMB-GIRDLE, TYPE 1B, LGMD1B
EMERY–DREYFUSS MUSCULAR DYSTROPHY (AUTOSOMAL DOMINANT), EDMD2
EMERY–DREYFUSS MUSCULAR DYSTROPHY (AUTOSOMAL RECESSIVE), EDMD3
LIPODYSTROPHY, FAMILIAL PARTIAL, TYPE 2, FPLD2 »
LIPODYSTROPHY, DUNNIGAN TYPE
DILATED CARDIOMYOPATHY WITH CARDIAC CONDUCTION DEFECTS, CMD1A
CHARCOT-MARIE-TOOTH DISEASE, AXONAL, TYPE 2B1, CMT2B1
MANDIBULOACRAL DYSPLASIA WITH TYPE A LIPODYSTROPHY, MADA » CRANIOMANDIBULAR DERMATODYSOSTOSIS
HUTCHINSON-GILFORD PROGERIA SYNDROME
WERNER SYNDROME
LIPOATROPHY WITH DIABETES, HEPATIC STEATOSIS, HYPERTROPHIC CARDIOMYOPATHY AND LEUKOMELANODERMIC PAPULES, LDHCP
NAIL-PATELLA SYNDROME, NPS

LMX1B

LONG QT PANEL 1: - KCNQ1 - HERG
LONG QT PANEL 2: - KCNQ1 - HERG - SCN5A - KCNE1 - KCNE2

LONG QT PANEL 3:

LOR (LORICRIN)

LONG QT SYNDROME
LONG QT SYNDROME

LONG QT SYNDROME

VOHWINKEL SYNDROME, VARIANT FORM »
VOHWINKEL SYNDROME WITH ICHTHYOSIS »
MUTILATING KERATODERMA WITH ICHTHYOSIS
ERYTHROKERATODERMIA, PROGRESSIVE SYMMETRIC

9 Exons with Mutation Hotspots
20 Exons with Mutation Hotspots

All 65 Exons

LPL (LIPOPROTEIN LIPASE)	HYPERLIPOPROTEINEMIA TYPE 1 » LIPOPROTEIN LIPASE DEFICIENCY » HYPERCHYLOMICRONEMIA	
LRAT (LECITHIN RETINOL ACYLTRANSFERASE)	RETINAL DYSTROPHY, EARLY-ONSET SEVERE	
LRRK2 (LEUCINE-RICH REPEAT KINASE)	PARKINSON DISEASE, TYPE 8, PARK8	Whole Gene or Exon 41
MANBA (MANNOSIDASE, BETA A, LYSOSOMAL)	MANNOSIDOSIS, BETA A, LYSOSOMAL » BETA-MANNOSIDASE DEFICIENCY	
MAP2K1 (MITOGEN-ACTIVATED PROTEIN KINASE KINASE 1, MEK1)	CARDIOFACIOCUTANEOUS SYNDROME, CFC » CFC SYNDROME	Whole Gene or Exons 2 and 3
MAP2K2 (MITOGEN-ACTIVATED PROTEIN KINASE KINASE 2, MEK2)	CARDIOFACIOCUTANEOUS SYNDROME, CFC » CFC SYNDROME	Whole Gene or Exons 2 and 3 (6% of CFC cases)
MAPT (MICROTUBULE-ASSOCIATED PROTEIN TAU)	FRONTOTEMPORAL DEMENTIA » MULTIPLE SYSTEM TAUOPATHY WITH PRESENILE DEMENTIA » DISINHIBITION-DEMENTIA-PARKINSONISM-AMYOTROPHY COMPLEX » WILHELMESEN-LYNCH DISEASE » FRONTOTEMPORAL DEMENTIA-AMYOTROPHIC LATERAL SCLEROSIS » PALLIDOPONTONIGRAL DEGENERATION	Whole Gene
	PICK DISEASE OF BRAIN	Whole Gene
	SUPRANUCLEAR PALSY, PROGRESSIVE, 1 » STEELE-RICHARDSON-OLSZEWSKI SYNDROME	Whole Gene
	17q21.31 MICRODELETION SYNDROME	Deletions
MASTL (MICROTUBULE-ASSOCIATED SERINE/THREONINE KINASE-LIKE)	THROMBOCYTOPENIA, TYPE 2 » THROMBOCYTOPENIA (AUTOSOMAL DOMINANT)	
MBL2 (LECTIN, MANNOSE-BINDING, SOLUBLE, 2, MANNAN-BINDING PROTEIN, COLLECTIN 1)	SUSCEPTIBILITY TO INFECTION	Whole Gene or 5 Mutations: ARG52CYS, GLY54ASP, GLY57GLU, -550G>C and -221G>C
	MANNANOSE-BINDING PROTEIN DEFICIENCY	Whole Gene or 5 Mutations: ARG52CYS, GLY54ASP, GLY57GLU, -550G>C and -221G>C

MC2R (MELANOCORTIN 2 RECEPTOR; ACTH RECEPTOR)	GLUCOCORTICOID DEFICIENCY 1 » ADRENAL UNRESPONSIVENESS TO ACTH » ACTH RESISTANCE	
MC4R (MELANOCORTIN 4 RECEPTOR)	OBESITY	
MCCC1 (3-@METHYLCROTONYL-CoA CARBOXYLASE 1; MCCA)	3-@METHYLCROTONYL-CoA CARBOXYLASE 1 DEFICIENCY » METHYLCROTONYLGLYCINURIA TYPE 1	
MCCC2 (3-@METHYLCROTONYL-CoA CARBOXYLASE 2; MCCB)	3-@METHYLCROTONYL-CoA CARBOXYLASE 2 DEFICIENCY » METHYLCROTONYLGLYCINURIA TYPE 2	
MCOLN1 (ML4, MUCOLYPIN)	MUCOLIPIDOSIS 4, ML4	
MCP (MEMBRANE COFACTOR PROTEIN, MEASLES VIRUS RECEPTOR, CD46, MIC10, TLX, TRA2.10)	HEMOLYTIC - UREMIC SYNDROME, HUS	
MECP2	RETT SYNDROME	Whole Gene or Deletion Analysis (MLPA)
	MENTAL RETARDATION WITH PSYCHOSIS, PYRAMIDAL SIGNS, AND MACROORCHIDISM	Whole Gene or Deletion Analysis (MLPA)
	NONSPECIFIC MENTAL RETARDATION (X-LINKED), MRX16 » MENTAL RETARDATION, NONSPECIFIC (X-LINKED), TYPE16, MRX16	Whole Gene or Deletion Analysis (MLPA)
MEFV	FAMILIAL MEDITERRANEAN FEVER, FMF	Exons 2,3,5 and 10 harbouring 98% of all mutations or Whole Gene
MEN1 (MENIN)	MULTIPLE ENDOCRINE NEOPLASIA, TYPE 1, MEN1	
MERTK (MER TYROSINE KINASE PROTOONCOGENE)	RETINITIS PIGMENTOSA, MERTK-RELATED	
MESP2 (MESODERM POSTERIOR 2)	SPONDYLOCOSTAL DYSOSTOSIS, TYPE 2 (AUTOSOMAL RECESSIVE)	
MEST (PEG1)	SILVER-RUSSELL SYNDROME	UPD Study by Methylation Analysis
MFN2 (MITOFUSIN 2)	CHARCOT-MARIE-TOOTH DISEASE, AXONAL, TYPE 2A, CMT2A	

MID1 (MIDLINE 1, MIDIN, MIDLINE 1 RING FINGER GENE)	OPITZ SYNDROME » OPITZ G/BBB SYNDROME (X-LINKED) » OPITZ SYNDROME (X-LINKED) » OPITZ-G SYNDROME	
MITF	WAARDENBURG SYNDROME, TYPE 2, WS2	Whole Gene or Deletion Analysis (MLPA)
MITOCHONDRIAL MUTATIONS	Listed Separately. » See also (Mitochondrial Molecular Tests)	
MLC1	MEGALENCEPHALIC LEUKOENCEPHALOPATHY WITH SUBCORTICAL CYSTS	
MLH1	COLORECTAL CANCER, HEREDITARY NONPOLYPOSIS, HNPCC » LYNCH CANCER FAMILY SYNDROME, TYPE 1	Whole Gene and MLPA See also MLH1, MSH2 and MSH6
	COLORECTAL CANCER, HEREDITARY NONPOLYPOSIS, HNPCC » LYNCH CANCER FAMILY SYNDROME, TYPE 2	Whole Gene and MLPA See also MLH1, MSH2 and MSH6
MLH1, MSH2 and MSH6	COLORECTAL CANCER, HEREDITARY NONPOLYPOSIS, HNPCC » LYNCH CANCER FAMILY SYNDROME, TYPE 1	3 Genes (Whole Gene and MLPA)
	COLORECTAL CANCER, HEREDITARY NONPOLYPOSIS, HNPCC » LYNCH CANCER FAMILY SYNDROME, TYPE 2	3 Genes (Whole Gene and MLPA)
MMAA	METHYLMALONIC ACIDURIA, cbIA TYPE » METHYLMALONIC ACIDURIA, VITAMIN B12-RESPONSIVE, DUE TO DEFECT IN SYNTHESIS OF ADENOSYLCOBALAMIN, cbIA TYPE	
MMAB (COBALAMIN ADENOSYLTRANSFERASE)	METHYLMALONIC ACIDURIA, cbIB TYPE » METHYLMALONIC ACIDURIA, VITAMIN B12-RESPONSIVE, DUE TO DEFECT IN SYNTHESIS OF ADENOSYLCOBALAMIN, cbIB TYPE	
MMACHC	METHYLMALONIC ACIDURIA AND HOMOCYSTINURIA, cbIC TYPE » VITAMIN B12 METABOLIC DEFECT WITH COMBINED DEFICIENCY OF METHYLMALONYL-CoA MUTASE AND HOMOCYSTEINE:METHYLTETRAHYDROFOLATE METHYLTRANSFERASE	

MPL (MYELOPROLIFERATIVE LEUKEMIA VIRUS ONCOGENE, TPOR)	AMEGAKARYOCYTIC THROMBOCYTOPENIA, CONGENITAL, CAMT
MPZ (MYELIN PROTEIN ZERO, P0)	CHARCOT-MARIE-TOOTH DISEASE, DEMYELINATING, TYPE 1B, CMT1B » HEREDITARY MOTOR AND SENSORY NEUROPATHY 1B, HMSN1B CHARCOT-MARIE-TOOTH DISEASE, AXONAL, TYPE 2I, CMT2I CHARCOT-MARIE-TOOTH DISEASE, AXONAL, TYPE 2J, CMT2J CHARCOT-MARIE-TOOTH DISEASE, DEMYELINATING, TYPE 4F, CMT4F » DEJERINE-SOTTAS HYPERTROPHIC NEUROPATHY CHARCOT-MARIE-TOOTH DISEASE, DOMINANT INTERMEDIATE D, CMTDID NEUROPATHY, CONGENITAL HYPOMYELINATING ROUSSY-LEVY HEREDITARY AREFLEXIC DYSTASIA
MR1 (MYOFIBRILLOGENESIS REGULATOR 1)	PAROXYSMAL NONKINESIGENIC DYSKINESIA » PAROXYSMAL DYSTONIC CHOREOATHETOSIS » MOUNT-REBACK SYNDROME » CHOREOATHETOSIS, NONKINESIGENIC » DYSTONIA 8, DYT8
MRX-Panel: ARX, PQBP1, JARID1C, TM4SF2, FAFL4, DLG3, FTSJ1, ZNF41	MENTAL RETARDATION, NONSPECIFIC (X-LINKED)
MRAP (MELANOCORTIN 2 RECEPTOR ACCESSORY PROTEIN)	GLUCOCORTICOID DEFICIENCY 2
MSK1	MECKEL SYNDROME, TYPE 1 » DYSENCEPHALIA SPLANCHNOCYSTICA » GRUBER SYNDROME » MECKEL-GRUBER SYNDROME
MSK3 (MECKELIN, TMEM67, TRANSMEMBRANE PROTEIN 67)	JOUBERT SYNDROME, TYPE 6 MECKEL SYNDROME, TYPE 3 » DYSENCEPHALIA SPLANCHNOCYSTICA » GRUBER SYNDROME »

	MECKEL-GRUBER SYNDROME	
MSH2	COLORECTAL CANCER, HEREDITARY NONPOLYPOSIS, HNPCC » LYNCH CANCER FAMILY SYNDROME, TYPE 1	Whole Gene and MLPA See also MLH1, MSH2 and MSH6
	COLORECTAL CANCER, HEREDITARY NONPOLYPOSIS, HNPCC » LYNCH CANCER FAMILY SYNDROME, TYPE 2	Whole Gene and MLPA See also MLH1, MSH2 and MSH6
MSH6	COLORECTAL CANCER, HEREDITARY NONPOLYPOSIS, HNPCC » LYNCH CANCER FAMILY SYNDROME, TYPE 1	Whole Gene and MLPA See also MLH1, MSH2 and MSH6
	COLORECTAL CANCER, HEREDITARY NONPOLYPOSIS, HNPCC » LYNCH CANCER FAMILY SYNDROME, TYPE 2	Whole Gene and MLPA See also MLH1, MSH2 and MSH6
MSX2 (MUSCLE SEGMENT HOMEBOX, DROSOPHILA, HOMOLOG OF, 2)	PARIETAL FORAMINA, TYPE 1 » FORAMINA PARIETALIA PERMAGNA » CATLIN MARKS PARIETAL FORAMINA WITH CLEIDOCRANIAL DYSPLASIA	
	CRANIOSYNOSTOSIS, TYPE 2 » CRANIOSYNOSTOSIS, BOSTON-TYPE	
MTM1 (MYOTUBULARIN)	MYOTUBULAR MYOPATHY (X-LINKED)	
MTP	ABETALIPOPROTEINEMIA » ACANTHOCYTOSIS » BASSEN-KORNZWEIG SYNDROME » APOLIPOPROTEIN B DEFICIENCY » MICROSOMAL TRIGLYCERIDE TRANSFER PROTEIN DEFICIENCY » MTP DEFICIENCY	
MUT (METHYLMALONYL CoA MUTASE,MCM)	METHYLMALONICACIDURIA DUE TO MCM DEFICIENCY » MMA DUE TO MCM DEFICIENCY » MCM DEFICIENCY	
MVK (MEVALONATE KINASE)	HYPER-IGD SYNDROME » PERIODIC FEVER, DUTCH TYPE	
MYBPC3 (MYOSIN-BINDING PROTEIN C, CARDIAC)	CARDIOMYOPATHY, FAMILIAL HYPERTROPHIC, TYPE 4	See also MYH7 MYBPC3, TNNT2, TNNI3 and TPM1
MYBPC3 (MYOSIN-BINDING PROTEIN C, CARDIAC)	HYPERTROPHIC CARDIOMYOPATHY, FAMILIAL, 4A, CMD4A » VENTRICULAR HYPERTROPHY, HEREDITARY » ASYMMETRIC SEPTAL HYPERTROPHY » HYPERTROPHIC SUBAORTIC STENOSIS, IDIOPATHIC	See also MYH7 MYBPC3, TNNT2, TNNI3 and TPM1

MYCN (V-MYC AVIAN
MYELOCYTOMATOSIS VIRAL-
RELATED ONCOGENE,
NEUROBLASTOMA-DERIVED)

FEINGOLD SYNDROME »
OCULODIGITOEOPHAGODUODENAL SYNDROME »
ODED SYNDROME » MICROCEPHALY-OCULO-DIGITO-
ESOPHAGEAL-DUODENAL SYNDROME » DIGITAL
ANOMALIES WITH SHORT PALPEBRAL FISSURES AND
ATRESIA OF ESOPHAGUSOR DUODENUM »
MICROCEPHALY, MENTAL RETARDATION, AND
TRACHEOESOPHAGEAL FISTULA SYNDROME » MMT
SYNDROME

Whole Gene or Deletions

MYH (MUTYH, MUTY, E. COLI,
HOMOLOG OF)
MYH3 (MYOSIN, HEAVY CHAIN 3,
SKELETAL MUSCLE, EMBRYONIC)

COLORECTAL ADENOMATOUS POLYPOSIS
(AUTOSOMAL RECESSIVE)
ARTHROGRYPOSIS, DISTAL, TYPE 2A » FREEMAN-
SHELDON SYNDROME » WHISTLING FACE-WINDMILL
VANE HAND SYNDROME » CRANIOCARPOTARSAL
DYSTROPHY
ARTHROGRYPOSIS, DISTAL, TYPE 2B »
ARTHROGRYPOSIS MULTIPLEX CONGENITA, DISTAL,
TYPE 2B » SHELDON-HALL SYNDROME » FREEMAN-
SHELDON SYNDROME VARIANT » ARTHROGRYPOSIS
MULTIPLEX CONGENITA, DISTAL, TYPE 2, WITH
CRANIOFACIAL ABNORMALITIES
HYPERTROPHIC CARDIOMYOPATHY, FAMILIAL, 1,
CMH1 » VENTRICULAR HYPERTROPHY, HEREDITARY »
ASYMMETRIC SEPTAL HYPERTROPHY »
HYPERTROPHIC SUBAORTIC STENOSIS, IDIOPATHIC
CARDIOMYOPATHY, DILATED, TYPE 1S

Whole Gene or 2 Common Mutations:
Y165C and G382D

All 39 Exons or Exons 5, 14, 15, 17, 21
or Exons 17 and 21

All 39 Exons or Exons 5, 14, 15, 17, 21
or Exons 17 and 21

MYH7 (MYOSIN, HEAVY CHAIN 7,
CARDIAC MUSCLE, BETA)

MYOPATHY, MYOSIN STORAGE » MYOPATHY,
HYALINE BODY (AUTOSOMAL DOMINANT)
MYOPATHY, DISTAL 1, MPD1 » LAING DISTAL
MYOPATHY » MYOPATHY, DISTAL, EARLY-ONSET
(AUTOSOMAL DOMINANT)

See also MYH7 MYBPC3, TNNT2, TNNI3
and TPM1

See also MYH7 MYBPC3, TNNT2, TNNI3
and TPM1

See also MYH7 MYBPC3, TNNT2, TNNI3
and TPM1

See also MYH7 MYBPC3, TNNT2, TNNI3
and TPM1

MYH7 MYBPC3 TNNT2 TNNI3 TPM1	HYPERTROPHIC CARDIOMYOPATHY, FAMILIAL » VENTRICULAR HYPERTROPHY, HEREDITARY » ASYMMETRIC SEPTAL HYPERTROPHY » HYPERTROPHIC SUBAORTIC STENOSIS, IDIOPATHIC » DILATED CARDIOMYOPATHY	106 Exons
MYH9 (MYOSIN, HEAVY CHAIN 9)	MAY-HEGGLIN ANOMALY » DOHLE LEUKOCYTE INCLUSIONS WITH GIANT PLATELETS » MACROTHROMBOCYTOPENIA WITH LEUKOCYTE INCLUSIONS SEBASTIAN SYNDROME » SEBASTIAN PLATELET SYNDROME FECHTNER SYNDROME » MACROTHROMBOCYTOPATHY, NEPHRITIS, DEAFNESS, AND LEUKOCYTE INCLUSIONS » ALPORT SYNDROME WITH LEUKOCYTE INCLUSIONS AND MACROTHROMBOCYTOPENIA EPSTEIN SYNDROME » MACROTHROMBOCYTOPATHY, NEPHRITIS, AND DEAFNESS » ALPORT SYNDROME WITH MACROTHROMBOCYTOPENIA DEAFNESS, DFNA17 » DEAFNESS, (AUTOSOMAL DOMINANT), NONSYNDROMIC SENSORINEURAL 17	
MYH11 (MYOSIN, HEAVY CHAIN 11, SMOOTH MUSCLE, SMOOTH MUSCLE MYOSIN HEAVY CHAIN)	AORTIC ANEURYSM, FAMILIAL THORACIC 4 » AORTIC ANEURYSM/AORTIC DISSECTION AND PATENT DUCTUS ARTERIOSUS	
MYL2 (MYOSIN, LIGHT CHAIN 2, REGULATORY, CARDIAC, SLOW)	HYPERTROPHIC CARDIOMYOPATHY, FAMILIAL, 10, CMH10 » VENTRICULAR HYPERTROPHY, HEREDITARY » ASYMMETRIC SEPTAL HYPERTROPHY » HYPERTROPHIC SUBAORTIC STENOSIS, IDIOPATHIC » CARDIOMYOPATHY, HYPERTROPHIC, MID-LEFT VENTRICULAR CHAMBER TYPE 2	See ACTC, MYL2 and MYL3

MYL3 (MYOSIN, LIGHT CHAIN 3, ALKALI, VENTRICULAR, SKELETAL, SLOW, ESSENTIAL LIGHT CHAIN OF MYOSIN)	HYPERTROPHIC CARDIOMYOPATHY, FAMILIAL, 8, CMH8 » VENTRICULAR HYPERTROPHY, HEREDITARY » ASYMMETRIC SEPTAL HYPERTROPHY » HYPERTROPHIC SUBAORTIC STENOSIS, IDIOPATHIC » CARDIOMYOPATHY, HYPERTROPHIC, MID-LEFT VENTRICULAR CHAMBER TYPE 1	See ACTC, MYL2 and MYL3
MYO7A (MYOSIN 7A)	USHER SYNDROME, TYPE 1B, USH1B » USHER SYNDROME, TYPE 1, NON-ACADIAN VARIETY	
MYOC (MYOCILIN)	GLAUCOMA, PRIMARY OPEN ANGLE, JUVENILE-ONSET, 1	See also CYP1B1, OPTN and MYOC
NAGLU (N-ACETYLGLUCOSAMINIDASE, ALPHA)	MUCOPOLYSACCHARIDOSIS TYPE 3B, MPS3B » SANFILIPPO SYNDROME B » N-ACETYL-ALPHA-D-GLUCOSAMINIDASE DEFICIENCY » NAGLU DEFICIENCY	
NBS1 (NIBRIN)	NIJMEGEN BREAKAGE SYNDROME, NBS	5 bp Deletion (90% of all mutations) or Whole Gene
NCF1 (p47 PHOX)	CHRONIC GRANULOMATOUS DISEASE (AUTOSOMAL RECESSIVE) CYTOCHROME-b-POSITIVE FORM TYPE 1	
NCF2 (p67 PHOX)	CHRONIC GRANULOMATOUS DISEASE (AUTOSOMAL RECESSIVE) CYTOCHROME-b-POSITIVE FORM TYPE 2	
NDP (NORRIN)	NORRIE DISEASE » ATROPHIA BULBORUM HEREDITARIA » PSEUDOGLIOMA » EPISKOPI BLINDNESS » EXUDATIVE RETINOPATHY (X-LINKED) COATS DISEASE	
NDUFS1 (NADH-UBIQUINONE OXIDOREDUCTASE Fe-S PROTEIN 1, COMPLEX 1, MITOCHONDRIAL RESPIRATORY CHAIN, 75-KD SUBUNIT)	CYTOCHROME c OXIDASE DEFICIENCY » COX DEFICIENCY » COMPLEX 1, MITOCHONDRIAL RESPIRATORY CHAIN, DEFICIENCY OF LEIGH SYNDROME	
NDUFS3 (NADH-UBIQUINONE OXIDOREDUCTASE Fe-S PROTEIN 3, NADH-COENZYME Q REDUCTASE, 30-KD COMPLEX 1, MITOCHONDRIAL RESPIRATORY	CYTOCHROME c OXIDASE DEFICIENCY » COX DEFICIENCY » COMPLEX 1, MITOCHONDRIAL RESPIRATORY CHAIN, DEFICIENCY OF LEIGH SYNDROME	

CHAIN, 30-KD SUBUNIT)		
NDUFS4 (NADH-UBIQUINONE OXIDOREDUCTASE Fe-S PROTEIN 4, NADH-COENZYME Q REDUCTASE, 18-KD COMPLEX 1, MITOCHONDRIAL RESPIRATORY CHAIN, 18-KD SUBUNIT, AQQDQ)	CYTOCHROME c OXIDASE DEFICIENCY » COX DEFICIENCY » COMPLEX 1, MITOCHONDRIAL RESPIRATORY CHAIN, DEFICIENCY OF LEIGH SYNDROME	
NDUFS7 (NADH-UBIQUINONE OXIDOREDUCTASE Fe-S PROTEIN 7, NADH-COENZYME Q REDUCTASE, 20-KD COMPLEX 1, MITOCHONDRIAL RESPIRATORY CHAIN, 20-KD SUBUNIT, PSST)	CYTOCHROME c OXIDASE DEFICIENCY » COX DEFICIENCY » COMPLEX 1, MITOCHONDRIAL RESPIRATORY CHAIN, DEFICIENCY OF LEIGH SYNDROME	
NDUFS8 (NADH-UBIQUINONE OXIDOREDUCTASE Fe-S PROTEIN 8, COMPLEX 1, MITOCHONDRIAL RESPIRATORY CHAIN, 23-KD SUBUNIT, TYKY)	CYTOCHROME c OXIDASE DEFICIENCY » COX DEFICIENCY » COMPLEX 1, MITOCHONDRIAL RESPIRATORY CHAIN, DEFICIENCY OF LEIGH SYNDROME	
NDUFV1 (NADH-UBIQUINONE OXIDOREDUCTASE FLAVOPROTEIN 1, COMPLEX 1, MITOCHONDRIAL RESPIRATORY CHAIN, 51-KD SUBUNIT, UQOR1)	CYTOCHROME c OXIDASE DEFICIENCY » COX DEFICIENCY » COMPLEX 1, MITOCHONDRIAL RESPIRATORY CHAIN, DEFICIENCY OF LEIGH SYNDROME	
NEB (NEBULIN)	NEMALINE MYOPATHY (AUTOSOMAL RECESSIVE), NEM2	Common Deletion of Exon 55
NEFL (NEUROFILAMENT PROTEIN, LIGHT POLYPEPTIDE)	CHARCOT-MARIE-TOOTH DISEASE, AXONAL, TYPE 2E, CMT2E CHARCOT-MARIE-TOOTH DISEASE, DEMYELINATING, TYPE 1F, CMT1F	
NEU1 (NEURAMINIDASE 1, SIALIDASE)	NEURAMINIDASE DEFICIENCY » SIALIDOSIS, TYPE 2 » SIALIDOSIS, TYPE 1 » MUCOLIPIDOSIS 1, ML1 » LIPOMUCOPOLYSACCHARIDOSIS » SIALIDASE DEFICIENCY » NEUG DEFICIENCY » CHERRY RED SPOT--MYOCLONUS SYNDROME » MYOCLONUS--CHERRY RED SPOT SYNDROME	

NEUROD1 (BETA2)	MATURITY-ONSET DIABETES OF THE YOUNG, TYPE 6, MODY6 DIABETES MELLITUS, NONINSULIN-DEPENDENT, NIDDM » DIABETES MELLITUS, TYPE 2 » NONINSULIN-DEPENDENT DIABETES MELLITUS » MATURITY-ONSET DIABETES » INSULIN RESISTANCE, SUSCEPTIBILITY TO	
NF1 (NEUROFIBROMIN)	NEUROFIBROMATOSIS, TYPE 1, NF1 » VON RECKLINGHAUSEN DISEASE	At least 15 (adults) or 5 ml (children) EDTA-blood has to arrive at room temperature within 48 hrs after withdrawal
NF2 (NEUROFIBROMIN 2, MERLIN, SCHWANNOMIN)	NEUROFIBROMATOSIS TYPE 2, NF2 » NEUROFIBROMATOSIS, CENTRAL TYPE » ACOUSTIC SCHWANNOMAS, BILATERAL » BILATERAL ACOUSTIC NEUROFIBROMATOSIS » ACOUSTIC NEURINOMA	
NHLRC1 (NHL REPEAT-CONTAINING 1 GENE, EPM2B, MALIN)	MYOCLONIC EPILEPSY OF LAFORA » LAFORA DISEASE » EPILEPSY, PROGRESSIVE MYOCLONIC TYPE 2	
NHS	NANCE-HORAN SYNDROME » CATARACT-DENTAL SYNDROME » CATARACT, X-LINKED, WITH HUTCHINSONIAN TEETH » MESIODENS-CATARACT SYNDROME	
NIPA1 (NONIMPRINTED GENE IN PRADER-WILLI SYNDROME/ANGELMAN SYNDROME CHROMOSOME REGION 1)	FAMILIAL SPASTIC PARAPLEGIA 6 (AUTOSOMAL DOMINANT), SPG6	Whole Gene
NIPBL	CORNELIA DE LANGE SYNDROME » BRACHMANN-DE LANGE SYNDROME	
NKX2E (NK2, DROSOPHILA, HOMOLOG OF, E, NKX2.5, CSX)	ATRIAL SEPTAL DEFECT WITH ATRIOVENTRICULAR CONDUCTION DEFECTS TETRALOGY OF FALLOT	

	ATRIOVENTRICULAR BLOCK, IDIOPATHIC SECOND-DEGREE	
	VARIOUS HEART MALFORMATIONS	
NLGN3 (NEUROLIGIN 3)	AUTISM (X-LINKED)	
	ASPERGER SYNDROME (X-LINKED)	
NLGN4 (NEUROLIGIN 4)	AUTISM (X-LINKED)	
	ASPERGER SYNDROME (X-LINKED)	
NLGN3 AND NLGN4	AUTISM (X-LINKED)	
	ASPERGER SYNDROME (X-LINKED)	
NOG (NOGGIN)	PROXIMAL SYMPHALANGISM	
	MULTIPLE SYNOSTOSIS SYNDROME 1	
	TARSAL-CARPAL COALITION SYNDROME	
	STAPES ANKYLOSIS » TEUNISSEN-CREMERS SYNDROME	
NOTCH3	CEREBRAL AUTOSOMAL DOMINANT ARTERIOPATHY WITH SUBCORTICAL INFARCTS AND LEUKOENCEPHALOPATHY, CADASIL	Whole Gene
		Exons 3 and 4 harboring the majority of mutations
NPC1	NIEMANN-PICK DISEASE, TYPE C1, NPC1 » NIEMANN-PICK DISEASE, TYPE D » NIEMANN-PICK DISEASE, NOVA SCOTIAN TYPE	
NPC2	NIEMANN-PICK DISEASE, TYPE C2	
NPHP1 (NEPHROCYSTIN)	NEPHRONOPHTHISIS, TYPE 1	
	SENIOR-LOKEN SYNDROME » RENAL - RETINAL SYNDROME » JUVENILE NEPHRONOPHTHISIS WITH LEBER AMAUROSIS » RENAL DYSPLASIA AND RETINAL APLASIA	
	JOUBERT SYNDROME, TYPE 4	
NPHP2 (NEPHROCYSTIN 2, INVS, INVERSIN)	NEPHRONOPHTHISIS, TYPE 2 » NEPHRONOPHTHISIS, INFANTILE	
NPHP3 (NEPHROCYSTIN 3)	NEPHRONOPHTHISIS, TYPE 3 » NEPHRONOPHTHISIS, ADOLESCENT	
NPHP4 (NEPHROCYSTIN 4,	NEPHRONOPHTHISIS, TYPE 4 » NEPHRONOPHTHISIS,	

NEPHRORETININ)	JUVENILE	
	SENIOR-LOKEN SYNDROME 4	
NPHS1 (NEPHRIN)	NEPHROSIS 1, CONGENITAL, FINNISH TYPE, NPHS1 » FINNISH CONGENITAL NEPHROSIS NEPHROTIC SYNDROME, STEROID-RESISTANT, (AUTOSOMAL RECESSIVE), SRN1 » FAMILIAL FOCAL SEGMENTAL GLOMERULOSCLEROSIS (AUTOSOMAL RECESSIVE)	
NPHS2 (PODOCIN)	NEPHROTIC SYNDROME, STEROID-RESISTANT, (AUTOSOMAL RECESSIVE), SRN1 » FAMILIAL FOCAL SEGMENTAL GLOMERULOSCLEROSIS (AUTOSOMAL RECESSIVE) NEPHROSIS 1, CONGENITAL, FINNISH TYPE, NPHS1 » FINNISH CONGENITAL NEPHROSIS	
NPR2 (NATRIURETIC PEPTIDE RECEPTOR B/GUANYLATE CYCLASE B, ATRIAL NATRIURETIC PEPTIDE RECEPTOR, TYPE B, ANPRB)	ACROMESOMELIC DYSPLASIA, MAROTEAUX TYPE » ST. HELENA DYSPLASIA	
NR3C1	GLUCOCORTICOID RECEPTOR DEFICIENCY » GLUCOCORTICOID RESISTANCE » CORTISOL RESISTANCE FROM GLUCOCORTICOID RECEPTOR DEFECT » PSEUDOHERMAPHRODITISM WITH HYPOKALEMIA DUE TO GLUCOCORTICOID RESISTANCE	
NR3C2 (MINERALOCORTICOID RECEPTOR, MLR, MCR, MR, ALDOSTERONE RECEPTOR)	PSEUDOHYPOALDOSTERONISM, TYPE 1 (AUTOSOMAL DOMINANT) HYPERTENSION, EARLY-ONSET (AUTOSOMAL DOMINANT)	
NSD1	CEREBRAL GIGANTISM » SOTOS SYNDROME	At least 20mg DNA is needed
NTRK1	INSENSITIVITY TO PAIN, CONGENITAL, WITH ANHIDROSIS, CIPA » NEUROPATHY, CONGENITAL SENSORY, WITH ANHIDROSIS » HEREDITARY SENSORY AND AUTONOMIC NEUROPATHY 4, HSN4 » FAMILIAL DYSAUTONOMIA, TYPE 2	

NYX (NYCTALOPIN)	NIGHT BLINDNESS, CONGENITAL STATIONARY, TYPE 1A » HEMERALOPIA-MYOPIA » MYOPIA-NIGHT BLINDNESS » NYCTALOPIA	
OA1	ALBINISM, OCULAR, TYPE 1, OA1 » NETTLESHIP-FALLS TYPE OCULAR ALBINISM	
OCA1 (TYR, TYROSINASE)	OCULOCUTANEOUS ALBINISM, TYPE 1, OCA1 (TYROSINASE-NEGATIVE)	Whole Gene
OCA1 and OCA2	OCULOCUTANEOUS ALBINISM	2 Whole Genes
OCA2 (P gene)	OCULOCUTANEOUS ALBINISM, TYPE 2, OCA2 (TYROSINASE - POSITIVE) » PINK-EYED DILUTION » BROWN OCULOCUTANEOUS ALBINISM, BOCA	Whole Gene or Common Deletion
OCRL1	LOWE OCULOCEREBRORENAL SYNDROME, OCRL DENT NEPHROCALCINOSIS	
OFD1 (CHROMOSOME X OPEN READING FRAME 5; CXORF5)	OROFACIODIGITAL SYNDROME, TYPE 1 » OFD SYNDROME, TYPE 1 » PAPILLON-LEAGUE-PSAUME SYNDROME	
OPA1	SIMPSON-GOLABI-BEHMEL SYNDROME, TYPE 2 OPTIC ATROPHY 1 » OPTIC ATROPHY, JUVENILE » KJER TYPE OPTIC ATROPHY » OPTIC ATROPHY, DEAFNESS, OPHTHALMOPLEGIA, AND MYOPATHY	
OPTN (OPTINEURIN)	GLAUCOMA, PRIMARY OPEN ANGLE, ADULT-ONSET, POAG	See also CYP1B1, OPTN and MYOC
OTC (ORNITHINE CARBAMOYL TRANSFERASE GENE)	ORNITHINE CARBAMOYLTRANSFERASE DEFICIENCY	
OTOF (OTOFERLIN)	DEAFNESS, DFNB9 » DEAFNESS, (AUTOSOMAL RECESSIVE), NONSYNDROMIC SENSORINEURAL 9 » AUDITORY NEUROPATHY, AUTOSOMAL RECESSIVE, TYPE 1	
P16 (CDKN2)	CUTANEOUS MALIGNANT MELANOMA 2, CMM2 MALIGNANT MELANOMA WITH NEURAL CELL TUMORS » MELANOMA-ASTROCYTOMA SYNDROME	
P53 (TP53)	LI-FRAUMENI SYNDROME 1 OSTEOSARCOMA	Whole Gene or Exons 5-8 Whole Gene or Exons 5-8

P63 (TP63)	BREAST CANCER, FAMILIAL	Whole Gene or Exons 5-8
	PEDIATRIC ADRENOCORTICAL CARCINOMA	Whole Gene or Exons 5-8
	ECTRODACTYLY-ECTODERMAL DYSPLASIA-CLEFTING, TYPE 1, EEC1 » ECTODERMAL DYSPLASIA, ECTRODACTYLY, CLEFTING, TYPE 1	Exons 5-8, 13 and 14
	ECTRODACTYLY-ECTODERMAL DYSPLASIA-CLEFTING, TYPE 3, EEC3 » ECTODERMAL DYSPLASIA, ECTRODACTYLY, CLEFTING, TYPE 3	Exons 5-8, 13 and 14
	ACRO-DERMATO-UNGUAL-LACRIMAL-TOOTH SYNDROME » ADULT SYNDROME	Exons 5-8, 13 and 14
	ANKYLOBLEPHARON-ECTODERMAL DEFECTS WITH CLEFT LIP AND PALATE » HAY-WELLS SYNDROME	Exons 5-8, 13 and 14
PABPN1 (PAB2, POLYA BINDING PROTEIN)	LIMB-MAMMARY SYNDROME	Exons 5-8, 13 and 14
	SPLIT HAND - SPLIT FOOT, TYPE 4, SHFM4	Exons 5-8, 13 and 14
	OCULOPHARYNGEAL MUSCULAR DYSTROPHY (AUTOSOMAL DOMINANT) » MUSCULAR DYSTROPHY, OCULOPHARYNGEAL MUSCULAR DYSTROPHY (AUTOSOMAL DOMINANT)	Repeat
	OCULOPHARYNGEAL MUSCULAR DYSTROPHY (AUTOSOMAL RECESSIVE) » MUSCULAR DYSTROPHY, OCULOPHARYNGEAL MUSCULAR DYSTROPHY (AUTOSOMAL RECESSIVE)	Repeat
PAFAH1B1 (LIS1)	LISSENCEPHALY 1, LIS1 » SUBCORTICAL LAMINAR HETEROTOPIA » SUBCORTICAL BAND HETEROTOPIA	Whole Gene or Deletions
PAH (PHENYLALANINE HYDROXYLASE)	MILLER-DIEKER LISSENCEPHALY SYNDROME, MDLS PHENYLKETONURIA, PKU »	Whole Gene or Deletions
PANK2 (PANTOTHENATE KINASE 2)	HYPERPHENYLALANINEMIA » FOLLING DISEASE	Whole Gene or Exons 3, 4, 5, 7 and 12
PARKIN (PARK2)	HALLERVORDEN-SPATZ DISEASE » PANTOTHENATE KINASE-ASSOCIATED NEURODEGENERATION, PKAN »	
	NEUROAXONAL DYSTROPHY, JUVENILE-ONSET HARP SYNDROME (HYPOPREBETALIPOPROTEINEMIA, ACANTHOCYTOSIS, RETINITIS PIGMENTOSA, AND PALLIDAL DEGENERATION)	
	PARKINSON DISEASE, TYPE 2, PARK2 (AUTOSOMAL RECESSIVE)	Whole Gene or Deletions

PAX2 (PAIRED BOX GENE 2)	RENAL-COLOBOMA SYNDROME » PAPHILORENAL SYNDROME RENAL HYPOPLASIA	
PAX3 (PAIRED BOX GENE 3)	WAARDENBURG SYNDROME, TYPE 1, WS1	Whole Gene or Deletion Analysis (MLPA)
PAX6 (PAIRED BOX GENE 6)	WAARDENBURG SYNDROME, TYPE 3, WS3 » KLEIN-WAARDENBURG SYNDROME ANIRIDIA, TYPE 2, AN2 KERATITIS ECTOPIA PUPILLAE FOVEAL HYPOPLASIA AND PRESENILE CATARACT SYNDROME PETERS ANOMALY CATARACT, CONGENITAL, WITH LATE-ONSET CORNEAL DYSTROPHY	Whole Gene or Deletion Analysis (MLPA) Whole Gene or Deletion Analysis (MLPA) Whole Gene or Deletion Analysis (MLPA) Whole Gene or Deletion Analysis (MLPA) Whole Gene or Deletion Analysis (MLPA) Whole Gene or Deletion Analysis (MLPA)
PAX8 (PAIRED BOX GENE 8)	HYPOTHYROIDISM, CONGENITAL, NONGOITROUS, TYPE 2 » THYROID DYSGENESIS » THYROID AGENESIS » THYROID HYPOPLASIA » THYROID, ECTOPIC » HYPOTHYROIDISM, CONGENITAL, DUE TO THYROID DYSGENESIS » HYPOTHYROIDISM, ATHYREOTIC » THYROTROPIN RESISTANCE	
PAX9 (PAIRED BOX GENE 9)	HYPODONTIA/OLIGODONTIA 3 » TOOTH AGENESIS, SELECTIVE, 3	
PCCA (PROPIONYL-CoA CARBOXYLASE, ALPHA SUBUNIT)	PROPIONIC ACIDEMIA » PROPIONYL-CoA CARBOXYLASE DEFICIENCY » GLYCINEMIA, KETOTIC » HYPERGLYCINEMIA WITH KETOACIDOSIS AND LEUKOPENIA » KETOTIC HYPERGLYCINEMIA	PCCA and PCCB

PCCB (PROPIONYL-CoA CARBOXYLASE, BETA SUBUNIT)	PROPIONIC ACIDEMIA » PROPIONYL-CoA CARBOXYLASE DEFICIENCY » GLYGINEMIA, KETOTIC » HYPERGLYGINEMIA WITH KETOACIDOSIS AND LEUKOPENIA » KETOTIC HYPERGLYGINEMIA	PCCA and PCCB
PDCD10 (PROGRAMMED CELL DEATH 10, TFAR15)	CEREBRAL CAVERNOUS MALFORMATIONS 3, CCM3	See also CCM1, CCM2 and PDCD10
PDHA1 (PYRUVATE DEHYDROGENASE COMPLEX, E1-ALPHA POLYPEPTIDE 1)	CYTOCHROME c OXIDASE DEFICIENCY » COX DEFICIENCY » COMPLEX 4, MITOCHONDRIAL RESPIRATORY CHAIN, DEFICIENCY OF LEIGH SYNDROME PYRUVATE DECARBOXYLASE DEFICIENCY » ATAXIA, INTERMITTENT, WITH ABNORMAL PYRUVATE METABOLISM » PYRUVATE DEHYDROGENASE DEFICIENCY » PDH DEFICIENCY » ATAXIA, INTERMITTENT, WITH PYRUVATE DEHYDROGENASE, OR DECARBOXYLASE, DEFICIENCY » ATAXIA WITH LACTIC ACIDOSIS I LACTIC ACIDEMIA, THIAMINE-RESPONSIVE » THIAMINE-RESPONSIVE LACTIC ACIDEMIA » PYRUVATE DEHYDROGENASE E1-ALPHA DEFICIENCY	
PEX1 (PEROXISOME BIOGENESIS FACTOR 1)	ZELLWEGER SYNDROME » CEREBROHEPATORENAL SYNDROME	Exons 13 and 15
	ADRENOLEUKODYSTROPHY, AUTOSOMAL NEONATAL FORM, NALD	Exons 13 and 15
	REFSUM DISEASE, INFANTILE FORM » INFANTILE PHYTANIC ACID STORAGE DISEASE	Exons 13 and 15
PEX2, PEX10, PEX12 and PEX26	ZELLWEGER SYNDROME » CEREBROHEPATORENAL SYNDROME	PEX2 (Exon 4), PEX10 (Exons 4 and 5), PEX12 (Exons 2 and 3) and PEX26 (Exons 2 and 3)
	ADRENOLEUKODYSTROPHY, AUTOSOMAL NEONATAL FORM, NALD	PEX2 (Exon 4), PEX10 (Exons 4 and 5), PEX12 (Exons 2 and 3) and PEX26 (Exons 2 and 3)
	REFSUM DISEASE, INFANTILE FORM » INFANTILE PHYTANIC ACID STORAGE DISEASE	PEX2 (Exon 4), PEX10 (Exons 4 and 5), PEX12 (Exons 2 and 3) and PEX26 (Exons 2 and 3)

PEX7 (PEROXISOME BIOGENESIS FACTOR 7, PEROXIN 7, PEROXISOMAL PTS2 RECEPTOR)	RHIZOMELIC CHONDRODYSPLASIA PUNCTATA, TYPE 1 » CHONDRODYSTROPHIA CALCIFICANS PUNCTATA	
PFKM (PHOSPHOFRUCTOKINASE, MUSCLE TYPE)	GLYCOGEN STORAGE DISEASE, TYPE 7 » GSD TYPE 7 » TARUI DISEASE » MUSCLE PHOSPHOFRUCTOKINASE DEFICIENCY	
PHEX	HYPOPHOSPHATEMIC RICKETS (X-LINKED) » VITAMIN D-RESISTANT RICKETS (X-LINKED) » RICKETS, HYPOPHOSPHATEMIC RICKETS (X-LINKED)	Whole Gene Sequencing or Deletion-Duplication
PHKA1 (PHOSPHORYLASE KINASE, MUSCLE, ALPHA-1 SUBUNIT)	GLYCOGEN STORAGE DISEASE, TYPE 9D » GSD TYPE 9D » MUSCLE PHOSPHORYLASE KINASE DEFICIENCY » MUSCLE GLYCOGENOSIS (X-LINKED)	
PHKA2 (PHOSPHORYLASE KINASE, MUSCLE, ALPHA-2 SUBUNIT)	GLYCOGEN STORAGE DISEASE, TYPE 9A » GSD TYPE 9A » LIVER PHOSPHORYLASE KINASE DEFICIENCY » LIVER GLYCOGENOSIS (X-LINKED) » GLYCOGEN STORAGE DISEASE, TYPE 8 » GSD TYPE 8	
PHKG1 (PHOSPHORYLASE KINASE, MUSCLE, GAMMA-1)		
PHKG2 (PHOSPHORYLASE KINASE, TESTIS/LIVER, GAMMA-2)	GLYCOGEN STORAGE DISEASE, TYPE 9C » GSD TYPE 9C » GLYCOGENOSIS, HEPATIC (AUTOSOMAL RECESSIVE) CIRRHOSIS DUE TO LIVER PHOSPHORYLASE KINASE DEFICIENCY	
PHOX2B	CONGENITAL CENTRAL HYPOVENTILATION SYNDROME » ONDINE CURSE » ONDINE-HIRSCHSPRUNG DISEASE » HADDAD SYNDROME HIRSCHSPRUNG DISEASE WITH NEUROBLASTOMA	
PI (ANTITRYPSINE, PROTEASE INHIBITOR)	ANTITRYPSINE DEFICIENCY, AAT	Alleles M, S and Z
PINK1	PARKINSON DISEASE, TYPE 6, PARK6 (AUTOSOMAL RECESSIVE)	Whole Gene or Deletions
PITX2 (PAIRED-LIKE HOMEODOMAIN TRANSCRIPTION FACTOR 2, PTX2)	RIEGER SYNDROME, TYPE 1, RIEG1 » AXENFELD-RIEGER SYNDROME	Whole Gene Sequencing or Deletion-Duplication Testing

	IRIDOGONIODYSGENESIS SYNDROME, TYPE 2, IRID2 » IRIS HYPOPLASIA WITH EARLY-ONSET GLAUCOMA (AUTOSOMAL DOMINANT), IHGA PETERS ANOMALY	Whole Gene Sequencing or Deletion-Duplication Testing
PKD1 (POLYCYSTIN 1) AND PKD2 (POLYCYSTIN 2)	ADULT POLYCYSTIC KIDNEYS, PKD, ADPKD » POTTER TYPE 3 POLYCYSTIC KIDNEY DISEASE	Whole Gene Sequencing or Deletion-Duplication Testing 2 Genes
PKHD1 (FIBROCYSTIN, POLYDUCTIN)	POLYCYSTIC KIDNEY DISEASE (AUTOSOMAL RECESSIVE), ARPKD » POLYCYSTIC KIDNEY DISEASE, INFANTILE, TYPE 1 » POLYCYSTIC KIDNEY AND HEPATIC DISEASE » CAROLI DISEASE » RENAL-HEPATIC-PANCREATIC DYSPLASIA	Various Exons (80% of Mutations)
PKP2 (PLAKOPHILIN 2)	ARRHYTHMOGENIC RIGHT VENTRICULAR DYSPLASIA, FAMILIAL, 9, ARVD9 » ARRHYTHMOGENIC RIGHT VENTRICULAR CARDIOMYOPATHY 9	
PLA2G6 (PHOSPHOLIPASE A2, GROUP 6)	HALLERVORDEN-SPATZ DISEASE » PANTOTHENATE KINASE-ASSOCIATED NEURODEGENERATION, PKAN » NEUROAXONAL DYSTROPHY, JUVENILE-ONSET KARAK SYNDROME NEUROAXONAL DYSTROPHY, INFANTILE (INAD)	
PLEC1 (PLECTIN 1)	NEURODEGENERATION WITH BRAIN IRON ACCUMULATION, PLA2G6-RELATED LIMB-GIRDLE MUSCULAR DYSTROPHY WITH EPIDERMOLYSIS BULLOSA SIMPLEX EPIDERMOLYSIS BULLOSA SIMPLEX, OGNA TYPE » EPIDERMOLYSIS BULLOSA SIMPLEX 1, EBS1	
PLOD1 (PROCOLLAGEN-LYSINE, 2-OXOGLUTARATE 5-DIOXYGENASE; LYSYL HYDROXYLASE)	EHLERS-DANLOS TYPE 6, EDS 6 » EHLERS-DANLOS SYNDROME, KYPHOSCOLIOTIC TYPE	Whole Gene Sequencing or Duplication Exons 10-16
PLP1 (PROTEOLIPID PROTEIN 1, PLP)	PELIZAEUS-MERZBACHER DISEASE, PMD FAMILIAL SPASTIC PARAPLEGIA 2 (X-LINKED), SPG2	Whole Gene or Duplication Preferably EDTA-blood Whole Gene or Duplication Preferably

PMP22 (PERIPHERAL MYELIN PROTEIN)	CHARCOT-MARIE-TOOTH DISEASE, DEMYELINATING, TYPE 1A, CMT1A » HEREDITARY MOTOR AND SENSORY NEUROPATHY 1A, HMSN1A	EDTA-blood Whole Gene or Duplication
	HEREDITARY NEUROPATHY WITH LIABILITY TO PRESSURE PALSIES, HNPP » TOMACULOUS NEUROPATHY	Whole Gene or Deletion
	CHARCOT-MARIE-TOOTH DISEASE, DEMYELINATING, TYPE 4F, CMT4F » DEJERINE-SOTTAS HYPERTROPHIC NEUROPATHY	Whole Gene
	CHARCOT-MARIE-TOOTH DISEASE AND DEAFNESS	Whole Gene or Duplication
PNPO (PYRIDOXAMINE 5-PRIME-PHOSPHATE OXIDASE, PYRIDOXAMINE-PHOSPHATE OXIDASE)	PYRIDOXAMINE 5-PRIME-PHOSPHATE OXIDASE DEFICIENCY » PNPO DEFICIENCY » EPILEPTIC ENCEPHALOPATHY, NEONATAL, PNPO-RELATED	
POLG (POLYMERASE, DNA, GAMMA)	PROGRESSIVE EXTERNAL OPHTHALMOPLEGIA WITH MITOCHONDRIAL DNA DELETIONS (AUTOSOMAL DOMINANT), 1 » PROGRESSIVE EXTERNAL OPHTHALMOPLEGIA (AUTOSOMAL DOMINANT), 1 » PROGRESSIVE EXTERNAL OPHTHALMOPLEGIA WITH HYPOGONADISM » OCULAR MYOPATHY WITH HYPOGONADISM » MYOPATHY, CATARACT, HYPOGONADISM SYNDROME PROGRESSIVE EXTERNAL OPHTHALMOPLEGIA WITH MITOCHONDRIAL DNA DELETIONS (AUTOSOMAL RECESSIVE) » SPINOCEREBELLAR ATAXIA WITH EPILEPSY » ALPERS SYNDROME SENSORY ATAXIC NEUROPATHY, DYSARTHRIA, AND OPHTHALMOPARESIS, SANDO » SPINOCEREBELLAR ATAXIA WITH EPILEPSY ALPERS DIFFUSE DEGENERATION OF CEREBRAL GRAY MATTER WITH HEPATIC CIRRHOSIS » ALPERS PROGRESSIVE INFANTILE POLIODYSTROPHY » ALPERS SYNDROME » ALPERS-HUTTENLOCHER SYNDROME » NEURONAL DEGENERATION OF CHILDHOOD WITH LIVER DISEASE, PROGRESSIVE	

POMC (PROOPIOMELANOCORTIN)	PROOPIOMELANOCORTIN DEFICIENCY » OBESITY, EARLY-ONSET, ADRENAL INSUFFICIENCY, AND RED HAIR	
POR (CYTOCHROME P450 OXIDOREDUCTASE)	ANTLEY-BIXLER SYNDROME » TRAPEZOIDOCEPHALY-SYNOSTOSIS SYNDROME » MULTISYNOSTOTIC OSTEODYSGENESIS WITH LONG BONE FRACTURES » OSTEODYSGENESIS, MULTISYNOSTOTIC, WITH FRACTURES207410. 400DISORDERED STEROIDOGENESIS, ISOLATED ADRENAL HYPERPLASIA, CONGENITAL, DUE TO APPARENT COMBINED P450C17 AND P450C21 DEFICIENCY » ADRENAL HYPERPLASIA, CONGENITAL, DUE TO DEFECTS IN SEVERAL STEROID-BIOSYNTHETIC ENZYMES » MALE PSEUDOHERMAPHRODITISM DUE TO MULTIPLE MICROSOMAL MIXED FUNCTION OXIDASE DEFICIENCY	
POU1F1 (POU DOMAIN, CLASS 1, TRANSCRIPTION FACTOR 1, GROWTH HORMONE FACTOR 1, PIT1)	PITUITARY HORMONE DEFICIENCY, COMBINED	
POU3F4 (POU DOMAIN, CLASS 3, TRANSCRIPTION FACTOR 4)	DEAFNESS, CONDUCTIVE, WITH STAPES FIXATION, DFN3 » PERILYMPHATIC GUSHER-DEAFNESS SYNDROME	
PPARG (PEROXISOME PROLIFERATOR-ACTIVATED RECEPTOR-GAMMA, PPARG1, PPARG2, PPARG3, PAX8/PPARG FUSION GENE)	OBESITY DIABETES MELLITUS, NONINSULIN-DEPENDENT » DIABETES MELLITUS, TYPE 2 » NONINSULIN-DEPENDENT DIABETES MELLITUS » MATURITY-ONSET DIABETES » INSULIN RESISTANCE, SUSCEPTIBILITY TO	2 Mutations: P12A and P115Q 2 Mutations: P12A and P115Q

PPP2R2B (PROTEIN PHOSPHATASE 2, REGULATORY SUBUNIT B, BETA)	SPINOCEREBELLAR ATAXIA 12, SCA12	
PPT (PALMITOYL-PROTEIN ESTERASE)	See CLN1	
PQBP1 (POLYGLUTAMINE-BINDING PROTEIN 1)	MENTAL RETARDATION, SYNDROMIC (X -LINKED), TYPE3, MRXS3 » SUTHERLAND-HAAN MENTAL RETARDATION SYNDROME (X-LINKED) » MENTAL RETARDATION, NONSPECIFIC (X-LINKED), TYPE55, MRX55	See also Mental Retardation Panel
PRF1 (PERFORIN 1, PORE-FORMING PROTEIN)	HEMOPHAGOCYTIC LYMPHOHISTIOCYTOSIS, FAMILIAL, TYPE 2, FHL2	See also PRF1, STX11, UNC13D
PRF1, STX11, UNC13D	HEMOPHAGOCYTIC LYMPHOHISTIOCYTOSIS, FAMILIAL	
PRKAG2 (PROTEIN KINASE, AMP-ACTIVATED, NONCATALYTIC, GAMMA-2, AMP-ACTIVATED PROTEIN KINASE, NONCATALYTIC, GAMMA-2 AMPK-GAMMA-2) PRKAR1A	CARDIOMYOPATHY, FAMILIAL HYPERTROPHIC, WITH WOLFF-PARKINSON-WHITE SYNDROME GLYCOGEN STORAGE DISEASE OF HEART, LETHAL CONGENITAL » PHOSPHORYLASE KINASE DEFICIENCY OF HEART » GLYCOGEN STORAGE DISEASE OF HEART CARNEY COMPLEX, TYPE 1 » PRIMARY PIGMENTED NODULAR ADRENOCORTICAL DISEASE CARDIAC MYXOMA	
PRKCG (PROTEIN KINASE C, GAMMA, PKCC)	SPINOCEREBELLAR ATAXIA 14, SCA14	
PRNP (PRION PROTEIN)	CREUTZFELDT-JAKOB DISEASE, CJD GERSTMANN-STRAUSSLER DISEASE, GSD INSOMNIA-DYSAUTONOMIA » FAMILIAL FATAL INSOMNIA	DNA obtained after phenol extraction DNA obtained after phenol extraction DNA obtained after phenol extraction
PROC (PROTEIN C)	HUNTINGTON DISEASE-LIKE 1, HDL1 THROMBOEMBOLISM	DNA obtained after phenol extraction

PROP1 (PROPHET OF PIT1, PAIRED-LIKE HOMEODOMAIN TRANSCRIPTION FACTOR)	PITUITARY DWARFISM 3 » PANHYPOPITUITARISM » ATELIOTIC DWARFISM WITH HYPOGONADISM » HANHART DWARFISM PITUITARY HORMONE DEFICIENCY, COMBINED » PITUITARY HORMONE DEFICIENCY, COMBINED WITH RIGID CERVICAL SPINE » PITUITARY HORMONE DEFICIENCY, COMBINED	
PROS1 (PROTEIN S) PRPH2 (PERIPHERIN 2, MOUSE, HOMOLOG OF, RDS)	THROMBOEMBOLISM RETINITIS PIGMENTOSA 7, RP7 FUNDUS ALBIPUNCTATUS » RETINITIS PUNCTATA ALBESCENS MACULAR DYSTROPHY MACULAR DYSTROPHY, VITELLIFORM, ADULT-ONSET » FOVEOMACULAR DYSTROPHY, ADULT-ONSET PATTERNED DYSTROPHY OF RETINAL PIGMENT EPITHELIUM » MACULAR DYSTROPHY, BUTTERFLY-SHAPED PIGMENTARY » BUTTERFLY DYSTROPHY OF RETINAL PIGMENT EPITHELIUM STARGARDT DISEASE, TYPE 1 » MACULAR DEGENERATION, JUVENILE » FUNDUS FLAVIMACULATUS » MACULAR DYSTROPHY WITH FLECKS, TYPE 1	
PRSS1 (PROTEASE, SERINE 1) PRX (PERIAXIN)	PANCREATITIS, HEREDITARY, PCTT CHARCOT-MARIE-TOOTH DISEASE, DEMYELINATING, TYPE 4F, CMT4F » DEJERINE-SOTTAS HYPERTROPHIC NEUROPATHY » HEREDITARY MOTOR AND SENSORY NEUROPATHY TYPE 3, HMSN3	Exons 1-3 Includes SPINK1 (Exon 3)
PSAP (PROSAPOSIN)	METACHROMATIC LEUKODYSTROPHY DUE TO SAP1 DEFICIENCY » GAUCHER DISEASE DUE TO SAP2 DEFICIENCY » PROSAPOSIN DEFICIENCY	
PSEN1 (PRESENILIN 1, PS1) PSEN2 (PRESENILIN 2, PS2)	ALZHEIMER DEMENTIA, EARLY-ONSET, TYPE 3, AD3 ALZHEIMER DEMENTIA, EARLY-ONSET, TYPE 4, AD4	

PSTPIP1 (PROLINE/ SERINE/ THREONINE PHOSPHATASE-INTERACTING PROTEIN 1)	PYOGENIC STERILE ARTHRITIS, PYODERMA GANGRENOSUM, AND ACNE » PAPA SYNDROME » FAMILIAL RECURRENT ARTHRITIS	
PTCH (PATCHED, PTC)	BASAL CELL NEVUS SYNDROME » GORLIN SYNDROME HOLOPROSENCEPHALY 7, HPE7	
PTEN	BANNAYAN-MYHRE-RILEY-RUVALCAVA-SMITH-ZONANA SYNDROME COWDEN DISEASE » LHERMITTE-DUCLOS DISEASE OLIGODENDROGLIOMA VATER-HYDROCEPHALUS SYNDROME	
PTPN11	NOONAN SYNDROME, TYPE 1 LEOPARD SYNDROME, TYPE 1 JUVENILE MYELOMONOCYTIC LEUKEMIA, JMML	Whole Gene Whole Gene Exons 3 and 13
PWS	PRADER-WILLI SYNDROME	Methylation or Deletion of Imprinting Centre
PYGL (GLYCOGEN PHOSPHORYLASE, LIVER)	GLYCOGEN STORAGE DISEASE, TYPE 6 » GSD TYPE 6 » HERS DISEASE » GLYCOGEN PHOSPHORYLASE DEFICIENCY	
PYGM (GLYCOGEN PHOSPHORYLASE, MUSCLE, MYOPHOSPHORYLASE)	GLYCOGEN STORAGE DISEASE, TYPE 5 » GSD TYPE 5 » MCARDLE DISEASE » MYOPHOSPHORYLASE DEFICIENCY	
RAB7 (RAS-ASSOCIATED PROTEIN RAB7)	CHARCOT-MARIE-TOOTH DISEASE, AXONAL, TYPE 2B, CMT2B	
RAF1 (V-RAF-1 MURINE LEUKEMIA VIRAL ONCOGENE HOMOLOG 1)	LEOPARD SYNDROME, TYPE 2 NOONAN SYNDROME, TYPE 5	
RAG1 (RECOMBINATION-ACTIVATING GENE 1) RAG2 (RECOMBINATION-ACTIVATING GENE 2)	SEVERE COMBINED IMMUNODEFICIENCY (AUTOSOMAL RECESSIVE), T CELL-NEGATIVE, B CELL-NEGATIVE, NK CELL-POSITIVE » SCID, T CELL-NEGATIVE, B CELL-NEGATIVE, NK CELL-POSITIVE OMENN SYNDROME » RETICULOENDOTHELIOSIS, FAMILIAL, WITH EOSINOPHILIA » SEVERE COMBINED IMMUNODEFICIENCY WITH HYPEREOSINOPHILIA	Testing of both RAG Genes Testing of both RAG Genes

RAI1 (RETINOIC ACID-INDUCED GENE 1, RAI1)	SMITH-MAGENIS SYNDROME	
RAPSN (RAPSIN, RECEPTOR-ASSOCIATED PROTEIN OF THE SYNAPSE, 43-KD)	MYASTHENIC SYNDROME, CONGENITAL, ASSOCIATED WITH ACETYLCHOLINE RECEPTOR DEFICIENCY » MYASTHENIC SYNDROME, CONGENITAL, TYPE 1d » MYASTHENIC SYNDROME, CONGENITAL, ASSOCIATED WITH FACIAL DYSMORPHISM FETAL AKINESIA DEFORMATION SEQUENCE » PENASHOKEIR SYNDROME, TYPE 1 » ARTHROGRYPOSIS MULTIPLEX CONGENITA WITH PULMONARY HYPOPLASIA	
RB1	RETINOBLASTOMA, RB1	
RDH5 (RETINOL DEHYDROGENASE 5)	FUNDUS ALBIPUNCTATUS » RETINITIS PUNCTATA ALBESCENS	
RDH12 (RETINOL DEHYDROGENASE 12)	LEBER CONGENITAL AMAUROSIS, TYPE 3, LCA3 » AMAUROSIS CONGENITA OF LEBER 3	
RDS, HPRP3 and PRPF8	RETINITIS PIGMENTOSA (AUTOSOMAL DOMINANT)	Exons 1-3 from RDS, exon 11 from HPRP3 and exon 42 from PRPF8, detecting 8-15% of autosomal dominant cases
RECQL2 (RECQ PROTEIN-LIKE 2)	WERNER SYNDROME	
RECQL3 (RECQ2)	BLOOM SYNDROME » (See also Molecular Screening Tests)	1 Mutation: 6del / 7ins
RECQL4 (RECQ PROTEIN-LIKE 4, DNA HELICASE, RECQ-LIKE, TYPE 4)	ROTHMUND-THOMSON SYNDROME » POIKILODERMA ATROPHICANS AND CATARACT RAPADILINO SYNDROME BALLER-GEROLD SYNDROME » CRANIOSYNOSTOSIS WITH RADIAL DEFECTS » CRANIOSYNOSTOSIS-RADIAL APLASIA SYNDROME	
RET (RET KINASE)	HIRSCHSPRUNG DISEASE, TYPE 1 » AGANGLIONIC MEGACOLON RENAL AGENESIS MEDULLARY THYROID CARCINOMA, MTC	Whole Gene Whole Gene Exons 10,11, 13-16

	MULTIPLE ENDOCRINE NEOPLASIA, TYPE 2A, MEN2A	Exons 10,11, 13-16
	MULTIPLE ENDOCRINE NEOPLASIA, TYPE 2B, MEN2B	Whole Gene or Exons 10,11, 13-16
RHCE (RHESUS BLOOD GROUP CcEe ANTIGENS)	HEMOLYTIC DISEASE, NEWBORN	3 Mutations: P226A, I60L, S103P
RHD (RHESUS BLOOD GROUP D ANTIGEN)	HEMOLYTIC DISEASE, NEWBORN	Deletion
RHO (RHODOPSIN, OPSIN 2, OPN2)	RETINITIS PIGMENTOSA 4, RP4 » NIGHT BLINDNESS, CONGENITAL STATIONARY	See also RHO, RDS, RP1, HPRP3 and PRPF8
RMRP (MITOCHONDRIAL RNA-PROCESSING ENDORIBONUCLEASE, RNA COMPONENT OF)	CARTILAGE-HAIR HYPOPLASIA » METAPHYSEAL CHONDRODYSPLASIA, MCKUSICK TYPE METAPHYSEAL DYSPLASIA WITHOUT HYPOTRICHOSIS » CARTILAGE-HAIR HYPOPLASIA-LIKE SKELETAL DYSPLASIA WITHOUT HYPOTRICHOSIS OR IMMUNODEFICIENCY » CARTILAGE-HAIR HYPOPLASIA VARIANT	
RNASEH2A (RIBONUCLEASE H2, LARGE SUBUNIT)	AICARDI-GOUTIERES SYNDROME 4	
RNASEH2B (RIBONUCLEASE H2, SUBUNIT B)	AICARDI-GOUTIERES SYNDROME 2	
RNASEH2C (RIBONUCLEASE H2, SUBUNIT C)	AICARDI-GOUTIERES SYNDROME 3	
ROR2 (NTRKR2)	ROBINOW SYNDROME (AUTOSOMAL RECESSIVE) » COVESDEM SYNDROME BRACHYDACTYLY, TYPE B, BDB	Exons 8 and 9
RP2	RETINITIS PIGMENTOSA 2 (X-LINKED)	
RPE65 (RETINAL PIGMENT EPITHELIUM-SPECIFIC PROTEIN, 65-KD)	RETINITIS PIGMENTOSA 20, RP20 LEBER CONGENITAL AMAUROSIS, TYPE 2, LCA2	
RPGR (RETINITIS PIGMENTOSA GTPase REGULATOR)	RETINITIS PIGMENTOSA 3, RP3 » CHOROIDORETINAL DEGENERATION WITH RETINAL REFLEX IN HETEROZYGOUS WOMEN	Exons 1-15 and ORF15
	RETINITIS PIGMENTOSA 15, RP15 » CONE-ROD DEGENERATION (X-LINKED)	Exons 1-15 and ORF15

	CONE-ROD DYSTROPHY, (X-LINKED) TYPE 1, CORDX1 » ACHROMATOPSIA, INCOMPLETE (X-LINKED)	Exons 1-15 and ORF15
RPGRIP1 (RETINITIS PIGMENTOSA GTPase REGULATOR-INTERACTING PROTEIN, RPGR-INTERACTING PROTEIN) RPGRIP1L (RPGRIP1-LIKE)	LEBER CONGENITAL AMAUROSIS, TYPE 1, LCA1 CONE-ROD DYSTROPHY, TYPE 9, CORD9 JOUBERT SYNDROME, TYPE 7 MECKEL SYNDROME, TYPE 5	
RPS19 RSK2 (RPS6KA3)	DIAMOND-BLACKFAN ANEMIA, DBA COFFIN-LOWRY SYNDROME, CLS MENTAL RETARDATION, NONSPECIFIC (X-LINKED), TYPE19, MRX19	
RUNX1 (RUNT-RELATED TRANSCRIPTION FACTOR 1, ACUTE MYELOID LEUKEMIA 1 GENE, AML1 CORE-BINDING FACTOR, RUNT DOMAIN, ALPHA SUBUNIT 2, CBFA2)	PLATELET DISORDER, FAMILIAL, WITH ASSOCIATED MYELOID MALIGNANCY » PLATELET DISORDER, ASPIRIN-LIKE » THROMBOCYTOPENIA, FAMILIAL, WITH PROPENSITY TO ACUTE MYELOGENOUS LEUKEMIA	
RUNX 2 (CBFA 1) RYR1 (RYANODINE RECEPTOR 1)	CLEIDOCRANIAL DYSPLASIA, CCD MALIGNANT HYPERTHERMIA SUSCEPTIBILITY 1, MHS1 » KING SYNDROME	41 Exons: Exons 2, 3, 6, 8-15, 17, 34, 38-40, 42-48, 51, 52, 67, 71, 73, 85, 86, 90, 94, 95, 97, 98-104
RYR2 (RYANODINE RECEPTOR 2)	VENTRICULAR TACHYCARDIA, CATECHOLAMINERGIC POLYMORPHIC » VENTRICULAR TACHYCARDIA, STRESS-INDUCED POLYMORPHIC ARRHYTHMOGENIC RIGHT VENTRICULAR DYSPLASIA, FAMILIAL, TYPE 2 » ARRHYTHMOGENIC RIGHT VENTRICULAR CARDIOMYOPATHY, TYPE 2	

SALL1 (SAL-LIKE 1)	TOWNES-BROCKS SYNDROME » RENAL-EAR-ANAL-RADIAL SYNDROME » REAR SYNDROME » ANUS, IMPERFORATE, WITH HAND, FOOT, AND EAR ANOMALIES » DEAFNESS, SENSORINEURAL, WITH IMPERFORATE ANUS AND THUMB ANOMALIES » TOWNES-BROCKS-BRANCHIOOTORENAL-LIKE SYNDROME	Whole Gene or Deletions
SALL4 (SAL-LIKE 4)	DUANE-RADIAL RAY SYNDROME » OKIHIRO SYNDROME » DUANE ANOMALY WITH RADIAL RAY ABNORMALITIES AND DEAFNESS » DR SYNDROME » ACRORENOOCULAR SYNDROME	Whole Gene or Deletions
SBDS	SHWACHMAN-DIAMOND SYNDROME, SDS » PANCREATIC INSUFFICIENCY AND BONE MARROW DYSFUNCTION » CONGENITAL LIPOMATOSIS OF PANCREAS	
SCA1, SCA2, SCA3, SCA6, SCA7	SPINOCEREBELLAR ATAXIA » AUTOSOMAL DOMINANT CEREBELLAR ATAXIA, ADCA	Screening for Repeats in 5 SCA Genes
SCA8, SCA10, SCA12, SCA17	SPINOCEREBELLAR ATAXIA » AUTOSOMAL DOMINANT CEREBELLAR ATAXIA, ADCA	Screening for Repeats in 4 SCA Genes
SCN1A (SODIUM CHANNEL, NEURONAL TYPE 1, ALPHA SUBUNIT)	GENERALIZED EPILEPSY WITH FEBRILE SEIZURES PLUS, GEFS+ » GENERALIZED EPILEPSY WITH FEBRILE SEIZURES PLUS, TYPE 2, GEFS+, TYPE 2 » FEBRILE SEIZURES ASSOCIATED WITH AFEBRILE SEIZURES SEVERE MYOCLONIC EPILEPSY OF INFANCY » DRAVET SYNDROME INTRACTABLE CHILDHOOD EPILEPSY WITH GENERALISED TONIC-CLONIC SEIZURES (ICEGTC) BORDERLINE SMEI (SMEIB)	
SCN1B (SODIUM CHANNEL, VOLTAGE-GATED, TYPE 1, BETA SUBUNIT)	GENERALIZED EPILEPSY WITH FEBRILE SEIZURES PLUS, GEFS+ » GENERALIZED EPILEPSY WITH FEBRILE SEIZURES PLUS, TYPE 2, GEFS+, TYPE 2 » FEBRILE SEIZURES ASSOCIATED WITH AFEBRILE SEIZURES	
SCN4A	HYPERKALEMIC PERIODIC PARALYSIS, HYPP »	Exons 9, 12-14, 19, 21-24

SCN4A and CACNA1S (CACNL1A3)	<p>MYOTONIA CONGENITA, ATYPICAL PARAMYOTONIA CONGENITA (VON EULENBURG) HYPOKALEMIC PERIODIC PARALYSIS, HOKPP PARAMYOTONIA WITHOUT COLD PARALYSIS HYPOKALEMIC PERIODIC PARALYSIS, HOKPP</p>	<p>Exons 9, 12-14, 19, 21-24 Exons 9, 12-14, 19, 21-24 Exons 9, 12-14, 19, 21-24 4 Mutations in CACNA1S: R528G, R528H, R1239H, R1239G and 5 Mutations in SCN4A: R672S, R672H, R672G, R672C, R669H</p>
SCN5A	<p>LONG QT SYNDROME 3, LQT3 » ROMANO-WARD SYNDROME BRUGADA SYNDROME IDIOPATHIC VENTRICULAR FIBRILLATION HEART BLOCK, FAMILIAL SUDDEN INFANT DEATH SYNDROME, SIDS PSEUDOHYPOALDOSTERONISM, TYPE 1 (AUTOSOMAL RECESSIVE)</p>	<p>See also LONG QT PANEL 1 and LONG QT PANEL 2 and LONG QT PANEL 3</p>
SCNN1A (SODIUM CHANNEL, NONVOLTAGE-GATED 1, ALPHA SUBUNIT, SCNN1, SCNEA)	<p>LIDDLE SYNDROME » PSEUDOALDOSTERONISM PSEUDOHYPOALDOSTERONISM, TYPE 1 (AUTOSOMAL RECESSIVE)</p>	
SCNN1B	<p>LIDDLE SYNDROME » PSEUDOALDOSTERONISM PSEUDOHYPOALDOSTERONISM, TYPE 1 (AUTOSOMAL RECESSIVE)</p>	
SCNN1G (SODIUM CHANNEL, NONVOLTAGE-GATED 1, GAMMA SUBUNIT, SCNEG)	<p>LIDDLE SYNDROME » PSEUDOALDOSTERONISM PSEUDOHYPOALDOSTERONISM, TYPE 1 (AUTOSOMAL RECESSIVE) HEPATIC FAILURE, EARLY-ONSET, AND NEUROLOGIC DISORDER DUE TO CYTOCHROME c OXIDASE DEFICIENCY</p>	
SCO2	<p>CYTOCHROME c OXIDASE DEFICIENCY » COX DEFICIENCY » COMPLEX 4, MITOCHONDRIAL RESPIRATORY CHAIN, DEFICIENCY OF CARDIOENCEPHALOMYOPATHY, FATAL INFANTILE, DUE TO CYTOCHROME c OXIDASE DEFICIENCY » CYTOCHROME c OXIDASE DEFICIENCY, FATAL INFANTILE, WITH CARDIOENCEPHALOMYOPATHY</p>	

	LEIGH SYNDROME
SDHA (SUCCINATE DEHYDROGENASE COMPLEX, SUBUNIT A, FLAVOPROTEIN, SUCCINATE DEHYDROGENASE 2, FLAVOPROTEIN SUBUNIT; SDH2)	CYTOCHROME c OXIDASE DEFICIENCY » COX DEFICIENCY » COMPLEX 2, MITOCHONDRIAL RESPIRATORY CHAIN, DEFICIENCY OF LEIGH SYNDROME
SDHB (SUCCINATE DEHYDROGENASE 1, SDH1)	PARAGANGLIOMAS, PGL4 » GLOMUS TUMORS, TYPE 4
SDHC (SUCCINATE DEHYDROGENASE 3, SDH3)	PARAGANGLIOMAS, PGL3 » GLOMUS TUMORS, TYPE 3
SDHD (SUCCINATE DEHYDROGENASE 4, SDH4)	PARAGANGLIOMAS, PGL1 » GLOMUS TUMORS, TYPE 1
SEDL (SEDLIN)	SPONDYLOEPIPHYSEAL DYSPLASIA TARDA (X-LINKED), SEDT
SETX (SENATAXIN)	AMYOTROPHIC LATERAL SCLEROSIS 4, JUVENILE, ALS4 » NEURONOPATHY, DISTAL HEREDITARY MOTOR, WITH PYRAMIDAL FEATURES SPINOCEREBELLAR ATAXIA (AUTOSOMAL RECESSIVE), 1 » ATAXIA-OCULOMOTOR APRAXIA 2
SFTPB (SURFACTANT PULMONARY-ASSOCIATED PROTEIN B)	PULMONARY ALVEOLAR PROTEINOSIS
SGCA (SARCOGLYCAN, ALPHA, ADHALIN, DYSTROGLYCAN 2)	MUSCULAR DYSTROPHY, LIMB-GIRDLE, TYPE 2D, LGMD2D » DUCHENNE - LIKE MUSCULAR DYSTROPHY, TYPE 2, AUTOSOMAL RECESSIVE, DMDA2 » ADHALINOPATHY, PRIMARY » MUSCULAR DYSTROPHY, DUCHENNE - LIKE MUSCULAR DYSTROPHY, TYPE 2
SGCB (SARCOGLYCAN, BETA)	MUSCULAR DYSTROPHY, LIMB-GIRDLE, TYPE 2E, LGMD2E
SGCD (SARCOGLYCAN DELTA)	LIMB-GIRDLE MUSCULAR DYSTROPHY 2F, LGMD2F » MUSCULAR DYSTROPHY, LIMB-GIRDLE MUSCULAR DYSTROPHY 2F, LGMD2F DILATED CARDIOMYOPATHY, 1L, CMD1L

SGCE (SARCOGLYCAN, EPSILON)	MYOCLONIC DYSTONIA » MYOCLONUS-DYSTONIA SYNDROME » DYSTONIA, ALCOHOL-RESPONSIVE » DYSTONIA 11, DYT11	
SGCG (SARCOGLYCAN GAMMA)	DUCHENNE-LIKE MUSCULAR DYSTROPHY, TYPE 1 » LIMB GIRDLE MUSCULAR DYSTROPHY 2C, LGMD2C » MUSCULAR DYSTROPHY, DUCHENNE-LIKE MUSCULAR DYSTROPHY, TYPE 1	
SGSH (HEPARAN SULFATE SULFATASE, SULFAMIDASE, N-SULFOGLUCOSAMINE SULFOHYDROLASE)	MUCOPOLYSACCHARIDOSIS TYPE 3A, MPS3A » SANFILIPPO SYNDROME A	
SH2D1A	LYMPHOPROLIFERATIVE SYNDROME » LYMPHOPROLIFERATIVE DISEASE (X-LINKED)	
SH3BP2	CHERUBISM	Exon 2
SH3TC2 (SH3 DOMAIN AND TETRATRICOPEPTIDE REPEAT DOMAIN 2)	CHARCOT-MARIE-TOOTH DISEASE, DEMYELINATING, TYPE 4C, CMT4C (AUTOSOMAL RECESSIVE)	
SH3TC2 (SH3 DOMAIN AND TETRATRICOPEPTIDE REPEAT DOMAIN 2)	CHARCOT-MARIE-TOOTH DISEASE, TYPE 4C, CMT4C » CHARCOT-MARIE-TOOTH DISEASE, DEMYELINATING (AUTOSOMAL RECESSIVE), TYPE 4C	
SHH (SONIC HEDGEHOG)	HOLOPROSENCEPHALY 3, HPE3 SOLITARY MEDIAN MAXILLARY CENTRAL INCISOR	See also SHH, SIX3, TGIF and ZIC2 See also SHH, SIX3, TGIF and ZIC2
SHH, SIX3, TGIF and ZIC2	HOLOPROSENCEPHALY	Screening for Frequent Mutations in 4 Genes
SHOX (SHORT STATURE HOMEBOX)	LERI-WEILL DYSCHONDROSTEOSIS » DYSCHONDROSTEOSIS » MADELUNG DEFORMITY LANGER MESOMELIC DYSPLASIA » DYSCHONDROSTEOSIS, HOMOZYGOUS » MESOMELIC DWARFISM OF THE HYPOPLASTIC ULNA, FIBULA, AND MANDIBLE TYPE SHORT STATURE	Whole Gene or MLPA for Deletions-Duplications of SHOX and PAR1 Whole Gene or MLPA for Deletions-Duplications of SHOX and PAR1 Whole Gene or MLPA for Deletions-Duplications of SHOX and PAR1
SIX3	HOLOPROSENCEPHALY 2, HPE2	See also SHH, SIX3, TGIF and ZIC2

SIX6	MICROPHthalmia, ISOLATED, WITH CATARACT 2
SLC7A7 (SOLUTE CARRIER FAMILY 7, MEMBER 7)	LYSINURIC PROTEIN INTOLERANCE » DIBASICAMINO ACIDURIA, TYPE 2
SLC12A1 (SOLUTE CARRIER FAMILY 12 (SODIUM/POTASSIUM/CHLORIDE TRANSPORTER), MEMBER 1, SODIUM-POTASSIUM-CHLORIDE TRANSPORTER 2, NKCC2)	BARTTER SYNDROME, ANTENATAL, TYPE 1 » HYPOKALEMIC ALKALOSIS WITH HYPERCALCIURIA, ANTENATAL, TYPE 1 » HYPERPROSTAGLANDIN E SYNDROME 1
SLC12A3 (SOLUTE CARRIER FAMILY 12, SODIUM/CHLORIDE TRANSPORTER, MEMBER 3, THIAZIDE-SENSITIVE NA-CL COTRANSPORTER)	GITELMAN SYNDROME » HYPOMAGNESEMIA-HYPOKALEMIA, PRIMARY RENOTUBULAR, WITH HYPOCALCIURIA » POTASSIUM AND MAGNESIUM DEPLETION
SLC12A6	CORPUS CALLOSUM, AGENESIS OF, WITH NEURONOPATHY » CHARLEVOIX DISEASE » ANDERMANN SYNDROME » POLYNEUROPATHY, SENSORIMOTOR, WITH OR WITHOUT AGENESIS OF THE CORPUS CALLOSUM » AGENESIS OF THE CORPUS CALLOSUM WITH PERIPHERAL NEUROPATHY
SLC17A5	SIALURIA, FINNISH TYPE » SALLA DISEASE INFANTILE SIALIC ACID STORAGE DISORDER » SIALURIA, INFANTILE FORM » N-ACETYLNEURAMINIC ACID STORAGE DISEASE » NANA STORAGE DISEASE
SLC22A5 (SOLUTE CARRIER FAMILY 22, ORGANIC CATION TRANSPORTER, MEMBER 5, OCTN2)	CARNITINE DEFICIENCY, SYSTEMIC PRIMARY » SYSTEMIC CARNITINE DEFICIENCY » CARNITINE DEFICIENCY, SYSTEMIC, DUE TO DEFECT IN RENAL REABSORPTION OF CARNITINE » CARNITINE DEFICIENCY, PRIMARY » CARNITINE TRANSPORTER, PLASMA-MEMBRANE, DEFICIENCY OF » CARNITINE UPTAKE DEFECT

<p>SLC22A12 (SOLUTE CARRIER FAMILY 22, URATE TRANSPORTER, MEMBER 12, ORGANIC ANION TRANSPORTER 4-LIKE, OAT4L, URATE TRANSPORTER 1, URAT1</p>	<p>HYPOURICEMIA, RENAL » DALMATIAN HYPOURICEMIA » RENAL HYPOURICEMIA » URIC ACID UROLITHIASIS</p>
<p>SLC25A4 (SOLUTE CARRIER FAMILY 25 (MITOCHONDRIAL CARRIER), MEMBER 4, ADENINE NUCLEOTIDE TRANSLOCATOR 1, ANT1 ADP/ATP TRANSLOCATOR OF SKELETAL MUSCLE, ADP/ATP TRANSLOCASE 1)</p>	<p>PROGRESSIVE EXTERNAL OPHTHALMOPLEGIA WITH MITOCHONDRIAL DNA DELETIONS, (AUTOSOMAL DOMINANT), 2 » PROGRESSIVE EXTERNAL OPHTHALMOPLEGIA (AUTOSOMAL DOMINANT), 2</p>
<p>SLC25A20 (SOLUTE CARRIER FAMILY 25, MEMBER 20, CARNITINE-ACYLCARNITINE TRANSLOCASE, CARNITINE-ACYLCARNITINE CARRIER)</p>	<p>CARNITINE-ACYLCARNITINE TRANSLOCASE DEFICIENCY » CACT DEFICIENCY</p>
<p>SLC26A2 (DTD SULFATE TRANSPORTER, DTDST)</p>	<p>DIASTROPHIC DYSPLASIA , DTD ATELOSTEOGENESIS TYPE 2 » AO TYPE 2 » DE LA CHAPELLE DYSPLASIA » NEONATAL OSSEOUS DYSPLASIA 1 ACHONDROGENESIS TYPE 1B » ACHONDROGENESIS, FRACCARO TYPE EPIPHYSEAL DYSPLASIA, MULTIPLE, TYPE 4, EDM4 » MULTIPLE EPIPHYSEAL DYSPLASIA (AUTOSOMAL RECESSIVE)</p>
<p>SLC26A4 (PENDRIN)</p>	<p>PENDRED SYNDROME, PDS ENLARGED VESTIBULAR AQUEDUCT SYNDROME, EVA DEAFNESS, DFNB4 » DEAFNESS, (AUTOSOMAL RECESSIVE), NEUROSENSORY 4</p>

SLC2A2 (SOLUTE CARRIER FAMILY 2 (FACILITATED GLUCOSE TRANSPORTER, MEMBER 2, GLUT2)	GLYCOGEN STORAGE DISEASE, TYPE 11 » GSD TYPE 11 » FANCONI-BICKEL SYNDROME » HEPATORENAL GLYCOGENOSIS WITH RENAL FANCONI SYNDROME » PSEUDO-PHLORIZIN DIABETES	
SLC2A10 (SOLUTE CARRIER FAMILY 2 - FACILITATED GLUCOSE TRANSPORTER, MEMBER 10, GLUCOSE TRANSPORTER 10, GLUT10)	ARTERIAL TORTUOSITY SYNDROME, ATS	
SLC3A1 (RBAT)	CYSTINURIA TYPE 1	Whole Gene 10 mutations in SLC3A1: Thr216Met, Ser217Arg, Arg365Trp, Arg365Leu, Arg362Cys, Met467Thr, Met467Lys, 1500+1G/T, Glu483X duplication of exons 5-9 includes 7 mutations in SLC7A9: Gly105Arg, Thr123Met, Phe140Ser, Ala182Thr, 747delG, 244delE, Arg333Trp
SLC3A1 (RBAT) (analyzed together with SLC7A9)	CYSTINURIA TYPE 1	
SLC40A1 (FERROPORTIN 1, IREG1, SLC11A3)	HEMOCHROMATOSIS, TYPE 4, HFE4	
SLC45A2 (SOLUTE CARRIER FAMILY 45, MEMBER 2, MATP (MEMBRANE-ASSOCIATED TRANSPORTER PROTEIN)	OCULOCUTANEOUS ALBINISM, TYPE 4, OCA4	
SLC4A1 (BAND 3 OF RED CELL MEMBRANE, ERYTHROID PROTEIN BAND 3, ANION EXCHANGE PROTEIN 1)	RENAL TUBULAR ACIDOSIS, DISTAL (AUTOSOMAL DOMINANT) » RENAL TUBULAR ACIDOSIS 1 RENAL TUBULAR ACIDOSIS, DISTAL (AUTOSOMAL RECESSIVE) » RENAL TUBULAR ACIDOSIS (AUTOSOMAL RECESSIVE) WITH PRESERVED HEARING » RENAL TUBULAR ACIDOSIS, DISTAL, (AUTOSOMAL RECESSIVE) WITH LATE-ONSET SENSORINEURAL HEARING LOSS HEMOLYTIC ANEMIA DUE TO BAND 3 MONTEFIORE	

	SPHEROCYTOSIS, HEREDITARY, DUE TO BAND 3 (TUSCALOOSA, PRAGUE, CHUR, NOIRTERRE, LYON, GENAS, FUKUOKA, TOKYO, COIMBRA, CAPE TOWN, PRAGUE III)	
	OVALOCYTOSIS (SOUTHEAST ASIAN , MALAYSIAN- MELANESIAN-FILIPINO TYPE)	
	ACANTHOCYTOSIS	
	ERYTHROCYTOSIS, STOMATOCYTIC HEREDITARY	
	ELLIPTOCYTOSIS (MALAYSIAN-MELANESIAN TYPE, ELLIPTOCYTOSIS 4)	
	WRIGHT BLOOD GROUP ANTIGEN	
	WALDNER BLOOD GROUP ANTIGEN WD(A)	
	DIEGO BLOOD GROUP ANTIGEN	
SLC4A4 (SODIUM BICARBONATE COTRANSPORTER 1, NBC1)	RENAL TUBULAR ACIDOSIS, PROXIMAL, WITH OCULAR ABNORMALITIES » RENAL TUBULAR ACIDOSIS 2	
SLC5A2	RENAL GLUCOSURIA	
SLC5A5 (SODIUM-IODIDE SYMPORTER, NIS)	THYROID HORMONOGENESIS	
SLC6A8 (CREATINE TRANSPORTER, CT1)	CREATINE DEFICIENCY SYNDROME (X-LINKED) MENTAL RETARDATION, WITH SEIZURES, SHORT STATURE AND MIDFACE HYPOPLASIA (X-LINKED)	Blood in RNA PAX tubes Blood in RNA PAX tubes
SLC7A9	CYSTINURIA TYPE 1	Whole Gene
SLC7A9 (analyzed together with SLC3A1)	CYSTINURIA TYPE 1	7 mutations in SLC7A9: Gly105Arg, Thr123Met, Phe140Ser, Ala182Thr, 747delG, 244delE, Arg333Trp includes 10 mutations in SLC3A1: Thr216Met, Ser217Arg, Arg365Trp, Arg365Leu, Arg362Cys, Met467Thr, Met467Lys, 1500+1G/T, Glu483X duplication of exons 5-9
SMAD4 (DPC4)	JUVENILE POLYPOSIS/HEREDITARY HEMORRHAGIC TELANGIECTASIA SYNDROME » POLYPOSIS, GENERALIZED JUVENILE, WITH PULMONARY ARTERIOVENOUS MALFORMATION	

SMARCA1 (SWI/SNF-RELATED, MATRIX-ASSOCIATED, ACTIN-DEPENDENT REGULATOR OF CHROMATIN, SUBFAMILY A-LIKE PROTEIN 1)	JUVENILE POLYPOSIS SYNDROME IMMUNOOSSEOUS DYSPLASIA, SCHIMKE TYPE	
SMC1A (STRUCTURAL MAINTENANCE OF CHROMOSOMES 1A, SMC1L1, SMC1)	CORNELIA DE LANGE SYNDROME (X-LINKED)	
SMCX (JARID1C)	MENTAL RETARDATION, SYNDROMIC (X –LINKED)	See also Mental Retardation Panel
SMN1 (SURVIVAL MOTOR NEURON PROTEIN)	SPINAL MUSCULAR ATROPHY 1, SMA1 SPINAL MUSCULAR ATROPHY 2, SMA2 SPINAL MUSCULAR ATROPHY 3, SMA3	
SMPD1 (SPHINGOMYELIN PHOSPHODIESTERASE 1, ACID SPHINGOMYELINASE)	NIEMANN-PICK DISEASE, TYPE A » SPHINGOMYELIN LIPIDOSIS » SPHINGOMYELINASE DEFICIENCY » (See also Molecular Screening Tests) NIEMANN-PICK DISEASE, TYPE B » (See also Molecular Screening Tests)	Whole Gene, 1 Mutation: DelR608 or 3 Mutations: R496L, FSP330, L302P
SNCA (ALPHA SYNUCLEIN)	PARKINSON DISEASE, TYPE 1, PARK1 (AUTOSOMAL DOMINANT) » LEWY BODY PARKINSONISM PARKINSON DISEASE, TYPE 4, PARK4 (AUTOSOMAL DOMINANT) LEWY BODY DEMENTIA	Whole Gene, 1 Mutation: DelR608 or 3 Mutations: R496L, FSP330, L302P Whole Gene or Deletions
SOD1	AMYOTROPHIC LATERAL SCLEROSIS, ALS » LOU GEHRIG'S DISEASE	Whole Gene or Deletions
SOS1 (SON OF SEVENLESS, DROSOPHILA, HOMOLOG 1)	NOONAN SYNDROME, TYPE 4	
SOX2 (SRY-BOX 2)	FIBROMATOSIS, GINGIVAL, TYPE 1	
SOX3 (SRY-BOX 3)	ANOPHTHALMIA, ANOP3	
SOX9 (SRY-BOX 9)	MENTAL RETARDATION (X-LINKED), WITH ISOLATED GROWTH HORMONE DEFICIENCY, MRGH CAMPOMELIC DYSPLASIA » ACAMPOMELIC CAMPOMELIC DYSPLASIA	

SPG3A (ATLASTIN)	FAMILIAL SPASTIC PARAPLEGIA 3 (AUTOSOMAL DOMINANT), SPG3A, FSP1 » STRUMPELL DISEASE	
SPG4 (SPASTIN, SPAST)	FAMILIAL SPASTIC PARAPLEGIA 4 (AUTOSOMAL DOMINANT), SPG4, FSP2	
SPG7 (PARAPLEGIN)	SPASTIC PARAPLEGIA 7 (AUTOSOMAL RECESSIVE), SPG7, FSP7	Whole Gene
SPINK1 (PANCREATIC SECRETORY TRYPSIN INHIBITOR, PSTI, TUMOR-ASSOCIATED TRYPSIN INHIBITOR, TATI)	PANCREATITIS, HEREDITARY, PCTT	Exon 3 Includes PRSS1 (Exons 1-3)
SPINK5 (LEKTI)	NETHERTON SYNDROME	
SPTLC1 (SERINE PALMITOYLTRANSFERASE, LONG-CHAIN BASE SUBUNIT 1)	NEUROPATHY, HEREDITARY SENSORY AND AUTONOMIC, TYPE 1 » HSN1 » NEUROPATHY, HEREDITARY SENSORY RADICULAR (AUTOSOMAL DOMINANT)	Exons 5-6
SRD5A2 (STEROID 5-ALPHA-REDUCTASE 2)	PSEUDO-VAGINAL PERINEOSCROTAL HYPOSPADIAS, PPSH » MALE PSEUDOHERMAPHRODITISM DUE TO 5-ALPHA-REDUCTASE DEFICIENCY » FAMILIAL INCOMPLETE MALE PSEUDOHERMAPHRODITISM, TYPE 2	
SRY (SEX-DETERMINING REGION Y, TESTIS-DETERMINING FACTOR, TDF)	GONADAL DYSGENESIS, XY FEMALE TYPE, GDXY » SWYER SYNDROME » SEX-REVERSAL	Whole Gene or Positive / Negative
STK11 (SERINE THREONINE KINASE)	PEUTZ-JEGHERS SYNDROME, PJS » HAMARTOUS INTESTINAL POLYPOSI	Whole Gene Sequencing or Deletions - Duplications
STX11 (SYNTAXIN 11)	HEMOPHAGOCYTIC LYMPHOHISTIOCYTOSIS, FAMILIAL, TYPE 4, FHL4	See also PRF1, STX11, UNC13D
SUMF1 (SULFATASE-MODIFYING FACTOR 1)	MULTIPLE SULFATASE DEFICIENCY » MUCOSULFATIDOSIS » SULFATIDOSIS, JUVENILE, AUSTIN TYPE	
SURF1 (SURFEIT 1)	CYTOCHROME c OXIDASE DEFICIENCY » COX DEFICIENCY » COMPLEX 4, MITOCHONDRIAL RESPIRATORY CHAIN, DEFICIENCY OF	

LEIGH SYNDROME

TAZ (TAFAZZIN)	BARTH SYNDROME	
TBCE (TUBULIN-SPECIFIC CHAPERONE E)	HYPOPARATHYROIDISM-RETARDATION-DYSMORPHISM SYNDROME » HYPOPARATHYROIDISM WITH SHORT STATURE, MENTAL RETARDATION, AND SEIZURES » SANJAD-SAKATI SYNDROME » HYPOPARATHYROIDISM, CONGENITAL, ASSOCIATED WITH DYSMORPHISM, GROWTH RETARDATION, AND DEVELOPMENTAL DELAY	Whole Gene or 12 bp Deletion in Exon 2
	KENNY-CAFFEY SYNDROME, TYPE 1, KCS1 (AUTOSOMAL RECESSIVE)	Whole Gene or 12 bp Deletion in Exon 2
TBP (TATA BOX-BINDING PROTEIN, SCA17)	SPINOCEREBELLAR ATAXIA 17, SCA17 » HUNTINGTON DISEASE-LIKE 4, HDL4	Repeat
TBX1 (T-BOX 1)	DIGEORGE SYNDROME, DGS » CATCH22 » 22q11.2 DELETION SYNDROME VELOCARDIOFACIAL SYNDROME, VCFS » SHPRINTZEN SYNDROME	
	CONOTRUNCAL ANOMALY FACE SYNDROME » DOUBLE-OUTLET RIGHT VENTRICLE » TRUNCUS ARTERIOSUS COMMUNIS	
TBX3 (T-BOX 3)	ULNAR - MAMMARY SYNDROME » SCHINZEL SYNDROME	
TBX5 (T-BOX 5)	HOLT-ORAM SYNDROME, HOS1 » HEART-HAND SYNDROME	Whole Gene or Deletions
TBX19 (T-BOX 19, T-BOX FACTOR, PITUITARY)	ACTH DEFICIENCY	
TCIRG1 (T CELL IMMUNE REGULATOR 1, TIRC7, IOC116)	OSTEOPETROSIS (AUTOSOMAL RECESSIVE) » MARBLE BONES (AUTOSOMAL RECESSIVE) » ALBERS-SCHONBERG DISEASE (AUTOSOMAL RECESSIVE)	
TCOF1 (TREACLE)	TREACHER COLLINS-FRANCESCHETTI SYNDROME (TCOF) » TREACHER COLLINS SYNDROME » MANDIBULOFACIAL DYSOSTOSIS	

TERC (TELOMERASE RNA COMPONENT)	DYSKERATOSIS CONGENITA (AUTOSOMAL DOMINANT) » DYSKERATOSIS CONGENITA, SCOGGINS TYPE APLASTIC ANEMIA	
TGFB1 (TGFB, TRANSFORMING GROWTH FACTOR, BETA-1)	CAMURATI-ENGELMANN DISEASE » DIAPHYSEAL DYSPLASIA 1	Whole Gene or Exon 24
TGFBR1 (TRANSFORMING GROWTH FACTOR-BETA RECEPTOR, TYPE 1, ALK5)	LOEYS-DIETZ SYNDROME	See also TGFBR1 and TGFBR2
TGFBR2 (TRANSFORMING GROWTH FACTOR-BETA RECEPTOR, TYPE 2)	MARFAN SYNDROME, TYPE 2 » MARFAN-LIKE CONNECTIVE TISSUE DISORDER	See also TGFBR1 and TGFBR2
	LOEYS-DIETZ SYNDROME	See also TGFBR1 and TGFBR2
TGFBR1 and TGFBR2	MARFAN SYNDROME, TYPE 2 » MARFAN-LIKE CONNECTIVE TISSUE DISORDER LOEYS-DIETZ SYNDROME	
TGIF	HOLOPROSENCEPHALY 4, HPE4	See SHH, SIX3, TGIF and ZIC2
TGM1 (TRANSGLUTAMINASE)	COLLODION FETUS » LAMELLAR ICHTHYOSIS ICHTHYOSIFORM ERYTHRODERMA, NONBULLOUS CONGENITAL	
THRB (THYROID HORMONE RECEPTOR, BETA, ERBA2)	THYROID HORMONE RESISTANCE, GENERALIZED (AUTOSOMAL DOMINANT) » HYPERTHYROXINEMIA, FAMILIAL EUTHYROID, SECONDARY TO PITUITARY AND PERIPHERAL RESISTANCE TO THYROID HORMONES THYROID HORMONE RESISTANCE, GENERALIZED (AUTOSOMAL RECESSIVE) » THYROID HORMONE UNRESPONSIVENESS » REFETOFF SYNDROME THYROID HORMONE RESISTANCE, SELECTIVE PITUITARY » HYPERTHYROIDISM, FAMILIAL, DUE TO INAPPROPRIATE THYROTROPIN SECRETION	

TIMP3 (TISSUE INHIBITOR OF METALLOPROTEINASE 3)	FUNDUS DYSTROPHY, PSEUDOINFLAMMATORY, OF SORSBY » SORSBY SYNDROME	
TITF1 (THYROID NUCLEAR FACTOR, NKX2A)	CHOREA, HEREDITARY BENIGN	
TM4SF2 (TRANSMEMBRANE 4 SUPERFAMILY, MEMBER 2)	MENTAL RETARDATION, NONSPECIFIC (X-LINKED), TYPE 58, MRX58	See also Mental Retardation Panel
TNFRSF1A (TNFR1)	HIBERNIAN FEVER, FAMILIAL » FAMILIAL PERIODIC FEVER (AUTOSOMAL DOMINANT) » TUMOR NECROSIS FACTOR RECEPTOR-ASSOCIATED PERIODIC SYNDROME, TRAPS	Exons 2-4 (> 95% of mutations)
TNFRSF6 (TUMOR NECROSIS FACTOR RECEPTOR SUPERFAMILY, MEMBER 6, APT1, FAS)	AUTOIMMUNE LYMPHOPROLIFERATIVE SYNDROME, TYPE 1, ALPS, ALPS1A, ALPS1B » CANALE-SMITH SYNDROME	Exons 7-9 (Majority of Mutations) or Exons 1-6 or Whole Gene
TNFRSF13 (TUMOR NECROSIS FACTOR RECEPTOR SUPERFAMILY, MEMBER 13B, TRANSMEMBRANE ACTIVATOR AND CAML INTERACTOR, TAC1)	COMMON VARIABLE IMMUNODEFICIENCY » COMMON VARIABLE HYPOGAMMAGLOBULINEMIA » HYPOGAMMAGLOBULINEMIA, ACQUIRED » IMMUNOGLOBULIN DEFICIENCY, LATE-ONSET	
TNFSF5 (TUMOR NECROSIS FACTOR LIGAND SUPERFAMILY, MEMBER 5, CD40 LIGAND, TRAP, GP39)	IMMUNODEFICIENCY WITH HYPER-IGM, TYPE 1, HIGM1 » HYPER-IGM IMMUNODEFICIENCY (X-LINKED) » HYPER-IGM SYNDROME 1	
TNNI2 (TROPONIN 1, FAST-TWITCH SKELETAL MUSCLE ISOFORM)	ARTHROGRYPOSIS, DISTAL, TYPE 2B » ARTHROGRYPOSIS MULTIPLEX CONGENITA, DISTAL, TYPE 2B » SHELDON-HALL SYNDROME » FREEMAN-SHELDON SYNDROME VARIANT » ARTHROGRYPOSIS MULTIPLEX CONGENITA, DISTAL, TYPE 2, WITH CRANIOFACIAL ABNORMALITIES	
TNNI3 (TROPONIN I, CARDIAC)	HYPERTROPHIC CARDIOMYOPATHY, FAMILIAL, 7, CMH7 » VENTRICULAR HYPERTROPHY, HEREDITARY » ASYMMETRIC SEPTAL HYPERTROPHY » HYPERTROPHIC SUBAORTIC STENOSIS, IDIOPATHIC CARDIOMYOPATHY, FAMILIAL RESTRICTIVE, 1	See also MYH7 MYBPC3, TNNT2, TNNI3 and TPM1 See also MYH7 MYBPC3, TNNT2, TNNI3

TNNT2 (TROPONIN T2, CARDIAC)	<p>HYPERTROPHIC CARDIOMYOPATHY, FAMILIAL, 2, CMH2 » VENTRICULAR HYPERTROPHY, HEREDITARY » ASYMMETRIC SEPTAL HYPERTROPHY » HYPERTROPHIC SUBAORTIC STENOSIS, IDIOPATHIC DILATED CARDIOMYOPATHY, 1D, CMD1D</p>	<p>and TPM1 See also MYH7 MYBPC3, TNNT2, TNNI3 and TPM1</p>
TNNT3 (TROPONIN T3, FAST SKELETAL)	<p>ARTHROGRYPOSIS, DISTAL, TYPE 2B » ARTHROGRYPOSIS MULTIPLEX CONGENITA, DISTAL, TYPE 2B » SHELDON-HALL SYNDROME » FREEMAN-SHELDON SYNDROME VARIANT » ARTHROGRYPOSIS MULTIPLEX CONGENITA, DISTAL, TYPE 2, WITH CRANIOFACIAL ABNORMALITIES</p>	<p>See also MYH7 MYBPC3, TNNT2, TNNI3 and TPM1</p>
TPM1 (TROPOMYOSIN 1)	<p>HYPERTROPHIC CARDIOMYOPATHY, FAMILIAL, 3, CMH3 » VENTRICULAR HYPERTROPHY, HEREDITARY » ASYMMETRIC SEPTAL HYPERTROPHY » HYPERTROPHIC SUBAORTIC STENOSIS, IDIOPATHIC</p>	<p>See also MYH7 MYBPC3, TNNT2, TNNI3 and TPM1</p>
TPM2 (TROPOMYOSIN 2, beta TROPOMYOSIN)	<p>ARTHROGRYPOSIS, DISTAL, TYPE 1 » ARTHROGRYPOSIS MULTIPLEX CONGENITA, DISTAL, TYPE 1 NEMALINE MYOPATHY TYPE 4 » NEMALINE MYOPATHY CAUSED BY MUTATION IN THE TROPOMYOSIN 2 GENE</p>	
TREM2	<p>PRESENILE DEMENTIA WITH BONE CYSTS » POLYCYSTIC LIPOMEMBRANOUS OSTEODYSPLASIA WITH SCLEROSING LEUKOENCEPHALOPATHY » DEMENTIA, PROGRESSIVE, WITH LIPOMEMBRANOUS POLYCYSTIC OSTEODYSPLASIA » BRAIN-BONE-FAT DISEASE » DEMENTIA, PREFRONTAL, WITH BONE CYSTS » NASU-HAKOLA SYNDROME</p>	

TREX1 (3-PRIME @REPAIR EXONUCLEASE 1, ATRIP)	AICARDI-GOUTIERES SYNDROME 1 » ENCEPHALOPATHY, FAMILIAL INFANTILE, WITH INTRACRANIAL CALCIFICATION AND CHRONIC CEREBROSPINAL FLUID LYMPHOCYTOSIS » CREE ENCEPHALITIS » PSEUDO-TORCH SYNDROME » PSEUDOTOXOPLASMOSIS SYNDROME	
TRIM37 (TRIPARTITE MOTIF-CONTAINING PROTEIN 37)	MULIBREY NANISM » MUSCLE-LIVER-BRAIN-EYE NANISM » PERHEENTUPA SYNDROME	
TRPC6 (TRANSIENT RECEPTOR POTENTIAL CATION CHANNEL, SUBFAMILY C, MEMBER 6)	FOCAL SEGMENTAL GLOMERULOSCLEROSIS, TYPE 2	
TRPS1 (ZINC FINGER TRANSCRIPTION FACTOR TRPS1)	TRICHORHINOPHALANGEAL SYNDROME, TYPE 1, TRPS1	
TSC1 (HAMARTIN)	TUBEROUS SCLEROSIS	Whole Gene Sequencing - At least 20mg DNA is needed See also TSC1 and TSC2
TSC1 and TSC2	TUBEROUS SCLEROSIS	2 Whole Genes and TSC2 Deletion Testing - At least 20mg DNA is needed
TSC2 (TUBERIN)	TUBEROUS SCLEROSIS	Whole Gene Sequencing - At least 20mg DNA is needed See also TSC1 and TSC2
TSHR (THYROID-STIMULATING HORMONE RECEPTOR)	GRAVES DISEASE THYROID ADENOMA, HYPERFUNCTIONING THYROTROPIN, UNRESPONSIVENESS TO HYPERTHYROIDISM HYPOTHYROIDISM THYROID CARCINOMA	
TSPY (TESTIS-SPECIFIC PROTEIN, Y-LINKED)	CRYPTIC Y-CHROMOSOMAL MATERIAL	
TTID (TITIN IMMUNOGLOBULIN DOMAIN PROTEIN)	MUSCULAR DYSTROPHY, LIMB-GIRDLE, TYPE 1A, LGMD1A	Whole Gene or Exon 2
	MYOTILINOPATHY » MYOPATHY, MYOFIBRILLAR, MYOTILIN-RELATED	Whole Gene or Exon 2

TTN (TITIN, CONNECTIN)	CARDIOMYOPATHY, FAMILIAL HYPERTROPHIC, 9 CARDIOMYOPATHY, DILATED, 1G, CMD1G TIBIAL MUSCULAR DYSTROPHY, TARDIVE » UDD MYOPATHY	Exons 212-213 Exons 212-213 Exons 212-213
TTPA (TOCOPHEROL TRANSFER PROTEIN, ALPHA, TTP1)	VITAMIN E, FAMILIAL ISOLATED DEFICIENCY OF » ATAXIA, FRIEDREICH-LIKE, WITH SELECTIVE VITAMIN E DEFICIENCY » FRIEDREICH-LIKE ATAXIA ATAXIA AND RETINITIS PIGMENTOSA WITH ISOLATED VITAMIN E DEFICIENCY	
TTR (TRANSTHYRETIN) TWIST	AMYLOID POLYNEUROPATHY » AMYLOIDOSIS 1 SAETHRE-CHOTZEN SYNDROME » CRANIOSYNOSTOSIS, SAETHRE-CHOTZEN SYNDROME ROBINOW-SORAUFG SYNDROME	Exons 2-4
UBE3A	ANGELMAN SYNDROME, AS	Whole Gene or IC Deletion or Methylation
UBR1 (UBIQUITIN-PROTEIN LIGASE E3 COMPONENT N-RECOGNIN 1)	JOHANSON-BLIZZARD SYNDROME » NASAL ALAR HYPOPLASIA, HYPOTHYROIDISM, PANCREATIC ACHYLIA, AND CONGENITAL DEAFNESS	
UGT1A1 (UDP-GLYCURONOSYL TRANSFERASE)	CRIGLER-NAJJAR SYNDROME, TYPE 1 CRIGLER-NAJJAR SYNDROME, TYPE 2 GILBERT SYNDROME	1 Mutation: TA insertion in promotor
UMOD (UROMODULIN)	MEDULLARY CYSTIC KIDNEY DISEASE 2, MCKD2 FAMILIAL JUVENILE HYPERURICEMIC NEPHROPATHY, HNFJ » GOUTY NEPHROPATHY	
UNC13D (UNC13, C. ELEGANS, HOMOLOG OF, D, MUNC13-4)	HEMOPHAGOCYTTIC LYMPHOHISTIOCYTOSIS, FAMILIAL, TYPE 3, FHL3	See also PRF1, STX11, UNC13D
UNG (URACIL-DNA GLYCOSYLASE)	IMMUNODEFICIENCY WITH HYPER-IgM, TYPE 5 » HYPER-IgM SYNDROME 5	
USH2A	USHER SYNDROME, TYPE 2A, USH2A RETINITIS PIGMENTOSA, (AUTOSOMAL RECESSIVE) WITHOUT HEARING LOSS	Exon 13 Exon 13

VDR (VITAMIN D HORMONE RECEPTOR)	VITAMIN D-DEPENDENT RICKETS, TYPE 2A » RICKETS-ALOPECIA SYNDROME » HYPOCALCEMIC VITAMIN D-RESISTANT RICKETS » RICKETS, VITAMIN D-DEPENDENT RICKETS, TYPE 2A	
VHL	VON HIPPEL-LINDAU SYNDROME, VHL	Whole Gene or Deletion Analysis (MLPA)
	RENAL CELL CARCINOMA 1	Whole Gene or Deletion Analysis (MLPA)
	PHEOCHROMOCYTOMA	Whole Gene or Deletion Analysis (MLPA)
	CHUVASH POLYCYTHEMIA	Whole Gene or Deletion Analysis (MLPA)
	CEREBELLAR HEMANGIOMA	Whole Gene or Deletion Analysis (MLPA)
VKORC1	VITAMIN K-DEPENDENT CLOTTING FACTORS, COMBINED DEFICIENCY OF, 2	
VMD2 (VITELLIFORM MACULAR DYSTROPHY GENE 2, BESTROPHIN)	MACULAR DYSTROPHY, VITELLIFORM, VMD » BEST MACULAR DYSTROPHY MACULAR DYSTROPHY, CONCENTRIC ANNULAR » BULL'S EYE MACULAR DYSTROPHY VITELLIFORM MACULAR DYSTROPHY, ADULT-ONSET » FOVEOMACULAR DYSTROPHY, ADULT-ONSET, WITH CHOROIDAL NEOVASCULARIZATION	
VSX1 (VISUAL SYSTEM HOMEBOX GENE 1, ZEBRAFISH, HOMOLOG OF)	KERATOCONUS 1, KTCN1 CORNEAL DYSTROPHY, POSTERIOR POLYMORPHOUS, 1 PPCD1 CRANIOFACIAL ANOMALIES, EMPTY SELLA TURCICA, CORNEAL ENDOTHELIAL CHANGES, AND ABNORMAL RETINAL AND AUDITORY BIPOLAR CELLS	
VWF (VON WILLEBRAND FACTOR)	VON WILLEBRAND DISEASE, TYPE NORMANDY	Whole Gene or Exons 18-20, Including Mutations T791M, R816W and R854Q
WAS (WASP)	WISKOTT-ALDRICH SYNDROME, WAS THROMBOCYTOPENIA (X-LINKED) NEUTROCYTOPENIA (X-LINKED)	

WDR36 (WD40-REPEAT 36)	GLAUCOMA, PRIMARY OPEN ANGLE, ADULT-ONSET, POAG	
WFS1 (WOLFRAMIN)	WOLFRAM SYNDROME » DIABETES INSIPIDUS AND MELLITUS WITH OPTIC ATROPHY AND DEAFNESS, DIDMOAD DEAFNESS, DFNA6 » DEAFNESS, (AUTOSOMAL DOMINANT), NONSYNDROMIC SENSORINEURAL 6 ARTHROPATHY, PROGRESSIVE	
WISP3 (WNT1-INDUCIBLE SIGNALING PATHWAY PROTEIN 3)	PSEUDORHEUMATOID, OF CHILDHOOD » SPONDYLOEPIPHYSEAL DYSPLASIA TARDA WITH PROGRESSIVE ARTHROPATHY » PROGRESSIVE PSEUDORHEUMATOID DYSPLASIA	
WNK1 (PROTEIN KINASE, LYSINE-DEFICIENT 1)	PSEUDOHYPOALDOSTERONISM, TYPE 2 » HYPERPOTASSEMIA AND HYPERTENSION, FAMILIAL » HYPERTENSIVE HYPERKALEMIA, FAMILIAL » GORDON HYPERKALEMIA-HYPERTENSION SYNDROME	
WNK4 (PROTEIN KINASE, LYSINE-DEFICIENT 4)	PSEUDOHYPOALDOSTERONISM, TYPE 2 » HYPERPOTASSEMIA AND HYPERTENSION, FAMILIAL » HYPERTENSIVE HYPERKALEMIA, FAMILIAL » GORDON HYPERKALEMIA-HYPERTENSION SYNDROME	
WNT3 (WINGLESS-TYPE MMTV INTEGRATION SITE FAMILY, MEMBER 3)	TETRA-AMELIA (AUTOSOMAL RECESSIVE)	
WNT7A (WINGLESS-TYPE MMTV INTEGRATION SITE FAMILY, MEMBER 7A)	FIBULAR APLASIA OR HYPOPLASIA, FEMORAL BOWING AND POLY-, SYN-, AND OLIGODACTYLY » FUHRMANN SYNDROME ULNA AND FIBULA, ABSENCE OF, WITH SEVERE LIMB DEFICIENCY » LIMB/PELVIS-HYPOPLASIA/APLASIA SYNDROME » AL-AWADI/RAAS-ROTHSCHILD SYNDROME » SCHINZEL PHOCOMELIA SYNDROME	
WT1 (WILMS TUMOR 1 GENE)	WILMS TUMOR 1 » NEPHROBLASTOMA DENYS-DRASH SYNDROME » WILMS TUMOR AND	Whole Gene Whole Gene

	PSEUDOHERMAPHRODITISM	
	FRASIER SYNDROME	Whole Gene
	NEPHROTIC SYNDROME, EARLY-ONSET, WITH DIFFUSE MESANGIAL SCLEROSIS » MESANGIAL SCLEROSIS, FAMILIAL	Whole Gene or Deletion Analysis (MLPA)
XLRS1 (RS1, RETINOSCHISIN) Y-DELETIONS	RETINOSCHISIS 1 (X-LINKED), JUVENILE, RS1 See AZFa, AZFb and AZFc	
ZAP70 (ZETA-CHAIN-ASSOCIATED PROTEIN KINASE, SYK-RELATED TYROSINE KINASE)	SELECTIVE T-CELL DEFECT, IMMUNODEFICIENCY DUE TO	
ZFX1B (ZINC FINGER HOMEBOX 1B; ZFH1B, SMAD-INTERACTING PROTEIN 1, SMADIP1, SIP1)	MOWAT-WILSON SYNDROME » MICROCEPHALY, MENTAL RETARDATION, AND DISTINCT FACIAL FEATURES, WITH OR WITHOUT HIRSCHSPRUNG DISEASE » HIRSCHSPRUNG DISEASE-MENTAL RETARDATION SYNDROME	Whole Gene or MLPA
	HIRSCHSPRUNG DISEASE-MENTAL RETARDATION SYNDROME, LATE INFANTILE	Whole Gene or MLPA
ZIC2 (ZINC FINGER PROTEIN OF CEREBELLUM, 2)	HOLOPROSENCEPHALY 5, HPES	See also SHH, SIX3, TGIF and ZIC2
ZIC3	HETEROTAXY, VISCERAL (X-LINKED) » SITUS INVERSUS (X-LINKED) TRANSPOSITION OF GREAT ARTERIES (X-LINKED) CONGENITAL HEART DISEASE (X-LINKED)	
ZMPSTE24 (ZINC METALLOPROTEINASE STE24)	MANDIBULOACRAL DYSPLASIA WITH TYPE B LIPODYSTROPHY » LIPODYSTROPHY, TYPE B, ASSOCIATED WITH MANDIBULOACRAL DYSPLASIA TIGHT SKIN CONTRACTURE SYNDROME, LETHAL » HYPERKERATOSIS-CONTRACTURE SYNDROME » RESTRICTIVE DERMOPATHY, LETHAL » FETAL HYPOKINESIA SEQUENCE DUE TO RESTRICTIVE DERMOPATHY	
ZNF41 (ZINC FINGER PROTEIN 41)	MENTAL RETARDATION, NONSPECIFIC (X-LINKED)	See also Mental Retardation Panel

ZNF9

MYOTONIC DYSTROPHY, TYPE 2 » PROXIMAL
MYOTONIC MYOPATHY, PROMM » MYOTONIC
MYOPATHY, PROXIMAL » RICKER SYNDROME

Repeat Expansion (CCTG) At least 20
microgram DNA with a concentration
higher than 300 nanogram per
microliter is needed

GENÉTICA MOLECULAR DE COLOMBIA

Calle 103 A No. 21-49, Chicó Navarra

Teléfono(Fax) 2361483 y 6217306, Celular 3158967774

E-mail: info@geneticamolecular.net. Página web: www.geneticamolecular.net

BOGOTA, DC. Colombia