UK Genetic Testing Network

# NHS Directory of Genetic Testing

## A list of diseases for which tests are offered by UK Genetic Testing Network Laboratories

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## NHS Directory of Genetic Testing available from UKGTN laboratories

### Introduction

The UK Genetic Testing Network (http: www.ukgtn.nhs.uk) aims to support genetic tests for patients and their families through promoting high quality, equitable, and appropriately validated laboratory tests in the diagnosis and management of inherited genetic disorders. It provides co-ordination of the evaluation and provision of these laboratory services, and supports their commissioning and prioritisation, an approach that is strongly supported by the Department of Health.

### Assessing the clinical validity and utility of proposals for new tests to be offered as part of the UKGTN

A core function of the UKGTN is the clinical and scientific evaluation of proposed new genetic tests for adoption as mainstream NHS services. In the context of the UKGTN a genetic test is defined, for this Directory listing, as a test for germ line disorders where nucleic acid is the analyte (i.e. testing for a specific genetic disease, through testing for a causative mutation in DNA, usually present from conception). A tool was developed by the UKGTN Steering Group to evaluate these tests, "Proposal form for the evaluation of a genetic test for NHS service: Gene Dossier" (which is available from the UKGTN website at: http://www.ukgtn.nhs.uk/gtn/Information/Services/UKGTN_Forms ).

A genetic test is described as a test for a specified population, for a particular disease and defined genetic variants and for a specific purpose. The UKGTN evaluation of a test considers each of these elements. The analytical sensitivity and specificity are determined by the technologies and methodologies used in the laboratory. The gene dossier evaluation process also considers how the technologies and methodologies have been validated in the providing laboratory. The clinical sensitivity and specificity is calculated for the target population where the target population is described by clinical features or family history of clinical features. The utility of the test describes how the management of the patient will be affected by testing. Tests that are recommended by the UKGTN for NHS service are recommended on the basis of the information provided in the gene dossier.

Gene Dossiers are submitted to the UKGTN Gene Dossier working group for initial evaluation against the agreed criteria. The working group makes recommendations to the UKGTN Steering Group. Tests that are accepted by the Steering Group are taken to the autumn meeting of the Department of Health Genetics Commissioning Advisory Group (GenCAG) for the tests to be considered for inclusion in the Directory and for recommendation to commissioners for funding.

The UKGTN will also evaluate Gene Dossiers for tests that are being proposed to be included in a National Commissioning Group - NCG (formerly the National Specialist Commissioning Advisory Group) service and provided by UKGTN laboratories. Services agreed for funding through NCG are highlighted in green in the Directory.

## Commissioning the services listed on the NHS Directory of Molecular Genetic Testing

The UKGTN and GenCAG recommend to all UK commissioners and provider organisations that genetic testing for the diseases listed on the Directory should be available to their local populations. This testing is considered to be clinically appropriate when referred by a clinical professional within agreed protocols.

The recommendation to fund and provide new tests that have passed the gene dossier process is for the relevant health authorities in the United Kingdom. In England services accepted by the Gene Dossier process and which require additional NHS funding will be recommended by UKGTN to the Genetics Commissioning Advisory Group for consideration of funding in the commissioning plans between Specialised Commissioning Groups and Primary Care Trusts. Scotland, Northern Ireland and Wales have different commissioning arrangements and therefore the recommendation for new tests is made to the National Services Division Scotland, the Regional Medical Services Consortium and the Health Commission Wales, respectively. It is recognised by the UKGTN that as NHS commissioning remains within local arrangements that inclusion in the Directory does not guarantee funding.

There are a number of other tests that tend to be of low volume and are provided by non-UKGTN specialist laboratories. These laboratories may be NHS specialist laboratories or overseas laboratories. The tests that are provided by these non-UKGTN specialist laboratories have not been evaluated by the Gene Dossier process and therefore are not included in the Directory. Information about these tests can be obtained by contacting one of the Regional Genetics Centres.

## Clinical Referral

The ordering of a genetics test is based on the same process of clinical judgement as is exercised in any aspect of patient care, particularly as to whether the result of the test will significantly affect management. Therefore, each new test listed from July 2005 is accompanied by details of associated testing criteria. The UKGTN has developed the concept of Testing Criteria as part of the Gene Dossier application process. Testing Criteria in essence defines the appropriateness of a genetic test referral, and can inform clinicians' decisions about which investigations are suitable for their patients. Further information about testing criteria is available on the UKGTN website at http://www.ukgtn.nhs.uk/gtn/Information/Services/Testing_Criteria . Each of the tests offered may have different clinical uses even in individual families with the same disorder. Usually a mutation has to be detected in an affected family member before specific testing can be offered to the rest of the family. As well as using the listed testing criteria for a condition, further advice regarding the appropriateness of testing in individual cases can be sought from the Regional Genetic Centres.

## Monitoring and development of the Directory

The Directory is a regularly updated document with each version identifying those diseases and tests recommended as NHS services. The UKGTN will continue to work with laboratories and clinicians to ensure that the range of tests offered within the Directory continue to meet the criteria for validity and utility.

## Structure of the UKGTN Directory of Genetic Testing – Version 5

The directory consists of two columns. The first column lists the name of the disease and where available the symbol and a six-digit number. The name, symbol and the number have been taken from the OMIM (**O**nline **M**endelian **I**nheritance in **M**an) database (http://www.ncbi.nlm.nih.gov/entrez/query.fcgi?db=OMIM). The second column contains the Human Gene Nomenclature Committee (http://www.gene.ucl.ac.uk/nomenclature/) name of the gene and where available the OMIM number of the gene(s) for which Network laboratories provide tests for each disease. The OMIM numbers have been provided so that information about the clinical features of the disease and the characteristics of the gene and its mutations may be accessed through the OMIM database.

**Section one**
Tests that are currently available from UKGTN laboratories.
*Also listed on the UKGTN online database*

Included in Section 1 are a number of inherited metabolic diseases (IMD). In many clinical situations, biochemical (metabolite or enzyme) analysis or other types of test may be preferred rather than genetic analysis. However, in certain situations, genetic testing may be the best or only option for a family. Examples where this may be the case include a lack of appropriate material for biochemical tests or invalidation of the biochemical test, e.g. blood transfusion. The test is therefore available as a service in these circumstances. The clinical and laboratory experts or the IMD services are the appropriate gatekeepers for these referrals.

**Section two**
Testing that has recently been assessed through the UKGTN Gene Dossier process and has been accepted by GenCAG for NHS service from the start of the current year (2008/09). These tests may not be available initially as the laboratory could be in the process of starting up testing in order for it to be available as a full NHS service.
*Tests will only be listed on the UKGTN online database once the test is available for full NHS service.*

**Section three**
Approved tests that are currently not available from any UKGTN laboratories but may be available from laboratories out with the UKGTN. This section is included as these tests should be considered in local funding discussions between UKGTN laboratories, NHS Trusts and commissioners as they have been previously evaluated and accepted for inclusion in the Directory but are currently not offered by a UKGTN laboratory.
*Not listed on the UKGTN online database as the database only shows services from UKGTN laboratories.*

**Additional information**
The tests listed in this Directory have been recommended by the UKGTN Steering Group to GenCAG for NHS service and have subsequently been adopted by GenCAG. However there are tests that are provided by UKGTN laboratories as routine NHS provision but which are funded outside the normal NHS commissioning processes and these tests are listed separately on the UKGTN website. In addition further information on the progress of Gene Dossiers is available on the UKGTN website.

## Section 1

This section contains a list of approved diseases and the associated tests that are currently being offered by UKGTN laboratories.

| DISEASE | | GENE | |
|---|---|---|---|
| NAME | OMIM | NAME | OMIM |
| ACANTHOSIS NIGRICANS; AN | 100600 | FIBROBLAST GROWTH FACTOR RECEPTOR 3 (ACHONDROPLASIA, THANATOPHORIC DWARFISM); FGFR3 | 134934 |
| ACHONDROPLASIA; ACH | 100800 | FIBROBLAST GROWTH FACTOR RECEPTOR 3 (ACHONDROPLASIA, THANATOPHORIC DWARFISM); FGFR3 | 134934 |
| ACYL-CoA DEHYDROGENASE, LONG-CHAIN, DEFICIENCY OF | 201460 | HYDROXYACYL-COENZYME A DEHYDROGENASE/3-KETOACYL-COENZYME A THIOLASE/ENOYL-COENZYME A HYDRATASE (TRIFUNCTIONAL PROTEIN), ALPHA SUBUNIT; HADHA | 600890 |
| ACYL-CoA DEHYDROGENASE, MEDIUM-CHAIN, DEFICIENCY OF | 201450 | ACYL-COENZYME A DEHYDROGENASE, C-4 TO C-12 STRAIGHT CHAIN; ACADM | 607008 |
| ADENOMATOUS POLYPOSIS OF THE COLON; APC | 175100 | ADENOMATOSIS POLYPOSIS COLI; APC | 175100 |
| ADENOSINE MONOPHOSPHATE DEAMINASE 1; AMPD1 | 102770 | ADENOSINE MONOPHOSPHATE DEAMINASE 1 (ISOFORM M); AMPD1 | 102770 |
| ADRENAL HYPERPLASIA, CONGENITAL, DUE TO 21-HYDROXYLASE DEFICIENCY | 201910 | CYTOCHROME P450, FAMILY 21, SUBFAMILY A, POLYPEPTIDE 2; CYP21A2 | 201910 |
| ADRENAL HYPOPLASIA, CONGENITAL; AHC | 300200 | nuclear receptor subfamily 0, group B, member 1; NR0B1 | 300473 |
| ADRENOLEUKODYSTROPHY; ALD | 300100 | ATP-BINDING CASSETTE, SUB-FAMILY D (ALD), MEMBER 1; ABCD1 | 300371 |
| ALAGILLE SYNDROME; AGS | 118450 | JAGGED 1 (ALAGILLE SYNDROME); JAG1 | 601920 |
| ALBRIGHT HEREDITARY OSTEODYSTROPHY; AHO | 103580 | GNAS COMPLEX LOCUS; GNAS | 139320 |
| ALEXANDER DISEASE | 203450 | GLIAL FIBRILLARY ACIDIC PROTEIN; GFAP | 137780 |
| ALLAN-HERNDON-DUDLEY SYNDROME; AHDS | 300523 | solute carrier family 16, member 2 (monocarboxylic acid transporter 8); SLC16A2 | 300095 |

| DISEASE | | GENE | |
|---|---|---|---|
| NAME | OMIM | NAME | OMIM |
| ALPERS DIFFUSE DEGENERATION OF CEREBRAL GRAY MATTER WITH HEPATIC CIRRHOSIS | 203700 | POLYMERASE (DNA DIRECTED), GAMMA; POLG | 174763 |
| ALPORT SYNDROME, X-LINKED; ATS | 301050 | COLLAGEN, TYPE IV, ALPHA 5 (ALPORT SYNDROME); COL4A5 | 303630 |
| ALSTROM SYNDROME; ALMS | 203800 | ALSTROM SYNDROME 1; ALMS1 | 606844 |
| ALZHEIMER DISEASE; AD | 104300 | PRESENILIN 1 (ALZHEIMER DISEASE 3); PSEN1 | 104311 |
| AMYOTROPHIC LATERAL SCLEROSIS 1; ALS1 | 105400 | SUPEROXIDE DISMUTASE 1, SOLUBLE (AMYOTROPHIC LATERAL SCLEROSIS 1 (ADULT)); SOD1 | 147450 |
| ANDROGEN INSENSITIVITY SYNDROME; AIS | 300068 | ANDROGEN RECEPTOR (DIHYDROTESTOSTERONE RECEPTOR; TESTICULAR FEMINIZATION; SPINAL AND BULBAR MUSCULAR ATROPHY; KENNEDY DISEASE); AR | 313700 |
| ANGELMAN SYNDROME; AS | 105830 | NONIMPRINTED GENE IN PRADER-WILLI SYNDROME/ANGELMAN SYNDROME CHROMOSOME REGION 2 | 608146 |
| | | SMALL NUCLEAR RIBONUCLEOPROTEIN POLYPEPTIDE N; SNRPN | 182279 |
| | | UBIQUITIN PROTEIN LIGASE E3A (HUMAN PAPILLOMA VIRUS E6-ASSOCIATED PROTEIN, ANGELMAN SYNDROME); UBE3A | 601623 |
| ANIRIDIA; AN1 | 106200 | PAIRED BOX 6; PAX6 | 607108 |
| ANTLEY-BIXLER SYNDROME; ABS | 207410 | FIBROBLAST GROWTH FACTOR RECEPTOR 2 (BACTERIA-EXPRESSED KINASE, KERATINOCYTE GROWTH FACTOR RECEPTOR, CRANIOFACIAL DYSOSTOSIS 1, CROUZON SYNDROME, PFEIFFER SYNDROME, JACKSON-WEISS SYNDROME); FGFR2 | 176943 |
| | | P450 (CYTOCHROME) OXIDOREDUCTASE; POR | 124015 |

| DISEASE | | GENE | |
|---|---|---|---|
| NAME | OMIM | NAME | OMIM |
| APERT SYNDROME | 101200 | FIBROBLAST GROWTH FACTOR RECEPTOR 2 (BACTERIA-EXPRESSED KINASE, KERATINOCYTE GROWTH FACTOR RECEPTOR, CRANIOFACIAL DYSOSTOSIS 1, CROUZON SYNDROME, PFEIFFER SYNDROME, JACKSON-WEISS SYNDROME); FGFR2 | 176943 |
| APOLIPOPROTEIN E; APOE | 107741 | APOLIPOPROTEIN E; APOE | 107741 |
| AUTOIMMUNE LYMPHOPROLIFERATIVE SYNDROME; ALPS | 601859 | Fas (TNF receptor superfamily, member 6); FAS | 134637 |
| AUTOIMMUNE POLYENDOCRINOPATHY SYNDROME, TYPE I | 240300 | autoimmune regulator; AIRE | 607358 |
| AUTONOMIC CONTROL, CONGENITAL FAILURE OF | 209880 | PAIRED-LIKE HOMEOBOX 2B; PHOX2B | 603851 |
| BARTH SYNDROME; BTHS | 302060 | TAFAZZIN (CARDIOMYOPATHY, DILATED 3A (X-LINKED); ENDOCARDIAL FIBROELASTOSIS 2; BARTH SYNDROME); TAZ | 300394 |
| BASAL CELL NEVUS SYNDROME; BCNS | 109400 | patched homolog 1 (Drosophila); PTCH1 | 601309 |
| BASAL GANGLIA DISEASE, ADULT-ONSET | 606159 | FERRITIN, LIGHT POLYPEPTIDE; FTL | 134790 |
| BECKWITH-WIEDEMANN SYNDROME; BWS | 130650 | CYCLIN-DEPENDENT KINASE INHIBITOR 1C (P57, KIP2); CDKN1C | 600856 |
| BETA THALASSEMIA, DOMINANT INCLUSION BODY TYPE | 603902 | HEMOGLOBIN, BETA; HBB | 141900 |
| BRACHYDACTYLY, TYPE B1; BDB1 | 113000 | RECEPTOR TYROSINE KINASE-LIKE ORPHAN RECEPTOR 2; ROR2 | 602337 |
| BRACHYDACTYLY, TYPE D; BDD | 113200 | homeobox D13; HOXD13 | 142989 |
| BRACHYDACTYLY, TYPE E; BDE | 113300 | homeobox D13; HOXD13 | 142989 |
| BRANCHIOOTORENAL DYSPLASIA | 113650 | EYES ABSENT HOMOLOG 1 (DROSOPHILA); EYA1 | 601653 |
| BREAST CANCER | 114480 | BREAST CANCER 1, EARLY ONSET; BRCA1 | 113705 |
| | | BREAST CANCER 2, EARLY ONSET; BRCA2 | 600185 |

| DISEASE | | GENE | |
|---|---|---|---|
| NAME | OMIM | NAME | OMIM |
| BRUGADA SYNDROME | 601144 | sodium channel, voltage-gated, type V, alpha subunit; SCN5A | 600163 |
| BRUTON AGAMMAGLOBULINEMIA TYROSINE KINASE; BTK | 300300 | BRUTON AGAMMAGLOBULINEMIA TYROSINE KINASE; BTK | 300300 |
| BULLOUS ERYTHRODERMA ICHTHYOSIFORMIS CONGENITA OF BROCQ | 113800 | KERATIN 1 (EPIDERMOLYTIC HYPERKERATOSIS); KRT1 | 139350 |
| | | KERATIN 10 (EPIDERMOLYTIC HYPERKERATOSIS; KERATOSIS PALMARIS ET PLANTARIS); KRT10 | 148080 |
| CANAVAN DISEASE | 271900 | aspartoacylase (Canavan disease); ASPA | 608034 |
| CARBAMOYL PHOSPHATE SYNTHETASE I DEFICIENCY, HYPERAMMONEMIA DUE TO | 237300 | CARBAMOYL PHOSPHATE SYNTHETASE I DEFICIENCY, HYPERAMMONEMIA DUE TO | 237300 |
| CARDIOMYOPATHY, DILATED, 1A; CMD1A | 115200 | LAMIN A/C; LMNA | 150330 |
| | | MYOSIN BINDING PROTEIN C, CARDIAC; MYBPC3 | 600958 |
| | | myosin, heavy chain 7, cardiac muscle, beta; MYH7 | 160760 |
| CARDIOMYOPATHY, FAMILIAL HYPERTROPHIC, 2; CMH2 | 115195 | troponin T type 2 (cardiac); TNNT2 | 191045 |
| CARDIOMYOPATHY, FAMILIAL HYPERTROPHIC, 4; CMH4 | 115197 | MYOSIN BINDING PROTEIN C, CARDIAC; MYBPC3 | 600958 |
| CARDIOMYOPATHY, FAMILIAL HYPERTROPHIC; CMH | 192600 | mitochondrially encoded tRNA leucine 1 (UUA/G); MT-TL1 | 590050 |
| | | MYOSIN BINDING PROTEIN C, CARDIAC; MYBPC3 | 600958 |
| | | myosin, heavy chain 7, cardiac muscle, beta; MYH7 | 160760 |
| | | troponin I type 3 (cardiac); TNNI3 | 191044 |
| | | troponin T type 2 (cardiac); TNNT2 | 191045 |
| CARNITINE PALMITOYLTRANSFERASE II DEFICIENCY, LATE-ONSET | 255110 | CARNITINE PALMITOYLTRANSFERASE II; CPT2 | 600650 |
| CEREBRAL ARTERIOPATHY, AUTOSOMAL DOMINANT, WITH SUBCORTICAL INFARCTS AND LEUKOENCEPHALOPATHY; CADASIL | 125310 | NOTCH HOMOLOG 3 (DROSOPHILA); NOTCH3 | 600276 |

| DISEASE | | GENE | |
|---|---|---|---|
| NAME | OMIM | NAME | OMIM |
| CEROID LIPOFUSCINOSIS, NEURONAL 1, INFANTILE; CLN1 | 256730 | PALMITOYL-PROTEIN THIOESTERASE 1 (CEROID-LIPOFUSCINOSIS, NEURONAL 1, INFANTILE); PPT1 | 600722 |
| CEROID LIPOFUSCINOSIS, NEURONAL 2, LATE INFANTILE; CLN2 | 204500 | TRIPEPTIDYL PEPTIDASE I; TPP1 | 607998 |
| CEROID LIPOFUSCINOSIS, NEURONAL 3, JUVENILE; CLN3 | 204200 | CEROID-LIPOFUSCINOSIS, NEURONAL 3, JUVENILE (BATTEN, SPIELMEYER-VOGT DISEASE); CLN3 | 607042 |
| CEROID LIPOFUSCINOSIS, NEURONAL 6; CLN6 | 601708 | CEROID-LIPOFUSCINOSIS, NEURONAL 3, JUVENILE (BATTEN, SPIELMEYER-VOGT DISEASE); CLN3 | 607042 |
| CHARCOT-MARIE-TOOTH DISEASE, AXONAL, TYPE 2B1 | 605588 | LAMIN A/C; LMNA | 150330 |
| CHARCOT-MARIE-TOOTH DISEASE, DEMYELINATING, TYPE 1A; CMT1A | 118220 | PERIPHERAL MYELIN PROTEIN 22; PMP22 | 601097 |
| CHARCOT-MARIE-TOOTH DISEASE, DEMYELINATING, TYPE 1B; CMT1B | 118200 | MYELIN PROTEIN ZERO (CHARCOT-MARIE-TOOTH NEUROPATHY 1B); MPZ | 159440 |
| CHARCOT-MARIE-TOOTH DISEASE, X-LINKED, 1; CMTX1 | 302800 | gap junction protein, beta 1, 32kDa; GJB1 | 304040 |
| CHONDRODYSPLASIA PUNCTATA 1, X-LINKED RECESSIVE; CDPX1 | 302950 | EMOPAMIL BINDING PROTEIN (STEROL ISOMERASE); EBP | 300205 |
| | | PEROXISOMAL BIOGENESIS FACTOR 7; PEX7 | 601757 |
| CHOROIDEREMIA; CHM | 303100 | CHOROIDEREMIA (RAB ESCORT PROTEIN 1); CHM | 300390 |
| CLEIDOCRANIAL DYSPLASIA; CCD | 119600 | RUNT-RELATED TRANSCRIPTION FACTOR 2; RUNX2 | 600211 |
| COAGULATION FACTOR II; F2 | 176930 | COAGULATION FACTOR II (THROMBIN); F2 | 176930 |
| COFFIN-LOWRY SYNDROME; CLS | 303600 | RIBOSOMAL PROTEIN S6 KINASE, 90KDA, POLYPEPTIDE 3; RPS6KA3 | 300075 |
| COLORECTAL ADENOMATOUS POLYPOSIS, AUTOSOMAL RECESSIVE | 608456 | MUTY HOMOLOG (E. COLI); MUTYH | 604933 |

| DISEASE | | GENE | |
|---|---|---|---|
| NAME | OMIM | NAME | OMIM |
| COLORECTAL CANCER, HEREDITARY NONPOLYPOSIS; HNPCC | 114500 | Microsatellite instability; MSI | |
| | | mutL homolog 1, colon cancer, nonpolyposis type 2 (E. coli).; MLH1 | 120436 |
| | | MUTS HOMOLOG 2, COLON CANCER, NONPOLYPOSIS TYPE 1 (E. COLI); MSH2 | 120435 |
| | | MUTS HOMOLOG 6 (E. COLI); MSH6 | 600678 |
| CONGENITAL MUSCULAR DYSTROPHY WITH GLYCOSYLATION DEFECTS | | Fukuyama type congenital muscular dystrophy (fukutin); FCMD | 607440 |
| | | like-glycosyltransferase; LARGE | 603590 |
| | | protein O-linked mannose beta1,2-N-acetylglucosaminyltransferase; POMGNT1 | 606822 |
| | | protein-O-mannosyltransferase 1; POMT1 | 607423 |
| | | protein-O-mannosyltransferase 2; POMT2 | 607439 |
| COPROPORPHYRIA | 121300 | coproporphyrinogen oxidase; CPOX | 121300 |
| COSTELLO SYNDROME | 218040 | v-Ha-ras Harvey rat sarcoma viral oncogene homolog; HRAS | 190020 |
| COWDEN DISEASE; CD | 158350 | PHOSPHATASE AND TENSIN HOMOLOG (MUTATED IN MULTIPLE ADVANCED CANCERS 1); PTEN | 601728 |
| CRANIOFRONTONASAL SYNDROME; CFNS | 304110 | ephrin-B1; EFNB1 | 300035 |
| CRIGLER-NAJJAR SYNDROME | 218800 | UDP glucuronosyltransferase 1 family, polypeptide A1; UGT1A1 | 191740 |
| CROUZON SYNDROME | 123500 | FIBROBLAST GROWTH FACTOR RECEPTOR 1 (FMS-RELATED TYROSINE KINASE 2, PFEIFFER SYNDROME); FGFR1 | 136350 |
| | | FIBROBLAST GROWTH FACTOR RECEPTOR 2 (BACTERIA-EXPRESSED KINASE, KERATINOCYTE GROWTH FACTOR RECEPTOR, CRANIOFACIAL DYSOSTOSIS 1, CROUZON SYNDROME, PFEIFFER SYNDROME, JACKSON-WEISS SYNDROME); FGFR2 | 176943 |

| DISEASE | | GENE | |
|---|---|---|---|
| NAME | OMIM | NAME | OMIM |
| CROUZON SYNDROME | 123500 | FIBROBLAST GROWTH FACTOR RECEPTOR 3 (ACHONDROPLASIA, THANATOPHORIC DWARFISM); FGFR3 | 134934 |
| | | TWIST HOMOLOG 1 (ACROCEPHALOSYNDACTYLY 3; SAETHRE-CHOTZEN SYNDROME) (DROSOPHILA); TWIST1 | 601622 |
| CUTIS GYRATA SYNDROME OF BEARE AND STEVENSON | 123790 | FIBROBLAST GROWTH FACTOR RECEPTOR 1 (FMS-RELATED TYROSINE KINASE 2, PFEIFFER SYNDROME); FGFR1 | 136350 |
| | | FIBROBLAST GROWTH FACTOR RECEPTOR 2 (BACTERIA-EXPRESSED KINASE, KERATINOCYTE GROWTH FACTOR RECEPTOR, CRANIOFACIAL DYSOSTOSIS 1, CROUZON SYNDROME, PFEIFFER SYNDROME, JACKSON-WEISS SYNDROME); FGFR2 | 176943 |
| | | FIBROBLAST GROWTH FACTOR RECEPTOR 3 (ACHONDROPLASIA, THANATOPHORIC DWARFISM); FGFR3 | 134934 |
| | | TWIST HOMOLOG 1 (ACROCEPHALOSYNDACTYLY 3; SAETHRE-CHOTZEN SYNDROME) (DROSOPHILA); TWIST1 | 601622 |
| CYSTIC FIBROSIS; CF | 219700 | cystic fibrosis transmembrane conductance regulator (ATP-binding cassette sub-family C, member 7); CFTR | 602421 |
| CYSTINOSIS, ADULT NONNEPHROPATHIC | 219750 | CYSTINOSIS, NEPHROPATHIC; CTNS | 606272 |
| CYSTINOSIS, LATE-ONSET JUVENILE OR ADOLESCENT NEPHROPATHIC TYPE | 219900 | CYSTINOSIS, NEPHROPATHIC; CTNS | 606272 |
| CYSTINOSIS, NEPHROPATHIC; CTNS | 219800 | CYSTINOSIS, NEPHROPATHIC; CTNS | 606272 |
| DENTATORUBRAL-PALLIDOLUYSIAN ATROPHY; DRPLA | 125370 | atrophin 1; ATN1 | 607462 |

| DISEASE | | GENE | |
|---|---|---|---|
| NAME | OMIM | NAME | OMIM |
| DIABETES MELLITUS, PERMANENT NEONATAL; PNDM | 606176 | POTASSIUM INWARDLY-RECTIFYING CHANNEL, SUBFAMILY J, MEMBER 11; KCNJ11 | 600937 |
| DIABETES MELLITUS, TRANSIENT NEONATAL | 601410 | POTASSIUM INWARDLY-RECTIFYING CHANNEL, SUBFAMILY J, MEMBER 11; KCNJ11 | 600937 |
| DIABETES-DEAFNESS SYNDROME, MATERNALLY TRANSMITTED | 520000 | mitochondrially encoded tRNA leucine 1 (UUA/G); MT-TL1 | 590050 |
| DILATED CARDIOMYOPATHY; DCM | | myosin, heavy chain 7, cardiac muscle, beta; MYH7 | 160760 |
| | | troponin T type 2 (cardiac); TNNT2 | 191045 |
| DYSTONIA, FAMILIAL, WITH VISUAL FAILURE AND STRIATAL LUCENCIES | 500001 | mitochondrially encoded NADH dehydrogenase 6; MT-ND6 | 516006 |
| DYSTONIA, PROGRESSIVE, WITH DIURNAL VARIATION | 128230 | GTP CYCLOHYDROLASE 1 (DOPA-RESPONSIVE DYSTONIA); GCH1 | 600225 |
| DYSTROPHIA MYOTONICA 1 | 160900 | DYSTROPHIA MYOTONICA-PROTEIN KINASE; DMPK | 605377 |
| DYSTROPHIA MYOTONICA 2; DM2 | 602668 | CCHC-type zinc finger, nucleic acid binding protein; CNBP | 116955 |
| ECTODERMAL DYSPLASIA 1, ANHIDROTIC; ED1 | 305100 | ECTODYSPLASIN A; EDA | 300451 |
| ECTODERMAL DYSPLASIA 2, HIDROTIC; ED2 | 129500 | gap junction protein, beta 6; GJB6 | 604418 |
| ECTODERMAL DYSPLASIA 3, ANHIDROTIC; ED3 | 129490 | ECTODYSPLASIN A RECEPTOR; EDAR | 604095 |
| ECTODERMAL DYSPLASIA, ANHIDROTIC | 224900 | ECTODYSPLASIN A RECEPTOR; EDAR | 604095 |
| EHLERS-DANLOS SYNDROME, TYPE IV, AUTOSOMAL DOMINANT | 130050 | collagen, type III, alpha 1 (Ehlers-Danlos syndrome type IV, autosomal dominant); COL3A1 | 120180 |
| EMERY-DREIFUSS MUSCULAR DYSTROPHY, AUTOSOMAL DOMINANT; EDMD2 | 181350 | LAMIN A/C; LMNA | 150330 |
| EMERY-DREIFUSS MUSCULAR DYSTROPHY, X-LINKED; EDMD | 310300 | EMERIN (EMERY-DREIFUSS MUSCULAR DYSTROPHY); EMD | 300384 |

| DISEASE | | GENE | |
|---|---|---|---|
| NAME | OMIM | NAME | OMIM |
| EPIDERMOLYSIS BULLOSA HERPETIFORMIS, DOWLING-MEARA TYPE | 131760 | KERATIN 14 (EPIDERMOLYSIS BULLOSA SIMPLEX, DOWLING-MEARA, KOEBNER); KRT14 | 148066 |
| | | KERATIN 5 (EPIDERMOLYSIS BULLOSA SIMPLEX, DOWLING-MEARA/KOBNER/WEBER-COCKAYNE TYPES); KRT5 | 148040 |
| EPIDERMOLYSIS BULLOSA OF HANDS AND FEET | 131800 | KERATIN 14 (EPIDERMOLYSIS BULLOSA SIMPLEX, DOWLING-MEARA, KOEBNER); KRT14 | 148066 |
| | | KERATIN 5 (EPIDERMOLYSIS BULLOSA SIMPLEX, DOWLING-MEARA/KOBNER/WEBER-COCKAYNE TYPES); KRT5 | 148040 |
| EPIDERMOLYSIS BULLOSA SIMPLEX, KOEBNER TYPE; EBS2 | 131900 | KERATIN 14 (EPIDERMOLYSIS BULLOSA SIMPLEX, DOWLING-MEARA, KOEBNER); KRT14 | 148066 |
| | | KERATIN 5 (EPIDERMOLYSIS BULLOSA SIMPLEX, DOWLING-MEARA/KOBNER/WEBER-COCKAYNE TYPES); KRT5 | 148040 |
| EPILEPSY, MYOCLONIC, X-LINKED, WITH MENTAL RETARDATION AND SPASTICITY | 300432 | ARISTALESS RELATED HOMEOBOX; ARX | 300382 |
| EPIPHYSEAL DYSPLASIA, MULTIPLE, WITH EARLY-ONSET DIABETES MELLITUS | 226980 | EUKARYOTIC TRANSLATION INITIATION FACTOR 2-ALPHA KINASE 3; EIF2AK3 | 604032 |
| EXTRAPYRAMIDAL DISORDER, PROGRESSIVE, WITH PRIMARY HYPOGONADISM AND ALOPECIA | 601164 | TYROSINE HYDROXYLASE; TH | 191290 |
| FABRY DISEASE | 301500 | GALACTOSIDASE, ALPHA; GLA | 301500 |
| FACIOSCAPULOHUMERAL MUSCULAR DYSTROPHY 1A; FSHMD1A | 158900 | FACIOSCAPULOHUMERAL MUSCULAR DYSTROPHY 1A; FSHMD1A | 158900 |
| FACTOR V DEFICIENCY | 227400 | PROTEIN C (INACTIVATOR OF COAGULATION FACTORS VA AND VIIIA); PROC | 176860 |
| FANCONI ANEMIA; FA | | FANCONI ANEMIA, COMPLEMENTATION GROUP A; FANCA | 607139 |

| DISEASE | | GENE | |
|---|---|---|---|
| NAME | OMIM | NAME | OMIM |
| FANCONI ANEMIA; FA | | FANCONI ANEMIA, COMPLEMENTATION GROUP C; FANCC | 227645 |
| FRAGILE SITE MENTAL RETARDATION 1 GENE; FMR1 | 309550 | FRAGILE X MENTAL RETARDATION 1; FMR1 | 309550 |
| FRAGILE SITE, FOLIC ACID TYPE, RARE, FRA(X)(q28); FRAXE | 309548 | AF4/FMR2 family, member 2; AFF2 | 309548 |
| FRASIER SYNDROME | 136680 | WILMS TUMOR 1; WT1 | 607102 |
| FRIEDREICH ATAXIA 1; FRDA | 229300 | FRATAXIN; FXN | 606829 |
| FRONTOMETAPHYSEAL DYSPLASIA; FMD | 305620 | FILAMIN A, ALPHA (ACTIN BINDING PROTEIN 280); FLNA | 300017 |
| FRONTOTEMPORAL DEMENTIA | 600274 | MICROTUBULE-ASSOCIATED PROTEIN TAU; MAPT | 157140 |
| FRUCTOSE INTOLERANCE, HEREDITARY | 229600 | ALDOLASE B, FRUCTOSE-BISPHOSPHATE; ALDOB | 229600 |
| FUNDUS DYSTROPHY, PSEUDOINFLAMMATORY, OF SORSBY; SFD | 136900 | TIMP metallopeptidase inhibitor 3 (Sorsby fundus dystrophy, pseudoinflammatory); TIMP3 | 188826 |
| GALACTOSEMIA | 230400 | GALACTOSE-1-PHOSPHATE URIDYLYLTRANSFERASE; GALT | 606999 |
| GAP JUNCTION PROTEIN, BETA-2; GJB2 | 121011 | gap junction protein, beta 2, 26kDa; GJB2 | 121011 |
| GASTRIC CANCER | 137215 | CADHERIN 1, TYPE 1, E-CADHERIN (EPITHELIAL); CDH1 | 192090 |
| GAUCHER DISEASE, TYPE I | 230800 | GLUCOSIDASE, BETA; ACID (INCLUDES GLUCOSYLCERAMIDASE); GBA | 606463 |
| GAUCHER DISEASE, TYPE II | 230900 | GLUCOSIDASE, BETA; ACID (INCLUDES GLUCOSYLCERAMIDASE); GBA | 606463 |
| GAUCHER DISEASE, TYPE III | 231000 | GLUCOSIDASE, BETA; ACID (INCLUDES GLUCOSYLCERAMIDASE); GBA | 606463 |
| GENETIC IDENTITY | | LABORATORY SPECIFIC TEST | |
| GILBERT SYNDROME | 143500 | UDP glucuronosyltransferase 1 family, polypeptide A1; UGT1A1 | 191740 |

| DISEASE | | GENE | |
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| NAME | OMIM | NAME | OMIM |
| GITELMAN SYNDROME; GS | 263800 | solute carrier family 12 (sodium/chloride transporters), member 3; SLC12A3 | 600968 |
| GLUCOSE-6-PHOSPHATE DEHYDROGENASE; G6PD | 305900 | GLUCOSE-6-PHOSPHATE DEHYDROGENASE; G6PD | 305900 |
| GLUTARICACIDEMIA I | 231670 | glutaryl-Coenzyme A dehydrogenase; GCDH | 608801 |
| GLYCINE ENCEPHALOPATHY; GCE | 605899 | aminomethyltransferase; AMT | 238310 |
| | | glycine dehydrogenase (decarboxylating); GLDC | 238300 |
| GLYCOGEN STORAGE DISEASE I | 232200 | solute carrier family 37 (glucose-6-phosphate transporter), member 4; SLC37A4 | 602671 |
| GLYCOGEN STORAGE DISEASE Ib | 232220 | solute carrier family 37 (glucose-6-phosphate transporter), member 4; SLC37A4 | 602671 |
| GLYCOGEN STORAGE DISEASE Ic GLYCOGEN STORAGE DISEASE Id, INCLUDED | 232240 | solute carrier family 37 (glucose-6-phosphate transporter), member 4; SLC37A4 | 602671 |
| GLYCOGEN STORAGE DISEASE III | 232400 | AMYLO-1, 6-GLUCOSIDASE, 4-ALPHA-GLUCANOTRANSFERASE (GLYCOGEN DEBRANCHING ENZYME, GLYCOGEN STORAGE DISEASE TYPE III); AGL | 232400 |
| GLYCOGEN STORAGE DISEASE V | 232600 | PHOSPHORYLASE, GLYCOGEN; MUSCLE (MCARDLE SYNDROME, GLYCOGEN STORAGE DISEASE TYPE V); PYGM | 608455 |
| GONADAL DYSGENESIS, XY FEMALE TYPE; GDXY | 306100 | SEX DETERMINING REGION Y; SRY | 480000 |
| GREIG CEPHALOPOLYSYNDACTYLY SYNDROME; GCPS | 175700 | GLI-Kruppel family member GLI3 (Greig cephalopolysyndactyly syndrome); GLI3 | 165240 |
| GTP CYCLOHYDROLASE I DEFICIENCY; GTP | 233910 | GTP CYCLOHYDROLASE 1 (DOPA-RESPONSIVE DYSTONIA); GCH1 | 600225 |
| HEMOCHROMATOSIS; HFE | 235200 | HEMOCHROMATOSIS; HFE | 235200 |
| HEMOGLOBIN F, HEREDITARY PERSISTENCE OF, PANCELLULAR | 141749 | HEMOGLOBIN, BETA; HBB | 141900 |
| | | HEMOGLOBIN, DELTA; HBD | 142000 |
| | | HEMOGLOBIN, GAMMA A; HBG1 | 142200 |
| | | HEMOGLOBIN, GAMMA G; HBG2 | 142250 |

| DISEASE | | GENE | |
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| NAME | OMIM | NAME | OMIM |
| HEMOGLOBIN--VARIANTS FOR WHICH THE CHAIN CARRYING THE MUTATION IS UNKNOWN OR UNCERTAIN | 142309 | HEMOGLOBIN, ALPHA 1; HBA1 | 141800 |
| | | HEMOGLOBIN, ALPHA 2; HBA2 | 141850 |
| | | HEMOGLOBIN, BETA; HBB | 141900 |
| HEMOLYTIC-UREMIC SYNDROME; HUS | 235400 | COMPLEMENT FACTOR H; CFH | 134370 |
| HEMOPHILIA A | 306700 | COAGULATION FACTOR VIII, PROCOAGULANT COMPONENT (HEMOPHILIA A); F8 | 306700 |
| HEMOPHILIA B; HEMB | 306900 | coagulation factor IX (plasma thromboplastic component, Christmas disease, hemophilia B); F9 | 306900 |
| HEREDITARY PERSISTENCE OF FETAL HEMOGLOBIN, HETEROCELLULAR, INDIAN TYPE; HPFH2 | 142335 | HEMOGLOBIN, BETA; HBB | 141900 |
| | | HEMOGLOBIN, DELTA; HBD | 142000 |
| | | HEMOGLOBIN, GAMMA A; HBG1 | 142200 |
| | | HEMOGLOBIN, GAMMA G; HBG2 | 142250 |
| HETEROCELLULAR HEREDITARY PERSISTENCE OF FETAL HEMOGLOBIN; HPFH | 142470 | HEMOGLOBIN, BETA; HBB | 141900 |
| | | HEMOGLOBIN, DELTA; HBD | 142000 |
| | | HEMOGLOBIN, GAMMA A; HBG1 | 142200 |
| | | HEMOGLOBIN, GAMMA G; HBG2 | 142250 |
| HETEROTOPIA, PERIVENTRICULAR, X-LINKED DOMINANT | 300049 | FILAMIN A, ALPHA (ACTIN BINDING PROTEIN 280); FLNA | 300017 |
| HIRSCHSPRUNG DISEASE | 142623 | ret proto-oncogene; RET | 164761 |
| HOLT-ORAM SYNDROME; HOS | 142900 | T-BOX 5; TBX5 | 601620 |
| HOMOCYSTINURIA | 236200 | CYSTATHIONINE-BETA-SYNTHASE; CBS | 236200 |
| HOMOCYSTINURIA DUE TO DEFICIENCY OF N(5,10)-METHYLENETETRAHYDROFOLATE REDUCTASE ACTIVITY | 236250 | 5,10-METHYLENETETRAHYDROFOLATE REDUCTASE (NADPH); MTHFR | 607093 |

| DISEASE | | GENE | |
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| NAME | OMIM | NAME | OMIM |
| HUNTINGTON DISEASE; HD | 143100 | HUNTINGTIN (HUNTINGTON DISEASE); HD | 143100 |
| HURLER-SCHEIE SYNDROME; HSS | 607015 | IDURONIDASE, ALPHA-L-; IDUA | 252800 |
| HUTCHINSON-GILFORD PROGERIA SYNDROME; HGPS | 176670 | LAMIN A/C; LMNA | 150330 |
| HYDROCEPHALUS DUE TO CONGENITAL STENOSIS OF AQUEDUCT OF SYLVIUS; HSAS | 307000 | L1 cell adhesion molecule; L1CAM | 308840 |
| HYPERALDOSTERONISM, FAMILIAL, TYPE I | 103900 | CYTOCHROME P450, FAMILY 11, SUBFAMILY B, POLYPEPTIDE 1; CYP11B1 | 202010 |
| | | CYTOCHROME P450, FAMILY 11, SUBFAMILY B, POLYPEPTIDE 2; CYP11B2 | 124080 |
| HYPERCHOLESTEROLEMIA, AUTOSOMAL DOMINANT | 143890 | LOW DENSITY LIPOPROTEIN RECEPTOR (FAMILIAL HYPERCHOLESTEROLEMIA); LDLR | 606945 |
| HYPERCHOLESTEROLEMIA, AUTOSOMAL DOMINANT, TYPE B | 144010 | APOLIPOPROTEIN B (INCLUDING AG(X) ANTIGEN); APOB | 107730 |
| HYPERINSULINISM, AUTOSOMAL DOMINANT | 602485 | GLUCOKINASE (HEXOKINASE 4, MATURITY ONSET DIABETES OF THE YOUNG 2); GCK | 138079 |
| HYPERINSULINISM-HYPERAMMONEMIA SYNDROME | 606762 | GLUTAMATE DEHYDROGENASE 1; GLUD1 | 138130 |
| HYPERKALEMIC PERIODIC PARALYSIS; HYPP | 170500 | sodium channel, voltage-gated, type IV, alpha subunit; SCN4A | 603967 |
| HYPEROXALURIA, PRIMARY, TYPE I | 259900 | alanine-glyoxylate aminotransferase (oxalosis I; hyperoxaluria I; glycolicaciduria; serine-pyruvate aminotransferase); AGXT | 604285 |
| HYPEROXALURIA, PRIMARY, TYPE II | 260000 | glyoxylate reductase/hydroxypyruvate reductase; GRHPR | 604296 |
| HYPERPARATHYROIDISM 2; HRPT2 | 145001 | cell division cycle 73, Paf1/RNA polymerase II complex component, homolog (S. cerevisiae); CDC73 | 607393 |
| HYPERPARATHYROIDISM, NEONATAL SEVERE PRIMARY; NSHPT | 239200 | CALCIUM-SENSING RECEPTOR (HYPOCALCIURIC HYPERCALCEMIA 1, SEVERE NEONATAL HYPERPARATHYROIDISM); CASR | 601199 |

| DISEASE | | GENE | |
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| NAME | OMIM | NAME | OMIM |
| HYPERTHYROIDISM, FAMILIAL GESTATIONAL | 603373 | THYROID STIMULATING HORMONE RECEPTOR; TSHR | 603372 |
| HYPERURICEMIC NEPHROPATHY, FAMILIAL JUVENILE; HNFJ | 162000 | uromodulin (uromucoid, Tamm-Horsfall glycoprotein); UMOD | 191845 |
| HYPOCALCIURIC HYPERCALCEMIA, FAMILIAL, TYPE I; HHC1 | 145980 | CALCIUM-SENSING RECEPTOR (HYPOCALCIURIC HYPERCALCEMIA 1, SEVERE NEONATAL HYPERPARATHYROIDISM); CASR | 601199 |
| HYPOCHONDROPLASIA; HCH | 146000 | FIBROBLAST GROWTH FACTOR RECEPTOR 3 (ACHONDROPLASIA, THANATOPHORIC DWARFISM); FGFR3 | 134934 |
| HYPOKALEMIC PERIODIC PARALYSIS; HOKPP | 170400 | CALCIUM CHANNEL, VOLTAGE-DEPENDENT, L TYPE, ALPHA 1S SUBUNIT; CACNA1S | 114208 |
| HYPOPARATHYROIDISM, FAMILIAL ISOLATED; FIH | 146200 | CALCIUM-SENSING RECEPTOR (HYPOCALCIURIC HYPERCALCEMIA 1, SEVERE NEONATAL HYPERPARATHYROIDISM); CASR | 601199 |
| | | GLIAL CELLS MISSING HOMOLOG 2 (DROSOPHILA); GCM2 | 603716 |
| | | PARATHYROID HORMONE; PTH | 168450 |
| HYPOPARATHYROIDISM, SENSORINEURAL DEAFNESS, AND RENAL DYSPLASIA | 146255 | GATA BINDING PROTEIN 3; GATA3 | 131320 |
| HYPOPHOSPHATEMIC RICKETS, AUTOSOMAL DOMINANT; ADHR | 193100 | FIBROBLAST GROWTH FACTOR 23; FGF23 | 605380 |
| | | PHOSPHATE REGULATING ENDOPEPTIDASE HOMOLOG, X-LINKED (HYPOPHOSPHATEMIA, VITAMIN D RESISTANT RICKETS); PHEX | 307800 |
| ICHTHYOSIS, BULLOUS TYPE | 146800 | keratin 2 (epidermal ichthyosis bullosa of Siemens); KRT2 | 600194 |
| ICHTHYOSIS, X-LINKED | 308100 | STEROID SULFATASE (MICROSOMAL), ISOZYME S; STS | 308100 |

| DISEASE | | GENE | |
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| NAME | OMIM | NAME | OMIM |
| IMMUNODEFICIENCY WITH HYPER-IgM, TYPE 1; HIGM1 | 308230 | CD40 ligand (TNF superfamily, member 5, hyper-IgM syndrome); CD40LG | 300386 |
| INCONTINENTIA PIGMENTI; IP | 308300 | INHIBITOR OF KAPPA LIGHT POLYPEPTIDE GENE ENHANCER IN B-CELLS, KINASE GAMMA; IKBKG | 300248 |
| INFANTILE SPASM SYNDROME, X-LINKED | 308350 | ARISTALESS RELATED HOMEOBOX; ARX | 300382 |
| JACKSON-WEISS SYNDROME; JWS | 123150 | FIBROBLAST GROWTH FACTOR RECEPTOR 2 (BACTERIA-EXPRESSED KINASE, KERATINOCYTE GROWTH FACTOR RECEPTOR, CRANIOFACIAL DYSOSTOSIS 1, CROUZON SYNDROME, PFEIFFER SYNDROME, JACKSON-WEISS SYNDROME); FGFR2 | 176943 |
| KEARNS-SAYRE SYNDROME; KSS | 530000 | KEARNS-SAYRE SYNDROME; KSS | 530000 |
| | | MITOCHONDRIAL GENOME | |
| KRABBE DISEASE | 245200 | galactosylceramidase; GALC | 606890 |
| LEBER OPTIC ATROPHY | 535000 | mitochondrially encoded ATP synthase 6; MT-ATP6 | 516060 |
| | | mitochondrially encoded NADH dehydrogenase 1; MT-ND1 | 516000 |
| | | mitochondrially encoded NADH dehydrogenase 4; MT-ND4 | 516003 |
| | | mitochondrially encoded NADH dehydrogenase 6; MT-ND6 | 516006 |
| LEIGH SYNDROME; LS | 256000 | mitochondrially encoded ATP synthase 6; MT-ATP6 | 516060 |
| | | mitochondrially encoded NADH dehydrogenase 6; MT-ND6 | 516006 |
| LERI-WEILL DYSCHONDROSTEOSIS; LWD | 127300 | SHORT STATURE HOMEOBOX; SHOX | 312865 |
| LESCH-NYHAN SYNDROME; LNS | 300322 | HYPOXANTHINE PHOSPHORIBOSYLTRANSFERASE 1 (LESCH-NYHAN SYNDROME); HPRT1 | 308000 |
| LI-FRAUMENI SYNDROME; LFS | 151623 | TUMOR PROTEIN P53 (LI-FRAUMENI SYNDROME); TP53 | 191170 |

| DISEASE | | GENE | |
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| NAME | OMIM | NAME | OMIM |
| LIDDLE SYNDROME | 177200 | sodium channel, nonvoltage-gated 1, beta (Liddle syndrome); SCNN1B | 600760 |
| | | sodium channel, nonvoltage-gated 1, gamma; SCNN1G | 600761 |
| LIPODYSTROPHY, FAMILIAL PARTIAL, TYPE 2; FPLD2 | 151660 | LAMIN A/C; LMNA | 150330 |
| LISSENCEPHALY I; LIS1 | 607432 | PLATELET-ACTIVATING FACTOR ACETYLHYDROLASE, ISOFORM IB, ALPHA SUBUNIT 45KDA; PAFAH1B1 | 601545 |
| LISSENCEPHALY, X-LINKED, WITH AMBIGUOUS GENITALIA; XLAG | 300215 | ARISTALESS RELATED HOMEOBOX; ARX | 300382 |
| LISSENCEPHALY, X-LINKED; LISX | 300067 | DOUBLECORTEX; LISSENCEPHALY, X-LINKED (DOUBLECORTIN); DCX | 300121 |
| LONG QT SYNDROME 1; LQT1 | 192500 | POTASSIUM VOLTAGE-GATED CHANNEL, KQT-LIKE SUBFAMILY, MEMBER 1; KCNQ1 | 607542 |
| LONG QT SYNDROME 3; LQT3 | 603830 | sodium channel, voltage-gated, type V, alpha subunit; SCN5A | 600163 |
| LOWE OCULOCEREBRORENAL SYNDROME; OCRL | 309000 | OCULOCEREBRORENAL SYNDROME OF LOWE; OCRL | 309000 |
| LYMPHEDEMA, HEREDITARY, I | 153100 | FMS-RELATED TYROSINE KINASE 4; FLT4 | 136352 |
| LYMPHEDEMA-DISTICHIASIS SYNDROME | 153400 | FORKHEAD BOX C2 (MFH-1, MESENCHYME FORKHEAD 1); FOXC2 | 602402 |
| LYMPHOPROLIFERATIVE SYNDROME, X-LINKED | 308240 | LYMPHOPROLIFERATIVE SYNDROME, X-LINKED | 308240 |
| MACHADO-JOSEPH DISEASE; MJD | 109150 | ATAXIN 3; ATXN3 | 607047 |
| MACROCEPHALY, MULTIPLE LIPOMAS, AND HEMANGIOMATA | 153480 | PHOSPHATASE AND TENSIN HOMOLOG (MUTATED IN MULTIPLE ADVANCED CANCERS 1); PTEN | 601728 |
| MALIGNANT HYPERTHERMIA, SUSCEPTIBILITY TO, 1; MHS1 | 145600 | RYANODINE RECEPTOR 1 (SKELETAL); RYR1 | 180901 |
| MANDIBULOACRAL DYSPLASIA WITH TYPE A LIPODYSTROPHY; MADA | 248370 | LAMIN A/C; LMNA | 150330 |
| MARFAN SYNDROME, TYPE II; MFS2 | 154705 | transforming growth factor, beta receptor II (70/80kDa); TGFBR2 | 190182 |

| DISEASE | | GENE | |
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| NAME | OMIM | NAME | OMIM |
| MARFAN SYNDROME; MFS | 154700 | fibrillin 1; FBN1 | 134797 |
| MATURITY-ONSET DIABETES OF THE YOUNG, TYPE V; MODY5 | 604284 | TRANSCRIPTION FACTOR 2, HEPATIC; LF-B3; VARIANT HEPATIC NUCLEAR FACTOR; TCF2 | 189907 |
| MATURITY-ONSET DIABETES OF THE YOUNG; MODY | 606391 | GLUCOKINASE (HEXOKINASE 4, MATURITY ONSET DIABETES OF THE YOUNG 2); GCK | 138079 |
| | | HEPATOCYTE NUCLEAR FACTOR 4, ALPHA; HNF4A | 600281 |
| | | NEUROGENIC DIFFERENTIATION 1; NEUROD1 | 601724 |
| | | pancreatic and duodenal homeobox 1; PDX1 | 600733 |
| | | TRANSCRIPTION FACTOR 1, HEPATIC; LF-B1, HEPATIC NUCLEAR FACTOR (HNF1), ALBUMIN PROXIMAL FACTOR; TCF1 | 142410 |
| MEDULLARY CYSTIC KIDNEY DISEASE 2; MCKD2 | 603860 | uromodulin (uromucoid, Tamm-Horsfall glycoprotein); UMOD | 191845 |
| MELANOMA, CUTANEOUS MALIGNANT; CMM | 155600 | CYCLIN-DEPENDENT KINASE INHIBITOR 2A (MELANOMA, P16, INHIBITS CDK4); CDKN2A | 600160 |
| MELNICK-NEEDLES SYNDROME; MNS | 309350 | FILAMIN A, ALPHA (ACTIN BINDING PROTEIN 280); FLNA | 300017 |
| MENKES DISEASE; MENKE | 309400 | ATPase, Cu++ transporting, alpha polypeptide (Menkes syndrome); ATP7A | 300011 |
| MENTAL RETARDATION, X-LINKED 36; MRX36 | 300430 | ARISTALESS RELATED HOMEOBOX; ARX | 300382 |
| METACHROMATIC LEUKODYSTROPHY | 250100 | ARYLSULFATASE A; ARSA | 607574 |
| MITOCHONDRIAL DISORDERS | | LABORATORY SPECIFIC TEST | |
| MITOCHONDRIAL DNA DEPLETION SYNDROME, HEPATOCEREBRAL FORM MITOCHONDRIAL DNA DEPLETION MYOPATHY, INCLUDED | 251880 | MITOCHONDRIAL GENOME | |
| MITOCHONDRIAL MYOPATHY | 251900 | MITOCHONDRIAL MYOPATHY | 251900 |

| DISEASE | | GENE | |
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| NAME | OMIM | NAME | OMIM |
| MITOCHONDRIAL MYOPATHY WITH DIABETES | 500002 | TRANSFER RNA, MITOCHONDRIAL, GLUTAMIC ACID; MTTE | 590025 |
| MITOCHONDRIAL MYOPATHY, ENCEPHALOPATHY, LACTIC ACIDOSIS, AND STROKE-LIKE EPISODES; MELAS | 540000 | mitochondrially encoded tRNA leucine 1 (UUA/G); MT-TL1 | 590050 |
| MITOCHONDRIAL NEUROGASTROINTESTINAL ENCEPHALOPATHY SYNDROME; MNGIE | 603041 | ENDOTHELIAL CELL GROWTH FACTOR 1 (PLATELET-DERIVED); ECGF1 | 131222 |
| | | MITOCHONDRIAL GENOME | |
| MITOCHONDRIAL-REALTED DEAFNESS | | mitochondrially encoded 12S RNA; MT-RNR1 | 561000 |
| MOWAT-WILSON SYNDROME | 235730 | zinc finger E-box binding homeobox 2; ZEB2 | 605802 |
| MUCOPOLYSACCHARIDOSIS TYPE II | 309900 | IDURONATE 2-SULFATASE (HUNTER SYNDROME); IDS | 309900 |
| MUCOPOLYSACCHARIDOSIS TYPE IIIA | 252900 | N-SULFOGLUCOSAMINE SULFOHYDROLASE (SULFAMIDASE); SGSH | 605270 |
| MUCOPOLYSACCHARIDOSIS TYPE IVA | 253000 | GALACTOSAMINE (N-ACETYL)-6-SULFATE SULFATASE (MORQUIO SYNDROME, MUCOPOLYSACCHARIDOSIS TYPE IVA); GALNS | 253000 |
| MUENKE SYNDROME | 602849 | FIBROBLAST GROWTH FACTOR RECEPTOR 3 (ACHONDROPLASIA, THANATOPHORIC DWARFISM); FGFR3 | 134934 |
| MUIR-TORRE SYNDROME; MTS | 158320 | mutL homolog 1, colon cancer, nonpolyposis type 2 (E. coli).; MLH1 | 120436 |
| | | MUTS HOMOLOG 2, COLON CANCER, NONPOLYPOSIS TYPE 1 (E. COLI); MSH2 | 120435 |
| MULTIPLE ENDOCRINE NEOPLASIA, TYPE I; MEN1 | 131100 | SECRETOGRANIN II (CHROMOGRANIN C); SCG2 | 118930 |
| MULTIPLE ENDOCRINE NEOPLASIA, TYPE II; MEN2 | 171400 | ret proto-oncogene; RET | 164761 |
| MULTIPLE ENDOCRINE NEOPLASIA, TYPE IIB; MEN2B | 162300 | ret proto-oncogene; RET | 164761 |

| DISEASE | | GENE | |
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| NAME | OMIM | NAME | OMIM |
| MUSCULAR DYSTROPHY, BECKER TYPE; BMD | 300376 | DYSTROPHIN (MUSCULAR DYSTROPHY, DUCHENNE AND BECKER TYPES); DMD | 300377 |
| MUSCULAR DYSTROPHY, CONGENITAL MEROSIN-DEFICIENT, 1A; MDC1A | 607855 | LAMININ, ALPHA 2 (MEROSIN, CONGENITAL MUSCULAR DYSTROPHY); LAMA2 | 156225 |
| MUSCULAR DYSTROPHY, CONGENITAL, 1C; MDC1C | 606612 | FUKUTIN RELATED PROTEIN; FKRP | 606596 |
| MUSCULAR DYSTROPHY, DUCHENNE TYPE; DMD | 310200 | DYSTROPHIN (MUSCULAR DYSTROPHY, DUCHENNE AND BECKER TYPES); DMD | 300377 |
| MUSCULAR DYSTROPHY, LIMB-GIRDLE, TYPE 1B; LGMD1B | 159001 | LAMIN A/C; LMNA | 150330 |
| MUSCULAR DYSTROPHY, LIMB-GIRDLE, TYPE 2A; LGMD2A | 253600 | FUKUTIN RELATED PROTEIN; FKRP | 606596 |
| | | LAMIN A/C; LMNA | 150330 |
| MutY, E. COLI, HOMOLOG OF; MUTYH | 604933 | MUTY HOMOLOG (E. COLI); MUTYH | 604933 |
| MYASTHENIC SYNDROME, CONGENITAL, ASSOCIATED WITH ACETYLCHOLINE RECEPTOR DEFICIENCY | 608931 | cholinergic receptor, nicotinic, epsilon; CHRNE | 100725 |
| | | receptor-associated protein of the synapse, 43kD; RAPSN | 601592 |
| MYASTHENIC SYNDROME, CONGENITAL, ASSOCIATED WITH EPISODIC APNEA | 254210 | CHOLINE ACETYLTRANSFERASE; CHAT | 118490 |
| MYASTHENIC SYNDROME, CONGENITAL, FAST-CHANNEL | 608930 | cholinergic receptor, nicotinic, alpha 1 (muscle); CHRNA1 | 100690 |
| | | cholinergic receptor, nicotinic, delta; CHRND | 100720 |
| | | cholinergic receptor, nicotinic, epsilon; CHRNE | 100725 |

| DISEASE | | GENE | |
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| NAME | OMIM | NAME | OMIM |
| MYASTHENIC SYNDROME, CONGENITAL, SLOW-CHANNEL; SCCMS | 601462 | CHOLINE ACETYLTRANSFERASE; CHAT | 118490 |
| | | cholinergic receptor, nicotinic, alpha 1 (muscle); CHRNA1 | 100690 |
| | | cholinergic receptor, nicotinic, beta polypeptide 1 (muscle) ; CHRNB1 | 100710 |
| | | cholinergic receptor, nicotinic, delta; CHRND | 100720 |
| | | cholinergic receptor, nicotinic, epsilon; CHRNE | 100725 |
| MYOCLONIC EPILEPSY ASSOCIATED WITH RAGGED-RED FIBERS; MERRF | 545000 | mitochondrially encoded tRNA lysine; MT-TK | 590060 |
| MYOCLONIC EPILEPSY OF UNVERRICHT AND LUNDBORG | 254800 | CYSTATIN B (STEFIN B); CSTB | 601145 |
| NAIL-PATELLA SYNDROME; NPS | 161200 | LIM HOMEOBOX TRANSCRIPTION FACTOR 1, BETA; LMX1B | 602575 |
| NEPHROLITHIASIS, X-LINKED RECESSIVE, WITH RENAL FAILURE; XRN | 310468 | CHLORIDE CHANNEL 5 (NEPHROLITHIASIS 2, X-LINKED, DENT DISEASE); CLCN5 | 300008 |
| NEPHRONOPHTHISIS 1; NPHP1 | 256100 | NEPHRONOPHTHISIS 1; NPHP1 | 256100 |
| NEPHROTIC SYNDROME, STEROID-RESISTANT, AUTOSOMAL RECESSIVE; SRN1 | 600995 | nephrosis 2, idiopathic, steroid-resistant (podocin); NPHS2 | 604766 |
| NEUROFIBROMATOSIS, TYPE I; NF1 | 162200 | NEUROFIBROMIN 1 (NEUROFIBROMATOSIS, VON RECKLINGHAUSEN DISEASE, WATSON DISEASE); NF1 | 162200 |
| NEUROFIBROMATOSIS, TYPE II; NF2 | 101000 | NEUROFIBROMIN 2 (BILATERAL ACOUSTIC NEUROMA); NF2 | 607379 |
| NEUROLOGICAL DISORDERS | | MITOCHONDRIAL GENOME | |
| | | mitochondrially encoded ATP synthase 6; MT-ATP6 | 516060 |
| | | mitochondrially encoded tRNA leucine 1 (UUA/G); MT-TL1 | 590050 |
| | | mitochondrially encoded tRNA lysine; MT-TK | 590060 |

| DISEASE | | GENE | |
|---|---|---|---|
| NAME | OMIM | NAME | OMIM |
| NEUROMUSCULAR DISEASE | | MITOCHONDRIAL GENOME | |
| | | mitochondrially encoded ATP synthase 6; MT-ATP6 | 516060 |
| | | mitochondrially encoded tRNA leucine 1 (UUA/G); MT-TL1 | 590050 |
| | | mitochondrially encoded tRNA lysine; MT-TK | 590060 |
| NEUROPATHY, ATAXIA, AND RETINITIS PIGMENTOSA | 551500 | mitochondrially encoded ATP synthase 6; MT-ATP6 | 516060 |
| NEUROPATHY, HEREDITARY SENSORY AND AUTONOMIC, TYPE I; HSAN1 | 162400 | SERINE PALMITOYLTRANSFERASE, LONG CHAIN BASE SUBUNIT 1; SPTLC1 | 605712 |
| NEUROPATHY, HEREDITARY SENSORY AND AUTONOMIC, TYPE III; HSAN3 | 223900 | INHIBITOR OF KAPPA LIGHT POLYPEPTIDE GENE ENHANCER IN B-CELLS, KINASE COMPLEX-ASSOCIATED PROTEIN; IKBKAP | 603722 |
| NEUROPATHY, HEREDITARY, WITH LIABILITY TO PRESSURE PALSIES; HNPP | 162500 | PERIPHERAL MYELIN PROTEIN 22; PMP22 | 601097 |
| NIEMANN-PICK DISEASE, TYPE C1; NPC1 | 257220 | NIEMANN-PICK DISEASE, TYPE C1; NPC1 | 607623 |
| NOONAN SYNDROME 1; NS1 | 163950 | PROTEIN TYROSINE PHOSPHATASE, NON-RECEPTOR TYPE 11 (NOONAN SYNDROME 1); PTPN11 | 176876 |
| NORRIE DISEASE; NDP | 310600 | NORRIE DISEASE (PSEUDOGLIOMA); NDP | 310600 |
| OCULOPHARYNGEAL MUSCULAR DYSTROPHY; OPMD | 164300 | POLY(A) BINDING PROTEIN, NUCLEAR 1; PABPN1 | 602279 |
| ORNITHINE TRANSCARBAMYLASE DEFICIENCY, HYPERAMMONEMIA DUE TO | 311250 | ORNITHINE CARBAMOYLTRANSFERASE; OTC | 300461 |
| OSTEOGENESIS IMPERFECTA CONGENITA; OIC | 166210 | COLLAGEN, TYPE I, ALPHA 1; COL1A1 | 120150 |
| | | COLLAGEN, TYPE I, ALPHA 2; COL1A2 | 120160 |
| OSTEOGENESIS IMPERFECTA, TYPE IV | 166220 | COLLAGEN, TYPE I, ALPHA 1; COL1A1 | 120150 |
| | | COLLAGEN, TYPE I, ALPHA 2; COL1A2 | 120160 |

| DISEASE | | GENE | |
|---|---|---|---|
| NAME | OMIM | NAME | OMIM |
| OSTEOPETROSIS, AUTOSOMAL RECESSIVE | 259700 | T-cell, immune regulator 1, ATPase, H+ transporting, lysosomal V0 subunit A3; TCIRG1 | 604592 |
| OTOPALATODIGITAL SYNDROME, TYPE I; OPD1 | 311300 | FILAMIN A, ALPHA (ACTIN BINDING PROTEIN 280); FLNA | 300017 |
| OTOPALATODIGITAL SYNDROME, TYPE II; OPD2 | 304120 | FILAMIN A, ALPHA (ACTIN BINDING PROTEIN 280); FLNA | 300017 |
| PACHYONYCHIA CONGENITA, TYPE 1; PC1 | 167200 | KERATIN 16 (FOCAL NON-EPIDERMOLYTIC PALMOPLANTAR KERATODERMA); KRT16 | 148067 |
| | | KERATIN 17; KRT17 | 148069 |
| | | KERATIN 6A; KRT6A | 148041 |
| | | KERATIN 6B; KRT6B | 148042 |
| PACHYONYCHIA CONGENITA, TYPE 2; PC2 | 167210 | KERATIN 16 (FOCAL NON-EPIDERMOLYTIC PALMOPLANTAR KERATODERMA); KRT16 | 148067 |
| | | KERATIN 17; KRT17 | 148069 |
| | | KERATIN 6A; KRT6A | 148041 |
| | | KERATIN 6B; KRT6B | 148042 |
| PALLISTER-HALL SYNDROME; PHS | 146510 | GLI-Kruppel family member GLI3 (Greig cephalopolysyndactyly syndrome); GLI3 | 165240 |
| PALMOPLANTAR KERATODERMA, EPIDERMOLYTIC; EPPK | 144200 | KERATIN 9 (EPIDERMOLYTIC PALMOPLANTAR KERATODERMA); KRT9 | 607606 |
| PANCREATITIS, HEREDITARY; PCTT | 167800 | PROTEASE, SERINE, 1 (TRYPSIN 1); PRSS1 | 276000 |
| PARAGANGLIOMAS 1; PGL1 | 168000 | SUCCINATE DEHYDROGENASE COMPLEX, SUBUNIT D, INTEGRAL MEMBRANE PROTEIN; SDHD | 602690 |
| PARKINSON DISEASE, JUVENILE, AUTOSOMAL RECESSIVE; PJD | 600116 | Parkinson disease (autosomal recessive, juvenile) 2, parkin; PARK2 | 602544 |
| PARTINGTON X-LINKED MENTAL RETARDATION SYNDROME; PRTS | 309510 | ARISTALESS RELATED HOMEOBOX; ARX | 300382 |
| PEARSON MARROW-PANCREAS SYNDROME | 557000 | MITOCHONDRIAL GENOME | |

| DISEASE | | GENE | |
|---|---|---|---|
| NAME | OMIM | NAME | OMIM |
| PELIZAEUS-MERZBACHER DISEASE; PMD | 312080 | PELIZAEUS-MERZBACHER DISEASE; PMD | 312080 |
| PENDRED SYNDROME; PDS | 274600 | solute carrier family 26, member 4; SLC26A4 | 605646 |
| PERSISTENT HYPERINSULINEMIC HYPOGLYCEMIA OF INFANCY | 601820 | ATP-BINDING CASSETTE, SUB-FAMILY C (CFTR/MRP), MEMBER 8; ABCC8 | 600509 |
| | | POTASSIUM INWARDLY-RECTIFYING CHANNEL, SUBFAMILY J, MEMBER 11; KCNJ11 | 600937 |
| PEUTZ-JEGHERS SYNDROME; PJS | 175200 | serine/threonine kinase 11; STK11 | 602216 |
| PFEIFFER SYNDROME | 101600 | FIBROBLAST GROWTH FACTOR RECEPTOR 1 (FMS-RELATED TYROSINE KINASE 2, PFEIFFER SYNDROME); FGFR1 | 136350 |
| | | FIBROBLAST GROWTH FACTOR RECEPTOR 2 (BACTERIA-EXPRESSED KINASE, KERATINOCYTE GROWTH FACTOR RECEPTOR, CRANIOFACIAL DYSOSTOSIS 1, CROUZON SYNDROME, PFEIFFER SYNDROME, JACKSON-WEISS SYNDROME); FGFR2 | 176943 |
| | | FIBROBLAST GROWTH FACTOR RECEPTOR 3 (ACHONDROPLASIA, THANATOPHORIC DWARFISM); FGFR3 | 134934 |
| | | TWIST HOMOLOG 1 (ACROCEPHALOSYNDACTYLY 3; SAETHRE-CHOTZEN SYNDROME) (DROSOPHILA); TWIST1 | 601622 |
| PHENYLKETONURIA | 261600 | PHENYLALANINE HYDROXYLASE; PAH | 261600 |
| PHEOCHROMOCYTOMA | 171300 | SUCCINATE DEHYDROGENASE COMPLEX, SUBUNIT B, IRON SULFUR (IP); SDHB | 185470 |
| | | SUCCINATE DEHYDROGENASE COMPLEX, SUBUNIT D, INTEGRAL MEMBRANE PROTEIN; SDHD | 602690 |
| PHOSPHATASE AND TENSIN HOMOLOG; PTEN | 601728 | PHOSPHATASE AND TENSIN HOMOLOG (MUTATED IN MULTIPLE ADVANCED CANCERS 1); PTEN | 601728 |

| DISEASE | | GENE | |
|---|---|---|---|
| NAME | OMIM | NAME | OMIM |
| PITUITARY DWARFISM III | 262600 | POU CLASS 1, HOMEOBOX 1; POU1F1 | 173110 |
| | | PROP PAIRED-LIKE HOMEOBOX 1; PROP1 | 601538 |
| POLYCYSTIC KIDNEY DISEASE, AUTOSOMAL RECESSIVE; ARPKD | 263200 | POLYCYSTIC KIDNEY AND HEPATIC DISEASE 1 (AUTOSOMAL RECESSIVE); PKHD1 | 606702 |
| POLYCYSTIC KIDNEYS | 173900 | POLYCYSTIC KIDNEY DISEASE 1 (AUTOSOMAL DOMINANT); PKD1 | 601313 |
| | | POLYCYSTIC KIDNEY DISEASE 2 (AUTOSOMAL DOMINANT); PKD2 | 173910 |
| POLYDACTYLY, PREAXIAL IV | 174700 | GLI-Kruppel family member GLI3 (Greig cephalopolysyndactyly syndrome); GLI3 | 165240 |
| POLYPOSIS, JUVENILE INTESTINAL | 174900 | BONE MORPHOGENETIC PROTEIN RECEPTOR, TYPE IA; BMPR1A | 601299 |
| | | MUTY HOMOLOG (E. COLI); MUTYH | 604933 |
| | | SMAD family member 4; SMAD4 | 600993 |
| POPLITEAL PTERYGIUM SYNDROME; PPS | 119500 | interferon regulatory factor 6; IRF6 | 607199 |
| PORPHYRIA CUTANEA TARDA | 176100 | uroporphyrinogen decarboxylase; UROD | 176100 |
| PORPHYRIA VARIEGATA | 176200 | protoporphyrinogen oxidase; PPOX | 600923 |
| PORPHYRIA, ACUTE INTERMITTENT | 176000 | hydroxymethylbilane synthase; HMBS | 176000 |
| PORPHYRIA, CONGENITAL ERYTHROPOIETIC | 263700 | uroporphyrinogen III synthase (congenital erythropoietic porphyria); UROS | 606938 |
| POTASSIUM CHANNEL, VOLTAGE-GATED, ISK-RELATED SUBFAMILY, MEMBER 1; KCNE1 | 176261 | POTASSIUM CHANNEL, VOLTAGE-GATED, ISK-RELATED SUBFAMILY, MEMBER 1; KCNE1 | 176261 |
| POTASSIUM CHANNEL, VOLTAGE-GATED, ISK-RELATED SUBFAMILY, MEMBER 2; KCNE2 | 603796 | POTASSIUM VOLTAGE-GATED CHANNEL, ISK-RELATED FAMILY, MEMBER 2; KCNE2 | 603796 |
| POTASSIUM CHANNEL, VOLTAGE-GATED, SUBFAMILY H, MEMBER 2; KCNH2 | 152427 | POTASSIUM VOLTAGE-GATED CHANNEL, SUBFAMILY H (EAG-RELATED), MEMBER 2; KCNH2 | 152427 |

| DISEASE | | GENE | |
|---|---|---|---|
| NAME | OMIM | NAME | OMIM |
| PRADER-WILLI SYNDROME; PWS | 176270 | NECDIN HOMOLOG (MOUSE); NDN | 602117 |
| | | SMALL NUCLEAR RIBONUCLEOPROTEIN POLYPEPTIDE N; SNRPN | 182279 |
| PROGRESSIVE EXTERNAL OPHTHALMOPLEGIA WITH MITOCHONDRIAL DNA DELETIONS | 157640 | mitochondrially encoded NADH dehydrogenase 5; MT-ND5 | 516005 |
| | | mitochondrially encoded NADH dehydrogenase 6; MT-ND6 | 516006 |
| | | POLYMERASE (DNA DIRECTED), GAMMA; POLG | 174763 |
| | | PROGRESSIVE EXTERNAL OPHTHALMOPLEGIA 1; PEO1 | 606075 |
| PROTEASE INHIBITOR 1; PI | 107400 | serpin peptidase inhibitor, clade A (alpha-1 antiproteinase, antitrypsin), member 1; SERPINA1 | 107400 |
| PROTOPORPHYRIA, ERYTHROPOIETIC | 177000 | ferrochelatase (protoporphyria); FECH | 177000 |
| PROXIMAL MYOPATHY WITH FOCAL DEPLETION OF MITOCHONDRIA | 600706 | MITOCHONDRIAL GENOME | |
| PSEUDOACHONDROPLASTIC DYSPLASIA; PSACH | 177170 | CARTILAGE OLIGOMERIC MATRIX PROTEIN; COMP | 600310 |
| RENAL CELL CARCINOMA, PAPILLARY | 605074 | MET PROTO-ONCOGENE (HEPATOCYTE GROWTH FACTOR RECEPTOR); MET | 164860 |
| RENAL TUBULAR ACIDOSIS, DISTAL, AUTOSOMAL DOMINANT | 179800 | solute carrier family 4, anion exchanger, member 1 (erythrocyte membrane protein band 3, Diego blood group); SLC4A1 | 109270 |
| RETICULOSIS, FAMILIAL HISTIOCYTIC; FHL | 267700 | PERFORIN 1 (PORE FORMING PROTEIN); PRF1 | 170280 |
| RETINITIS PIGMENTOSA 2, X-LINKED; RP2 | 312600 | RETINITIS PIGMENTOSA GTPASE REGULATOR; RPGR | 312610 |
| RETINOBLASTOMA; RB1 | 180200 | RETINOBLASTOMA 1 (INCLUDING OSTEOSARCOMA); RB1 | 180200 |
| RETINOSCHISIS 1, X-LINKED, JUVENILE; RS1 | 312700 | RETINOSCHISIS (X-LINKED, JUVENILE) 1; RS1 | 312700 |

| DISEASE | | GENE | |
|---|---|---|---|
| NAME | OMIM | NAME | OMIM |
| RETT SYNDROME; RTT | 312750 | METHYL CPG BINDING PROTEIN 2 (RETT SYNDROME); MECP2 | 300005 |
| RIGID SPINE MUSCULAR DYSTROPHY 1; RSMD1 | 602771 | SELENOPROTEIN N, 1; SEPN1 | 606210 |
| ROBINOW SYNDROME, AUTOSOMAL RECESSIVE | 268310 | RECEPTOR TYROSINE KINASE-LIKE ORPHAN RECEPTOR 2; ROR2 | 602337 |
| SAETHRE-CHOTZEN SYNDROME; SCS | 101400 | TWIST HOMOLOG 1 (ACROCEPHALOSYNDACTYLY 3; SAETHRE-CHOTZEN SYNDROME) (DROSOPHILA); TWIST1 | 601622 |
| SEGAWA SYNDROME, AUTOSOMAL RECESSIVE | 605407 | GTP CYCLOHYDROLASE 1 (DOPA-RESPONSIVE DYSTONIA); GCH1 | 600225 |
| | | TYROSINE HYDROXYLASE; TH | 191290 |
| SEVERE COMBINED IMMUNODEFICIENCY, AUTOSOMAL RECESSIVE, T CELL-NEGATIVE, B CELL-NEGATIVE, NK CELL-POSITIVE | 601457 | RECOMBINATION ACTIVATING GENE 1; RAG1 | 179615 |
| | | RECOMBINATION ACTIVATING GENE 2; RAG2 | 179616 |
| SEVERE COMBINED IMMUNODEFICIENCY, AUTOSOMAL RECESSIVE, T CELL-NEGATIVE, B CELL-POSITIVE, NK CELL-NEGATIVE | 600802 | JANUS KINASE 3 (A PROTEIN TYROSINE KINASE, LEUKOCYTE); JAK3 | 600173 |
| SEVERE COMBINED IMMUNODEFICIENCY, X-LINKED; SCIDX1 | 300400 | INTERLEUKIN 2 RECEPTOR, GAMMA (SEVERE COMBINED IMMUNODEFICIENCY); IL2RG | 308380 |
| SEX DETERMINATION | | AMELOGENIN (AMELOGENESIS IMPERFECTA 1, X-LINKED); AMELX | 300391 |
| | | AMELOGENIN, Y-LINKED; AMELY | 410000 |
| | | SEX DETERMINING REGION Y; SRY | 480000 |
| SHPRINTZEN-GOLDBERG CRANIOSYNOSTOSIS SYNDROME; SGS | 182212 | FIBROBLAST GROWTH FACTOR RECEPTOR 2 (BACTERIA-EXPRESSED KINASE, KERATINOCYTE GROWTH FACTOR RECEPTOR, CRANIOFACIAL DYSOSTOSIS 1, CROUZON SYNDROME, PFEIFFER SYNDROME, JACKSON-WEISS SYNDROME); FGFR2 | 176943 |

| DISEASE | | GENE | |
|---|---|---|---|
| NAME | OMIM | NAME | OMIM |
| SHWACHMAN-DIAMOND SYNDROME; SDS | 260400 | SHWACHMAN-BODIAN-DIAMOND SYNDROME; SBDS | 607444 |
| SICKLE CELL ANEMIA | 603903 | HEMOGLOBIN, BETA; HBB | 141900 |
| SILVER-RUSSELL SYNDROME; SRS | 180860 | SILVER-RUSSELL SYNDROME; SRS | 180860 |
| SIMPSON-GOLABI-BEHMEL SYNDROME, TYPE 1; SGBS1 | 312870 | GLYPICAN 3; GPC3 | 300037 |
| SMITH-MAGENIS SYNDROME; SMS | 182290 | RETINOIC ACID INDUCED 1; RAI1 | 607642 |
| SOLUTE CARRIER FAMILY 25 (CARNITINE/ACYLCARNITINE TRANSLOCASE), MEMBER 20; SLC25A20 | 212138 | SOLUTE CARRIER FAMILY 25 (CARNITINE/ACYLCARNITINE TRANSLOCASE), MEMBER 20; SLC25A20 | 212138 |
| SOTOS SYNDROME | 117550 | NUCLEAR RECEPTOR BINDING SET DOMAIN PROTEIN 1; NSD1 | 606681 |
| SPASTIC PARAPLEGIA 4, AUTOSOMAL DOMINANT; SPG4 | 182601 | chromosome 1 open reading frame 2; C1orf2 | 604277 |
| SPINAL AND BULBAR MUSCULAR ATROPHY, X-LINKED 1; SMAX1 | 313200 | ANDROGEN RECEPTOR (DIHYDROTESTOSTERONE RECEPTOR; TESTICULAR FEMINIZATION; SPINAL AND BULBAR MUSCULAR ATROPHY; KENNEDY DISEASE); AR | 313700 |
| SPINAL MUSCULAR ATROPHY WITH RESPIRATORY DISTRESS 1; SMARD1 | 604320 | IMMUNOGLOBULIN MU BINDING PROTEIN 2; IGHMBP2 | 600502 |
| SPINAL MUSCULAR ATROPHY, TYPE I; SMA1 | 253300 | SURVIVAL OF MOTOR NEURON 1, TELOMERIC; SMN1 | 600354 |
| SPINAL MUSCULAR ATROPHY, TYPE II; SMA2 | 253550 | SURVIVAL OF MOTOR NEURON 1, TELOMERIC; SMN1 | 600354 |
| SPINAL MUSCULAR ATROPHY, TYPE III; SMA3 | 253400 | SURVIVAL OF MOTOR NEURON 1, TELOMERIC; SMN1 | 600354 |
| SPINOCEREBELLAR ATAXIA 12; SCA12 | 604326 | protein phosphatase 2 (formerly 2A), regulatory subunit B, beta isoform; PPP2R2B | 604325 |
| SPINOCEREBELLAR ATAXIA 17; SCA17 | 607136 | TATA BOX BINDING PROTEIN; TBP | 600075 |
| SPINOCEREBELLAR ATAXIA 1; SCA1 | 164400 | ATAXIN 1; ATXN1 | 601556 |
| SPINOCEREBELLAR ATAXIA 2; SCA2 | 183090 | ATAXIN 2; ATXN2 | 601517 |
| SPINOCEREBELLAR ATAXIA 6; SCA6 | 183086 | CALCIUM CHANNEL, VOLTAGE-DEPENDENT, P/Q TYPE, ALPHA 1A SUBUNIT; CACNA1A | 601011 |

| DISEASE | | GENE | |
|---|---|---|---|
| NAME | OMIM | NAME | OMIM |
| SPINOCEREBELLAR ATAXIA 7; SCA7 | 164500 | ATAXIN 7; ATXN7 | 607640 |
| SPONDYLOCOSTAL DYSOSTOSIS, AUTOSOMAL RECESSIVE 1; SCDO1 | 277300 | DELTA-LIKE 3 (DROSOPHILA); DLL3 | 602768 |
| STICKLER SYNDROME, TYPE I; STL1 | 108300 | COLLAGEN, TYPE II, ALPHA 1 (PRIMARY OSTEOARTHRITIS, SPONDYLOEPIPHYSEAL DYSPLASIA, CONGENITAL); COL2A1 | 120140 |
| | | COLLAGEN, TYPE XI, ALPHA 1; COL11A1 | 120280 |
| | | COLLAGEN, TYPE XI, ALPHA-2; COL11A2 | 120290 |
| STICKLER SYNDROME, TYPE II; STL2 | 604841 | COLLAGEN, TYPE II, ALPHA 1 (PRIMARY OSTEOARTHRITIS, SPONDYLOEPIPHYSEAL DYSPLASIA, CONGENITAL); COL2A1 | 120140 |
| | | COLLAGEN, TYPE XI, ALPHA 1; COL11A1 | 120280 |
| | | COLLAGEN, TYPE XI, ALPHA-2; COL11A2 | 120290 |
| STICKLER SYNDROME, TYPE III; STL3 | 184840 | COLLAGEN, TYPE II, ALPHA 1 (PRIMARY OSTEOARTHRITIS, SPONDYLOEPIPHYSEAL DYSPLASIA, CONGENITAL); COL2A1 | 120140 |
| | | COLLAGEN, TYPE XI, ALPHA 1; COL11A1 | 120280 |
| | | COLLAGEN, TYPE XI, ALPHA-2; COL11A2 | 120290 |
| STREPTOMYCIN OTOTOXICITY | 580000 | mitochondrially encoded 12S RNA; MT-RNR1 | 561000 |
| SURFACTANT, PULMONARY-ASSOCIATED PROTEIN B; SFTPB | 178640 | SURFACTANT, PULMONARY-ASSOCIATED PROTEIN B; SFTPB | 178640 |
| SYNDACTYLY, TYPE II | 186000 | homeobox D13; HOXD13 | 142989 |
| TELANGIECTASIA, HEREDITARY HEMORRHAGIC, OF RENDU, OSLER, AND WEBER; HHT | 187300 | ACTIVIN A RECEPTOR TYPE II-LIKE 1; ACVRL1 | 601284 |
| | | ENDOGLIN (OSLER-RENDU-WEBER SYNDROME 1); ENG | 131195 |

| DISEASE | | GENE | |
|---|---|---|---|
| NAME | OMIM | NAME | OMIM |
| THALASSEMIAS | 604131 | HEMOGLOBIN, ALPHA 1; HBA1 | 141800 |
| | | HEMOGLOBIN, ALPHA 2; HBA2 | 141850 |
| | | HEMOGLOBIN, BETA; HBB | 141900 |
| | | HEMOGLOBIN, DELTA; HBD | 142000 |
| THANATOPHORIC DYSPLASIA; TD | 187600 | FIBROBLAST GROWTH FACTOR RECEPTOR 3 (ACHONDROPLASIA, THANATOPHORIC DWARFISM); FGFR3 | 134934 |
| THROMBOPHILIA DUE TO DEFICIENCY OF ACTIVATED PROTEIN C COFACTOR | 188055 | coagulation factor V (proaccelerin, labile factor); F5 | 227400 |
| THYROID CARCINOMA, FAMILIAL MEDULLARY; MTC | 155240 | ret proto-oncogene; RET | 164761 |
| TORSION DYSTONIA 1, AUTOSOMAL DOMINANT; DYT1 | 128100 | TORSIN FAMILY 1, MEMBER A (TORSIN A); TOR1A | 605204 |
| TRANSTHYRETIN; TTR | 176300 | TRANSTHYRETIN (PREALBUMIN, AMYLOIDOSIS TYPE I); TTR | 176300 |
| TREACHER COLLINS-FRANCESCHETTI SYNDROME; TCOF | 154500 | Treacher Collins-Franceschetti syndrome 1; TCOF1 | 606847 |
| TUBEROUS SCLEROSIS; TS | 191100 | TUBEROUS SCLEROSIS 1; TSC1 | 605284 |
| | | TUBEROUS SCLEROSIS 2; TSC2 | 191092 |
| TURNERS SYNDROME | | SEX DETERMINING REGION Y; SRY | 480000 |
| TYROSINEMIA, TYPE I | 276700 | FUMARYLACETOACETATE HYDROLASE (FUMARYLACETOACETASE); FAH | 276700 |
| ULLRICH CONGENITAL MUSCULAR DYSTROPHY | 254090 | COLLAGEN, TYPE VI, ALPHA 1; COL6A1 | 120220 |
| | | COLLAGEN, TYPE VI, ALPHA 2; COL6A2 | 120240 |
| | | COLLAGEN, TYPE VI, ALPHA 3; COL6A3 | 120250 |
| UNIPARENTAL DISOMY CHROMOSOME 15 | | UNIPARENTAL DISOMY CHROMOSOME 15 | |
| UNIPARENTAL DISOMY, CHROMOSOME 16 | | UNIPARENTAL DISOMY, CHROMOSOME 16 | |
| UNIPARENTAL DISOMY, PATERNAL, CHROMOSOME 14 | 608149 | UNIPARENTAL DISOMY, PATERNAL, CHROMOSOME 14 | 608149 |
| VAN DER WOUDE SYNDROME; VWS | 119300 | interferon regulatory factor 6; IRF6 | 607199 |

| DISEASE | | GENE | |
|---|---|---|---|
| NAME | OMIM | NAME | OMIM |
| VON HIPPEL-LINDAU SYNDROME; VHL | 193300 | VON HIPPEL-LINDAU SYNDROME; VHL | 193300 |
| WAGR SYNDROME | 194072 | PAIRED BOX 6; PAX6 | 607108 |
| WILMS TUMOR 1; WT1 | 194070 | WILMS TUMOR 1; WT1 | 607102 |
| WILMS TUMOR AND PSEUDOHERMAPHRODITISM | 194080 | WILMS TUMOR 1; WT1 | 607102 |
| WILSON DISEASE | 277900 | ATPase, Cu++ transporting, beta polypeptide; ATP7B | 606882 |
| WISKOTT-ALDRICH SYNDROME; WAS | 301000 | WISKOTT-ALDRICH SYNDROME (ECZEMA-THROMBOCYTOPENIA); WAS | 300392 |
| WITKOP SYNDROME | 189500 | msh homeobox 1; MSX1 | 142983 |
| WOLFRAM SYNDROME | 222300 | WOLFRAM SYNDROME 1 (WOLFRAMIN); WFS1 | 606201 |
| X LINKED DISEASE | | LABORATORY SPECIFIC TEST | |
| Y CHROMOSOME MICRODELETIONS | | LABORATORY SPECIFIC TEST | |
| ZYGOSITY ANALYSIS | | LABORATORY SPECIFIC TEST | |

## Section 2

New tests that have been evaluated through the UKGTN Gene Dossier process, have been accepted by GenCAG and are currently being set up as NHS service

| DISEASE | | GENE | |
|---|---|---|---|
| NAME | OMIM | NAME | OMIM |
| AICARDI-GOUTIERES SYNDROME 1; AGS1 | 225750 | 3-PRIME @REPAIR EXONUCLEASE 1; TREX1 | 606609 |
| AICARDI-GOUTIERES SYNDROME 2; AGS2 | 610181 | RIBONUCLEASE H2, SUBUNIT B; RNASEH2B | 610326 |
| AICARDI-GOUTIERES SYNDROME 3; AGS3 | 610329 | RIBONUCLEASE H2, SUBUNIT C; RNASEH2C | 610330 |
| AICARDI-GOUTIERES SYNDROME 4; AGS4 | 610333 | RIBONUCLEASE H2, SUBUNIT A; RNASEH2A | 606034 |
| AICARDI-GOUTIERES SYNDROME 5; AGS5 | 610905 | 3-PRIME @REPAIR EXONUCLEASE 1; TREX1 | 606609 |
| CEREBRAL CAVERNOUS MALFORMATIONS; CCM | 116860 | KRIT1, ankyrin repeat containing; KRIT1 | 604214 |
| | | cerebral cavernous malformation 2; CCM2 | 603284 |
| | | programmed cell death 10; PDCD10 | 603285 |
| Charcot Marie Tooth disease, demyelinating, type 1D; CMT1D | 607678 | early growth response 2 (Krox-20 homolog, Drosophila); EGR2 | 129010 |
| CHARCOT-MARIE-TOOTH DISEASE, AXONAL, TYPE 2A2; CMT2A2 | 609260 | MITOFUSIN 2; MFN2 | 608507 |
| CHARCOT-MARIE-TOOTH DISEASE, AXONAL, TYPE 2E | 607684 | NEUROFILAMENT, LIGHT POLYPEPTIDE 68KDA; NEFL | 162280 |
| CHARCOT-MARIE-TOOTH DISEASE, AXONAL, TYPE 2K; CMT2K | 607831 | GANGLIOSIDE-INDUCED DIFFERENTIATION-ASSOCIATED PROTEIN 1; GDAP1 | 606598 |
| CHARCOT-MARIE-TOOTH DISEASE, DEMYELINATING, TYPE 1F | 607734 | NEUROFILAMENT, LIGHT POLYPEPTIDE 68KDA; NEFL | 162280 |
| CHARCOT-MARIE-TOOTH DISEASE, TYPE 4A; CMT4A | 214400 | GANGLIOSIDE-INDUCED DIFFERENTIATION-ASSOCIATED PROTEIN 1; GDAP1 | 606598 |
| Charge Syndrome | 214800 | Chromodomain helicase DNA-binding protein-7; CHD7 | 608892 |
| EPISODIC ATAXIA, TYPE 2; EA2 | 108500 | CALCIUM CHANNEL, VOLTAGE-DEPENDENT, P/Q TYPE, ALPHA 1A SUBUNIT; CACNA1A | 601011 |
| Fibrodysplasia Ossificans Progressiva; FOP | 135100 | Activin A, type 1 receptor ; ACVR1 | 102576 |

| DISEASE | | GENE | |
|---|---|---|---|
| NAME | OMIM | NAME | OMIM |
| Hypertrophic Neuropathy of Dejerine Sottas | 145900 | early growth response 2 (Krox-20 homolog, Drosophila); EGR2 | 129010 |
| | | PERIAXIN; PRX | 605725 |
| INFANTILE SPASM SYNDROME, X-LINKED | 308350 | CYCLIN-DEPENDENT KINASE-LIKE 5; CDKL5 | 300203 |
| MIGRAINE, FAMILIAL HEMIPLEGIC, 1; FHM1 | 141500 | CALCIUM CHANNEL, VOLTAGE-DEPENDENT, P/Q TYPE, ALPHA 1A SUBUNIT; CACNA1A | 601011 |
| NEUROPATHY, CONGENITAL HYPOMYELINATING | 605253 | early growth response 2 (Krox-20 homolog, Drosophila); EGR2 | 129010 |
| OSTEOGENESIS IMPERFECTA TYPE 3; OI3 | 259420 | collagen, type I, alpha 1; COL1A1 | 120150 |
| | | collagen, type I, alpha 2; COL1A2 | 120160 |
| RETINITIS PIGMENTOSA; RP | 268000 | retinitis pigmentosa GTPase regulator; RPGR | 312610 |
| | | retinitis pigmentosa 2 (X-linked recessive); RP2 | 312600 |
| | | rhodopsin (opsin 2, rod pigment) (retinitis pigmentosa 4, autosomal dominant); RHO | 180380 |
| | | peripherin 2 (retinal degeneration, slow); PRPH2 | 179605 |
| | | PRP31 pre-mRNA processing factor 31 homolog (S. cerevisiae); PRPF31 | 606419 |
| | | PRP8 pre-mRNA processing factor 8 homolog (S. cerevisiae); PRPF8 | 607300 |
| | | neural retina leucine zipper; NRL | 162080 |
| | | MP (inosine monophosphate) dehydrogenase 1;IMPDH1 | 146690 |
| | | PDGFA associated protein 1; PDAP1 | 607075 |
| | | retinitis pigmentosa 1 (autosomal dominant); RP1 | 603937 |
| SPASTIC PARAPLEGIA 3, AUTOSOMAL DOMINANT; SPG3A | 182600 | SPASTIC PARAPLEGIA 3A (AUTOSOMAL DOMINANT); SPG3A | 606439 |
| Spastic Paraplegia 31, autosomal dominant; SPG31 | 610250 | RECEPTOR ACCESSORY PROTEIN 1; REEP1 | 609139 |

## Section 3

Approved tests that are currently not available from any UKGTN laboratories

| DISEASE | | GENE | |
|---|---|---|---|
| NAME | OMIM | NAME | OMIM |
| JERVELL AND LANGE-NIELSEN SYNDROME; JLNS1 | 220400 | POTASSIUM VOLTAGE-GATED CHANNEL, ISK-RELATED FAMILY, MEMBER 1; KCNE1 | 176261 |
| MICROCEPHALY, PRIMARY AUTOSOMAL RECESSIVE, 1; MCPH1 | 251200 | ASP (ABNORMAL SPINDLE)-LIKE, MICROCEPHALY ASSOCIATED (DROSOPHILA); ASPM | 605481 |
| | | CDK5 REGULATORY SUBUNIT ASSOCIATED PROTEIN 2; CDK5RAP2 | 608201 |
| | | centromere protein J; CENPJ | 609279 |

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