Screening for autosomal recessive and X-linked conditions during pregnancy and preconception: a practice resource of the American College of Medical Genetics and Genomics (ACMG)

Supplemental Material

Methods

Rationale for Tier 3 screening

Figure S1. The relationship between Carrier Frequency and identification of an At-Risk couple.

Technology Considerations

Table S1. Autosomal recessive conditions recommended for screening and scored as definitive in ClinGen, listed as a reportable secondary finding, included in newborn screening programs.

Table S2. X-linked conditions listed in OMIM (November 30, 2020) and initially considered for screening

Methods: Literature search of publically available databases

We performed literature searches using the terms "genetic carrier screening and expanded" after examination of the hierarchy of terms in Medical Subjects Headings (MeSH). We searched PubMed multiple times between September 22, 2020 and December 20, 2020 using the year range 2010 through 2020; 262 results were returned. A second strategy performed over the same time-period incorporated the term "utility" and used the same year filter, which returned 67 results. This was further refined to exclude reviews ("not (reviews)"), which yielded 57 results. The abstracts of these search results were reviewed. Snowball sampling of the

articles' reference list identified additional relevant articles that fell outside the date range of 2010 through 2020.

Several publicly available databases were used to inform questions 5 and 6. These are shown in **Box S1**. Group consensus informed all ACMG recommendations.

Box S1. Publicly Available Data Bases Utilized	
Name	**URL**
gnomAD v 2.1.1	https://gnomad.broadinstitute.org/
OMIM	https://omim.org/
Orphanet	https://www.orpha.net/consor/cgi-bin/index.php
MedlinePlus	https://medlineplus.gov/
ClinGen	https://www.clinicalgenome.org/

Rationale for ≥ 1/200 autosomal recessive genes included in Tier 3 screening

The rationale for selecting ≥1/200 rests on two studies. One study utilized a diagnostic laboratory's database,[1] the other utilized gnomAD version 2.0.2 a large-scale dataset of unrelated individuals.[2,3] This version of gnomAD consists of 123,136 exome sequencing samples. Variant analysis, within this version, is stratified for seven populations: African/African American, Ashkenazi Jewish, East Asian, Finnish, Hispanic/Admixed American, Non-Finnish European, and South Asian. In this report Finnish were excluded since they represent a very small portion of the US population and a theoretical US population was constructed based on census data. gnomAD allowed investigators to view pathogenic and likely pathogenic allele frequencies within 415 autosomal recessive genes (referenced in ClinVar) by ancestry. Both studies demonstrated a log curve relationship with carrier frequency or total number of screened genes on the X-axis and identified at-risk couples or carriers on the Y-axis (**Figure S1**). At-risk couples (both partners are carrier of a pathogenic or likely pathogenic variant within the same gene) were more common within populations where endogamy was more likely (e.g., Ashkenazi Jewish). When moving from Tier 2 (≥1/100 carrier frequency) to Tier 3 (1/200 carrier frequency) or from point X to Y (**Figure S1**), there were an additional 9/10,000 at-risk couples identified. At a carrier frequency of 1/100 there were 241 per 10,000 at-risk AJ couples identified and this increased to 250/10,000 at 1/200 carrier frequency. This represents a 4% increase in at-risk couples. Additional at-risk couples identified in this interval ranged from 4-9 per 10,000 depending in the endogamous population examined. When the population evaluated was weighted by US census data, at-risk couples identified increased by six per 10,000 couples (45 to 51 per 10,000) when moving from the Tier 2 (≥1/100) carrier frequency to that of Tier 3 (≥1/200). Assuming ~4 million births per year, this translates to an annual increase of 2,400

additional US couples that will have the opportunity to make reproductive decisions following a positive carrier screening result if Tier 3 autosomal recessive conditions are screened rather than Tier 2.

Importantly, for carrier frequency of less than 1/200 the added number of at-risk couples gets diminishingly small (Z-Y in **Figure S1**). In populations where endogamy is common, modeling[2] suggested that screening for conditions with a carrier frequency of 1/1000 would identify only two additional couples per 10,000 couples screened (0.02%) or 252 vs. 250 couples per 10,000). The range of additional at-risk couples identified across the six populations evaluated was 2-5 per 10,000.[2]

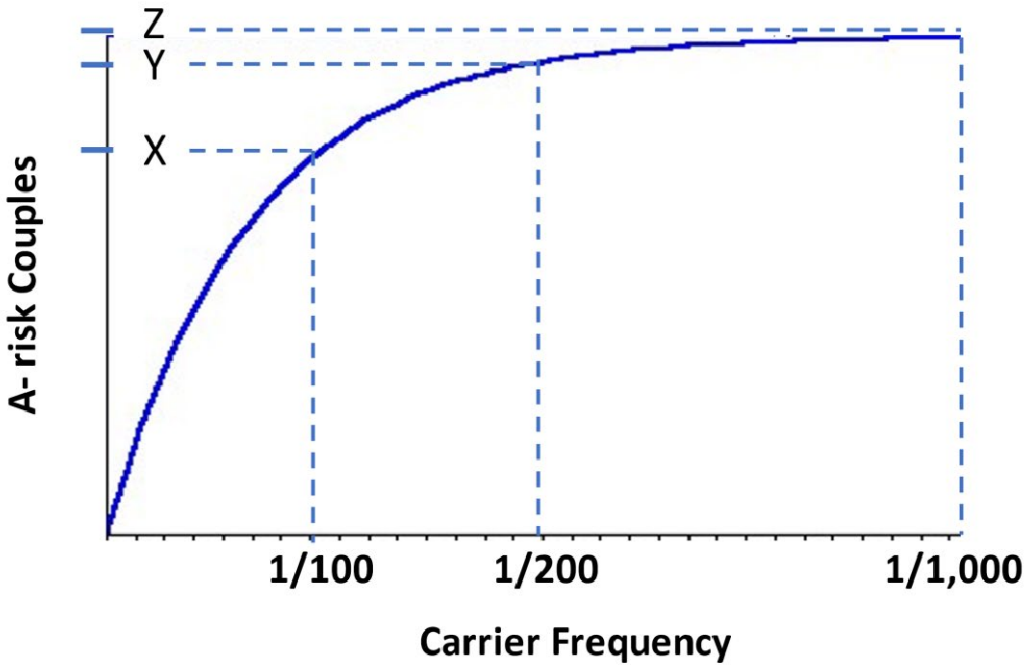

Figure S1. The relationship between Carrier Frequency and identification of an At-Risk couple. The gain in identification of at-risk couples (X-Y) is greatest when moving between conditions with higher frequency (1/100 to 1/200). As we move from higher to lower carrier

frequency (1/200 to 1/1000) the gain in identification of at-risk couples (Y-Z) gets diminishingly small.

Rationale for 1/40,000 disease prevalence of X-linked genes included in Tier 3 screening

It seems logical to apply the risk analysis used for screening autosomal recessive conditions. A 1/200 carrier frequency (Tier 3) results in a 1/160,000 risk of an affected fetus (1/200 carrier frequency threshold x 1/200 carrier frequency threshold x 1/4 risk of an affected fetus = 1/160,000). To reach 1/160,000 for an X-linked condition, the carrier frequency of 1/40,000 is required (1/40,000 X-linked condition carrier frequency x 1/2 chance of inheriting the variant X chromosome x 1/2 chance of inheriting Y = 1/160,000). However, this approach relies on accurate carrier frequency data for the X-linked conditions considered and this is precisely what confounds this approach. Currently there is no gnomAD peer-reviewed study with a comprehensive assessment of variant frequencies in X-linked genes across populations.

Among X-linked genes, variants are often *de novo* and may be high as 25% for some X-linked conditions. In other words, population prevalence for any condition is a function of heritable cases plus *de novo* variants. We chose to include conditions with a disease prevalence of at least 1/40,000 because it approaches the calculated frequency of at-risk couples for autosomal recessive conditions. The conditions we recommend have a prevalence that ranges from 1/3,500 to 1/40,000. With nearly 4 million births each year in the US, a condition with a prevalence as high as of 1/3,500 is expected to result in more than 500 affected XY patients each year; for conditions with a prevalence of 1/40,000, 50 affected XY patients will be born each year. We anticipate that screening for these X-linked conditions has the potential to impact at least 1,000 US families annually.

Technology Considerations

This consensus group recognizes that not all sequence variants and structural rearrangements leading to clinical pathology can be detected using high throughout low-cost laboratory methods. It is beyond the scope of this document to consider the laboratory methods required to make accurate determinations that can reliably classify patients as carriers. The ACMG Laboratory Quality Assurance Committee is assessing the genes proposed for carrier screening to identify appropriate laboratory methods that will result in the greatest sensitivity and specificity while preserving the need for high throughput and low cost.

We also recognize that it will be necessary to reevaluate the genes proposed for screening in this document as there is a continuous growth in accumulation, assessment, and interpretation of the data in human genetic variation. Databases cataloging human sequence accrue new samples leading to a more diverse and representative population composition. Advances in sequencing technology and bioinformatics enlarge the scope of assessed genetic material and improve the number and the type of variants identified. The ClinVar database grows with new submissions and the refinement of variant interpretation is an ongoing process. New genetic etiologies in human disease are being discovered. To address this dynamic nature of available information, a working group of the ACMG Board of Directors is proposed to provide continuing curation of the genes recommended for screening. As ClinGen curates more genes we may find other examples where curation identifies limited gene-disease association as we did for *BCS1L*. Most importantly, new information will be garnered as laboratories use this list of genes for screening across the United States. Using a standardized list of genes that considers many ancestral groups and is built around a transparent process for including and excluding genes will improve our attention to distributive justice. We believe this process recognizes and begins to

address the disparities of genetics and genomics in delivering better health to diverse populations.

Table S1. Autosomal recessive conditions recommended for screening and scored as definitive in ClinGen, listed as a reportable secondary finding, included in newborn screening programs.

Carrier Frequency (Table 1-5) see text	≥1/50 N=19	<1/50 to ≥1/100 N=19	<1/100 to ≥1/150 N=25	< 1/150 to ≥1/200 N=23	≥1/200 del/dup or ≥1/200 US Sub-Population (see text) N=11
ClinGen*	Definitive = 13 genes (68%)	Definitive = 5 genes (26%)	Definitive = 8 genes (32%)	Definitive = 9 genes (39%)	Definitive = 4 genes (36%)
ACMG SF V3.0	*ATP7B*	*GAA*		*BTD*	
Newborn Screening	**N=7 phenotypes** *HBB, CYP21A2, PAH, CFTR, GJB2 (deafness), ACADM, SLC26A4 (deafness)*	**N=3 phenotypes** *GAA, BCKDHB, MMUT*	**N=2 phenotypes** *GALT, FAH,*	**N=6 phenotypes** *ACADVL, ASL, BTD, MCCC2 CBS, IDUA,*	

ACMG, American College of Medical Genetics and Genomics; SF, secondary findings

*sum does not equal N for each carrier frequency because some genes have been curated in ClinGen but there is no statement regarding gene-disease association. One gene demonstrated limited association and was removed from the Tier 3 panel of genes and one gene was not identified in ClinGen.

Table S2: X-linked conditions listed in OMIM (November 30, 2020) and initially considered for screening

OMIM Search - 'prefix:# AND chromosome:X' Downloaded: Nov 30, 2020 Copyright (c) 1966-2020 Johns Hopkins University OMIM, data are provided for research purposes only.			
MIM Number	**Title**[a]	**Included Titles**	**Cytogenetic Location**
#127300	LERI-WEILL DYSCHONDROSTEOSIS; LWD	MADELUNG DEFORMITY, INCLUDED	Xp22.33, Yp11.2
#300009	DENT DISEASE 1		Xp11.23
#300018	46,XY SEX REVERSAL 2; SRXY2		Xp21.2
#300029	RETINITIS PIGMENTOSA 3; RP3		Xp11.4
#300048	INTESTINAL PSEUDOOBSTRUCTION, NEURONAL, CHRONIC IDIOPATHIC, X-LINKED	CONGENITAL SHORT BOWEL SYNDROME, X-LINKED, INCLUDED - FLNA	Xq28
#300049	PERIVENTRICULAR NODULAR HETEROTOPIA 1; PVNH1	HETEROTOPIA, PERIVENTRICULAR NODULAR, WITH FRONTOMETAPHYSEAL DYSPLASIA, INCLUDED	Xq28
#300055	MENTAL RETARDATION, X-LINKED, SYNDROMIC 13; MRXS13		Xq28
#300066	DEAFNESS, X-LINKED 4; DFNX4		Xp22.12
#300067	LISSENCEPHALY, X-LINKED, 1; LISX1	SUBCORTICAL LAMINAR HETEROTOPIA, X-LINKED, INCLUDED; SCLH, INCLUDED	Xq23
#300068	ANDROGEN INSENSITIVITY SYNDROME; AIS		Xq12
#300071	NIGHT BLINDNESS, CONGENITAL STATIONARY, TYPE 2A; CSNB2A		Xp11.23
#300087	X INACTIVATION, FAMILIAL SKEWED, 1; SXI1		Xq13.2
#300088	DEVELOPMENTAL AND EPILEPTIC ENCEPHALOPATHY 9; DEE9 - PCDH19 gene		Xq22.1
#168600	PARKINSON DISEASE, LATE-ONSET; PD		1q22, 4q23, 6q27, 12q24.12, 13q21.33, 17q21.31, Xq24
#176807	PROSTATE CANCER		7p22.3, 10p15.2, 10q23.31, 10q25.2, 13q13.1, 16q22.1, 16q22.2-q22.3, 22q12.1, Xq12
#194070	WILMS TUMOR 1; WT1		11p13, 13q13.1,

			Xq26.2 (somatic)
#300100	ADRENOLEUKODYSTROPHY; ALD	ADRENOMYELONEUROPATHY, INCLUDED; AMN, INCLUDED	Xq28
#300106	SPONDYLOEPIMETAPHYSEAL DYSPLASIA, X-LINKED; SEMDX - BGN gene		Xq28
#300114	RAYNAUD-CLAES SYNDROME; MRXSRC		Xp22.2
#300123	MENTAL RETARDATION, X-LINKED, WITH PANHYPOPITUITARISM	MENTAL RETARDATION, X-LINKED, WITH ISOLATED GROWTH HORMONE DEFICIENCY, INCLUDED; MRGH, INCLUDED	Xq27.1
#300143	MENTAL RETARDATION, X-LINKED 21; MRX21 - IL1RAPL1 gene		Xp21.3-p21.2
#300148	MEHMO SYNDROME; MEHMO - EIF2S3 gene		Xp22.11
#300166	MICROPHTHALMIA, SYNDROMIC 2; MCOPS2		Xp11.4
#300194	AMME COMPLEX		Xq22.3
#300200	ADRENAL HYPOPLASIA, CONGENITAL; AHC - NR0B1		Xp21.2
#300209	SIMPSON-GOLABI-BEHMEL SYNDROME, TYPE 2; SGBS2		Xp22.2
#300210	MENTAL RETARDATION, X-LINKED 58; MRX58 TSPAN7 gene		Xp11.4
#300215	LISSENCEPHALY, X-LINKED, 2; LISX2	HYDRANENCEPHALY AND ABNORMAL GENITALIA, INCLUDED - - ARX gene	Xp21.3
#300219	MYOTUBULAR MYOPATHY WITH ABNORMAL GENITAL DEVELOPMENT - - likely MTM1 contig gene deletion		
#300232	SPONDYLOEPIMETAPHYSEAL DYSPLASIA, X-LINKED, WITH HYPOMYELINATING LEUKODYSTROPHY; SEMDHL; AIFM1 gene		Xq26.1
#300238	MENTAL RETARDATION, X-LINKED, SYNDROMIC 11; MRXS11		Xq26.3
#300243	MENTAL RETARDATION, X-LINKED, SYNDROMIC, CHRISTIANSON TYPE; MRXSCH; SLC9A6 gene		Xq26.3
#300244	TERMINAL OSSEOUS DYSPLASIA; TOD		Xq28
#300257	DANON DISEASE		Xq24
#300260	LUBS X-LINKED MENTAL RETARDATION SYNDROME; MRXSL		Xq28
#300261	INTELLECTUAL DEVELOPMENTAL DISORDER, X-LINKED, SYNDROMIC, ARMFIELD TYPE; MRXSA		Xq28
#300263	SIDERIUS X-LINKED MENTAL RETARDATION SYNDROME; MRXSSD		Xp11.22

#300271	MENTAL RETARDATION, X-LINKED 72; MRX72; RAB39B gene		Xq28
#300280	URUGUAY FACIOCARDIOMUSCULOSKELETAL SYNDROME; FCMSU; FHL1 gene		Xq26.3
#300291	ECTODERMAL DYSPLASIA AND IMMUNODEFICIENCY 1; EDAID1		Xq28
#300299	NEUTROPENIA, SEVERE CONGENITAL, X-LINKED; SCNX; allelic with Wiskott Aldrich		Xp11.23
#300310	IMMUNODEFICIENCY 61; IMD61; SH3KBP1 gene; 2 brothers reported		Xp22.12
#300321	FG SYNDROME 2; FGS2; FLNA gene		Xq28
#300322	LESCH-NYHAN SYNDROME; LNS	HPRT DEFICIENCY, NEUROLOGIC VARIANT, INCLUDED	Xq26.2-q26.3
#300323	HYPERURICEMIA, HPRT-RELATED; HRH		Xq26.2-q26.3
#300337	HYPOMELANOSIS OF ITO; HMI; Incontinentia pigmenti Type 1 (not classic type); mosaic translocation		
#300352	CEREBRAL CREATINE DEFICIENCY SYNDROME 1; CCDS1		Xq28
#300354	MENTAL RETARDATION, X-LINKED, SYNDROMIC, CABEZAS TYPE; MRXSC		Xq24
#300367	THROMBOCYTOPENIA, X-LINKED, WITH OR WITHOUT DYSERYTHROPOIETIC ANEMIA; XLTDA; GATA1 gene		Xp11.23
#300373	OSTEOPATHIA STRIATA WITH CRANIAL SCLEROSIS; OSCS; WTX aka AMER1 gene		Xq11.2
#300376	MUSCULAR DYSTROPHY, BECKER TYPE; BMD		Xp21.2-p21.1
#300387	MENTAL RETARDATION, X-LINKED 63; MRX63; ACSL4 gene		Xq23
#300400	SEVERE COMBINED IMMUNODEFICIENCY, X-LINKED; SCIDX1		Xq13.1
#300419	MENTAL RETARDATION, X-LINKED, WITH OR WITHOUT SEIZURES, ARX-RELATED; MRXARX		Xp21.3
#300422	FG SYNDROME 4; FGS4	MENTAL RETARDATION, X-LINKED, WITH OR WITHOUT NYSTAGMUS, INCLUDED; FG4 in OMIM; CASK gene	Xp11.4
#300423	MENTAL RETARDATION, X-LINKED, SYNDROMIC, HEDERA TYPE; MRXSH; ATP6AP2 gene		Xp11.4
#300424	RETINITIS PIGMENTOSA 23; RP23; OFD1 gene		Xp22.2
#300425	AUTISM, SUSCEPTIBILITY TO, X-LINKED 1; AUTSX1; NLGN3 gene		Xq13.1

#300434	STOCCO DOS SANTOS X-LINKED MENTAL RETARDATION SYNDROME; SDSX; SHROOM4 gene		Xp11.22
#300438	HSD10 MITOCHONDRIAL DISEASE; HSD10MD		Xp11.22
#300448	ALPHA-THALASSEMIA MYELODYSPLASIA SYNDROME; ATMDS		Xq21.1
#300455	RETINITIS PIGMENTOSA, X-LINKED, AND SINORESPIRATORY INFECTIONS, WITH OR WITHOUT DEAFNESS		Xp11.4
#300472	CORPUS CALLOSUM, AGENESIS OF, WITH MENTAL RETARDATION, OCULAR COLOBOMA, AND MICROGNATHIA		Xq13.1
#300475	DEAFNESS, DYSTONIA, AND CEREBRAL HYPOMYELINATION; DDCH	CONTIGUOUS ABCD1/DXS1375E DELETION SYNDROME, INCLUDED; CADDS, INCLUDED	Xq28
#300476	CONE-ROD DYSTROPHY, X-LINKED, 3; CORDX3		Xp11.23
#300486	MENTAL RETARDATION, X-LINKED, WITH CEREBELLAR HYPOPLASIA AND DISTINCTIVE FACIAL APPEARANCE		Xq12
#300489	SPINAL MUSCULAR ATROPHY, DISTAL, X-LINKED 3; SMAX3		Xq21.1
#300491	EPILEPSY, X-LINKED, WITH VARIABLE LEARNING DISABILITIES AND BEHAVIOR DISORDERS		Xp11.3-p11.2
#300494	ASPERGER SYNDROME, X-LINKED, SUSCEPTIBILITY TO, 1; ASPGX1		Xq13.1
#300495	AUTISM, SUSCEPTIBILITY TO, X-LINKED 2; AUTSX2	MENTAL RETARDATION, X-LINKED, INCLUDED	Xp22.32-p22.31
#300496	AUTISM, SUSCEPTIBILITY TO, X-LINKED 3; AUTSX3		Xq28
#300497	ASPERGER SYNDROME, X-LINKED, SUSCEPTIBILITY TO, 2; ASPGX2		Xp22.32-p22.31
#300500	ALBINISM, OCULAR, TYPE I; OA1		Xp22.2
#300510	OVARIAN DYSGENESIS 2; ODG2	PREMATURE OVARIAN FAILURE 4, INCLUDED; POF4, INCLUDED	Xp11.22
#300511	PREMATURE OVARIAN FAILURE 2A; POF2A		Xq21.33
#300514	FANCONI ANEMIA, COMPLEMENTATION GROUP B; FANCB		Xp22.2
#300523	ALLAN-HERNDON-DUDLEY SYNDROME; AHDS		Xq13.2
#300534	MENTAL RETARDATION, X-LINKED, SYNDROMIC, CLAES-JENSEN TYPE; MRXSCJ		Xp11.22

#300539	NEPHROGENIC SYNDROME OF INAPPROPRIATE ANTIDIURESIS; NSIAD		Xq28
#300554	HYPOPHOSPHATEMIC RICKETS, X-LINKED RECESSIVE		Xp11.23
#300555	DENT DISEASE 2		Xq26.1
#300558	MENTAL RETARDATION, X-LINKED 30; MRX30		Xq23
#300559	GLYCOGEN STORAGE DISEASE, TYPE IXd; GSD9D		Xq13.1
#300578	CHROMOSOME Xp11.3 DELETION SYNDROME		Xp11.3
#300582	SHORT STATURE, IDIOPATHIC, X-LINKED; ISS		Xp22.33, Yp11.2
#300590	CORNELIA DE LANGE SYNDROME 2; CDLS2		Xp11.22
#300600	ALAND ISLAND EYE DISEASE; AIED		Xp11.23
#300604	PREMATURE OVARIAN FAILURE 2B; POF2B		Xq21.1
#300607	DEVELOPMENTAL AND EPILEPTIC ENCEPHALOPATHY 8; DEE8		Xq11.1
#300614	DEAFNESS, X-LINKED 5, WITH PERIPHERAL NEUROPATHY; DFNX5		Xq26.1
#300615	BRUNNER SYNDROME; BRNRS	ANTISOCIAL BEHAVIOR, SUSCEPTIBILITY TO, INCLUDED	Xp11.3
#300622	TN POLYAGGLUTINATION SYNDROME; TNPS		Xq24
#300623	FRAGILE X TREMOR/ATAXIA SYNDROME; FXTAS		Xq27.3
#300624	FRAGILE X SYNDROME; FXS		Xq27.3
#300633	HYPOSPADIAS 1, X-LINKED; HYSP1		Xq12
#300635	LYMPHOPROLIFERATIVE SYNDROME, X-LINKED, 2; XLP2		Xq25
#300636	IMMUNODEFICIENCY 33; IMD33		Xq28
#300643	ROLANDIC EPILEPSY, MENTAL RETARDATION, AND SPEECH DYSPRAXIA, X-LINKED; RESDX		Xq22.1
#300645	IMMUNODEFICIENCY 34; IMD34		Xp21.1-p11.4
#300653	PHOSPHOGLYCERATE KINASE 1 DEFICIENCY		Xq21.1
#300659	MENTAL RETARDATION, X-LINKED 93; MRX93		Xq21.1
#300661	PHOSPHORIBOSYLPYROPHOSPHATE SYNTHETASE SUPERACTIVITY	GOUT, PRPS-RELATED, INCLUDED	Xq22.3
#300672	DEVELOPMENTAL AND EPILEPTIC ENCEPHALOPATHY 2; DEE2		Xp22.13
#300673	ENCEPHALOPATHY, NEONATAL SEVERE, DUE TO MECP2 MUTATIONS		Xq28
#300676	MENTAL RETARDATION, X-LINKED, SYNDROMIC 14; MRXS14		Xq24
#300679	CHROMOSOME Xp21 DELETION SYNDROME		Xp21
#300695	SCAPULOPERONEAL MYOPATHY, X-LINKED DOMINANT; SPM		Xq26.3

#300696	MYOPATHY, X-LINKED, WITH POSTURAL MUSCLE ATROPHY; XMPMA	EMERY-DREIFUSS MUSCULAR DYSTROPHY 6, X-LINKED, INCLUDED; EDMD6, INCLUDED	Xq26.3
#300699	INTELLECTUAL DEVELOPMENTAL DISORDER, X-LINKED, SYNDROMIC, WU TYPE; MRXSW		Xq25
#300705	CHROMOSOME Xp11.22 DUPLICATION SYNDROME		Xp11.22
#300707	TOE SYNDACTYLY, TELECANTHUS, AND ANOGENITAL AND RENAL MALFORMATIONS; STAR		Xq28
#300717	REDUCING BODY MYOPATHY, X-LINKED 1A, SEVERE, WITH INFANTILE OR EARLY CHILDHOOD ONSET; RBMX1A		Xq26.3
#300718	REDUCING BODY MYOPATHY, X-LINKED 1B, WITH LATE CHILDHOOD OR ADULT ONSET; RBMX1B		Xq26.3
#300749	MENTAL RETARDATION AND MICROCEPHALY WITH PONTINE AND CEREBELLAR HYPOPLASIA; MICPCH		Xp11.4
#300751	ANEMIA, SIDEROBLASTIC, 1; SIDBA1		Xp11.21
#300752	PROTOPORPHYRIA, ERYTHROPOIETIC, X-LINKED; XLEPP		Xp11.21
#300755	AGAMMAGLOBULINEMIA, X-LINKED; XLA	HYPOGAMMAGLOBULINEMIA, X-LINKED, INCLUDED	Xq22.1
#300758	HYPOSPADIAS 2, X-LINKED; HYSP2		Xq28
#300770	SURFACTANT METABOLISM DYSFUNCTION, PULMONARY, 4; SMDP4		Xp22.33
#300799	INTELLECTUAL DEVELOPMENTAL DISORDER, X-LINKED, SYNDROMIC, RAYMOND TYPE; MRXSR		Xq26.1
#300801	CHROMOSOME Xp11.23-p11.22 DUPLICATION SYNDROME		Xp11.23-p11.22
#300802	MENTAL RETARDATION, X-LINKED 96; MRX96		Xp11.23
#300803	MENTAL RETARDATION, X-LINKED 97; MRX97		Xq21.1
#300804	JOUBERT SYNDROME 10; JBTS10		Xp22.2
#300807	THROMBOPHILIA, X-LINKED, DUE TO FACTOR IX DEFECT; THPH8	DEEP VENOUS THROMBOSIS, PROTECTION AGAINST, INCLUDED	Xq27.1
#300814	NYSTAGMUS 6, CONGENITAL, X-LINKED; NYS6		Xp22.2
#300815	CHROMOSOME Xq28 DUPLICATION SYNDROME		Xq28
#300816	COMBINED OXIDATIVE PHOSPHORYLATION DEFICIENCY 6; COXPD6		Xq26.1
#300818	PAROXYSMAL NOCTURNAL HEMOGLOBINURIA 1; PNH1		Xp22.2
#300830	AUTISM, SUSCEPTIBILITY TO, X-LINKED 4; AUTSX4		Xp22.11
#300831	CK SYNDROME		Xq28

#300833	46,XX SEX REVERSAL 3; SRXX3	CHROMOSOME Xq26 DELETION SYNDROME, INCLUDED	Xq26.3
#300834	MACULAR DEGENERATION, X-LINKED ATROPHIC		Xp11.4
#300835	ANEMIA, X-LINKED, WITH OR WITHOUT NEUTROPENIA AND/OR PLATELET ABNORMALITIES; XLANP		Xp11.23
#300842	MCLEOD SYNDROME; MCLDS	MCLEOD SYNDROME WITH CHRONIC GRANULOMATOUS DISEASE, INCLUDED	Xp21.1
#300844	MENTAL RETARDATION, X-LINKED 19; MRX19		Xp22.12
#300845	MOYAMOYA DISEASE 4 WITH SHORT STATURE, HYPERGONADOTROPIC HYPOGONADISM, AND FACIAL DYSMORPHISM; MYMY4		Xq28
#300847	AUTISM, SUSCEPTIBILITY TO, X-LINKED 5; AUTSX5		Xq28
#300849	MENTAL RETARDATION, X-LINKED 41; MRX41		Xq28
#300850	MENTAL RETARDATION, X-LINKED 90; MRX90		Xq13.1
#300853	IMMUNODEFICIENCY, X-LINKED, WITH MAGNESIUM DEFECT, EPSTEIN-BARR VIRUS INFECTION, AND NEOPLASIA; XMEN		Xq21.1
#300854	RENAL CELL CARCINOMA, Xp11-ASSOCIATED; RCCX1		Xp11.23
#300855	OGDEN SYNDROME; OGDNS		Xq28
#300857	AMYOTROPHIC LATERAL SCLEROSIS 15 WITH OR WITHOUT FRONTOTEMPORAL DEMENTIA; ALS15		Xp11.21
#300860	MENTAL RETARDATION, X-LINKED, SYNDROMIC, NASCIMENTO TYPE; MRXSN		Xq24
#300863	CHONDRODYSPLASIA WITH PLATYSPONDYLY, DISTINCTIVE BRACHYDACTYLY, HYDROCEPHALY, AND MICROPHTHALMIA		Xp11.23
#300867	KABUKI SYNDROME 2; KABUK2		Xp11.3
#300868	MULTIPLE CONGENITAL ANOMALIES-HYPOTONIA-SEIZURES SYNDROME 2; MCAHS2		Xp22.2
#300869	CHROMOSOME Xq27.3-q28 DUPLICATION SYNDROME		Xq27.3-q28
#300872	AUTISM, SUSCEPTIBILITY TO, X-LINKED 6; AUTSX6		Xq28
#300882	CORNELIA DE LANGE SYNDROME 5; CDLS5		Xq13.1
#300884	DEVELOPMENTAL AND EPILEPTIC ENCEPHALOPATHY 36; DEE36	CONGENITAL DISORDER OF GLYCOSYLATION, TYPE Is, INCLUDED; CDG1S, INCLUDED	Xq23

#300886	MENTAL RETARDATION, X-LINKED, SYNDROMIC 32; MRXS32		Xq28
#300887	LINEAR SKIN DEFECTS WITH MULTIPLE CONGENITAL ANOMALIES 2; LSDMCA2		Xq21.1
#300888	HYPOTHYROIDISM, CENTRAL, WITH TESTICULAR ENLARGEMENT; CHTE		Xq26.1
#300894	NEURODEGENERATION WITH BRAIN IRON ACCUMULATION 5; NBIA5		Xp11.23
#300895	OHDO SYNDROME, X-LINKED; OHDOX		Xq13.1
#300896	CONGENITAL DISORDER OF GLYCOSYLATION, TYPE IIm; CDG2M		Xp11.23
#300905	CHARCOT-MARIE-TOOTH DISEASE, X-LINKED DOMINANT, 6; CMTX6		Xp22.11
#300908	ANEMIA, NONSPHEROCYTIC HEMOLYTIC, DUE TO G6PD DEFICIENCY		Xq28
#300909	ANGIOEDEMA INDUCED BY ACE INHIBITORS, SUSCEPTIBILITY TO; AEACEI		Xq26.1
#300910	BONE MINERAL DENSITY QUANTITATIVE TRAIT LOCUS 18; BMND18		Xq23
#300911	PARKINSONISM WITH SPASTICITY, X-LINKED; XPDS		Xp11.4
#300912	MENTAL RETARDATION, X-LINKED 98; MRX98		Xq13.3
#300914	DEAFNESS, X-LINKED 6; DFNX6		Xq22.3
#300915	MICROPHTHALMIA, SYNDROMIC 13; MCOPS13		Xq28
#300918	PALMOPLANTAR KERATODERMA, MUTILATING, WITH PERIORIFICIAL KERATOTIC PLAQUES, X-LINKED		Xp22.12
#300919	MENTAL RETARDATION, X-LINKED 99; MRX99		Xp11.4
#300923	MENTAL RETARDATION, X-LINKED 100; MRX100		Xq13.1
#300928	MENTAL RETARDATION, X-LINKED 101; MRX101		Xq22.3
#300932	THYROXINE-BINDING GLOBULIN QUANTITATIVE TRAIT LOCUS; TBGQTL		Xq22.3
#300934	CONGENITAL DISORDER OF GLYCOSYLATION, TYPE Iy; CDG1Y		Xq28
#300942	CHROMOSOME Xq26.3 DUPLICATION SYNDROME		Xq26.3
#300943	PITUITARY ADENOMA 2, GROWTH HORMONE-SECRETING; PITA2		Xq26.3
#300946	DIAMOND-BLACKFAN ANEMIA 14 WITH MANDIBULOFACIAL DYSOSTOSIS; DBA14		Xp11.22
#300952	LINEAR SKIN DEFECTS WITH MULTIPLE CONGENITAL ANOMALIES 3; LSDMCA3		Xp11.3

#300953	TRICHOTHIODYSTROPHY 5, NONPHOTOSENSITIVE; TTD5		Xq24
#300957	MENTAL RETARDATION, X-LINKED 12; MRX12		Xq25
#300958	INTELLECTUAL DEVELOPMENTAL DISORDER, X-LINKED, SYNDROMIC, SNIJDERS BLOK TYPE; MRXSSB		Xp11.4
#300960	MEND SYNDROME; MEND		Xp11.23
#300963	RITSCHER-SCHINZEL SYNDROME 2; RTSC2		Xp11.23
#300966	MENTAL RETARDATION, X-LINKED, SYNDROMIC 33; MRXS33		Xq13.1
#300967	MENTAL RETARDATION, X-LINKED, SYNDROMIC 34; MRXS34		Xq13.1
#300968	MENTAL RETARDATION, X-LINKED 99, SYNDROMIC, FEMALE-RESTRICTED; MRXS99F		Xp11.4
#300971	BARTTER SYNDROME, TYPE 5, ANTENATAL, TRANSIENT; BARTS5		Xp11.21
#300972	IMMUNODEFICIENCY 47; IMD47		Xq28
#300978	TONNE-KALSCHEUER SYNDROME; TOKAS		Xq13.2
#300979	Xq25 DUPLICATION SYNDROME	Xq25 TRIPLICATION SYNDROME, INCLUDED	Xq25
#300982	MENTAL RETARDATION, X-LINKED 103; MRX103		Xp22.11
#300983	MENTAL RETARDATION, X-LINKED 104; MRX104		Xp22.2
#300984	MENTAL RETARDATION, X-LINKED 105; MRX105		Xp11.23
#300985	VAS DEFERENS, CONGENITAL BILATERAL APLASIA OF, X-LINKED; CBAVDX		Xp22.13
#300986	MENTAL RETARDATION, X-LINKED, SYNDROMIC, BAIN TYPE; MRXSB		Xq22.1
#300988	IMMUNODEFICIENCY 50; IMD50		Xq12
#300989	MEESTER-LOEYS SYNDROME; MRLS		Xq28
#300990	MIDFACE HYPOPLASIA, HEARING IMPAIRMENT, ELLIPTOCYTOSIS, AND NEPHROCALCINOSIS; MFHIEN		Xq23
#300991	CILIARY DYSKINESIA, PRIMARY, 36, X-LINKED; CILD36		Xq22.3
#300997	MENTAL RETARDATION, X-LINKED 106; MRX106		Xq13.1
#300998	MENTAL RETARDATION, X-LINKED, SYNDROMIC, 35; MRXS35		Xq28
#301000	WISKOTT-ALDRICH SYNDROME; WAS		Xp11.23
#301006	GALLOWAY-MOWAT SYNDROME 2, X-LINKED; GAMOS2		Xq28
#301008	MENTAL RETARDATION, X-LINKED, SYNDROMIC, HOUGE TYPE; MRXSHG		Xp22.12
#301010	MYOPIA 26, X-LINKED, FEMALE-RESTRICTED; MYP26		Xq13.1

#301013	MENTAL RETARDATION, X-LINKED 107; MRX107		Xq24
#301014	OSTEOGENESIS IMPERFECTA, TYPE XIX; OI19		Xp22.12
#301015	HEMOLYTIC ANEMIA, CONGENITAL, X-LINKED		Xq27.1
#301018	DEAFNESS, X-LINKED 7; DFNX7		Xq22.1
#301020	MITOCHONDRIAL COMPLEX I DEFICIENCY, NUCLEAR TYPE 12; MC1DN12		Xq24
#301021	MITOCHONDRIAL COMPLEX I DEFICIENCY, NUCLEAR TYPE 30; MC1DN30		Xp11.3
#301022	MULLEGAMA-KLEIN-MARTINEZ SYNDROME; MKMS		Xq25
#301024	INTELLECTUAL DEVELOPMENTAL DISORDER, X-LINKED 108; MRX108		Xp11.3
#301025	PAGANINI-MIOZZO SYNDROME; MRXSPM		Xq26.2
#301026	KEIPERT SYNDROME; KPTS		Xq26.2
#301028	NEPHROTIC SYNDROME, TYPE 20; NPHS20		Xq22.3
#301029	SHUKLA-VERNON SYNDROME; SHUVER		Xq26.1
#301030	VAN ESCH-O'DRISCOLL SYNDROME; VEODS		Xp22.1-p21.3
#301031	CONGENITAL DISORDER OF GLYCOSYLATION, TYPE Icc; CDG1CC		Xq21.1
#301032	BASILICATA-AKHTAR SYNDROME; MRXSBA		Xp22.2
#301033	HYPOTHYROIDISM, CONGENITAL, NONGOITROUS, 8; CHNG8		Xp22.3-p22.2
#301035	HYPOTHYROIDISM, CONGENITAL, NONGOITROUS, 9; CHNG9		Xq22.3
#301039	INTELLECTUAL DEVELOPMENTAL DISORDER, X-LINKED, SYNDROMIC, HACKMANN-DI DONATO TYPE; MRXSHD		Xq24
#301040	ALPHA-THALASSEMIA/MENTAL RETARDATION SYNDROME, X-LINKED; ATRX		Xq21.1
#301041	WIEACKER-WOLFF SYNDROME, FEMALE-RESTRICTED; WRWFFR		Xq11.2
#301043	HOLOPROSENCEPHALY 13, X-LINKED; HPE13		Xq25
#301044	EPILEPTIC ENCEPHALOPATHY, EARLY INFANTILE, 85, WITH OR WITHOUT MIDLINE BRAIN DEFECTS; EIEE85		Xp11.22
#301045	CONGENITAL DISORDER OF GLYCOSYLATION, TYPE IIr; CDG2R		Xp11.4
#301050	ALPORT SYNDROME 1, X-LINKED; ATS1		Xq22.3
#301051	IMMUNODEFICIENCY 74, COVID19-RELATED, X-LINKED; IMD74		Xp22.2

#301052	WARFARIN SENSITIVITY, X-LINKED		Xq27.1
#301054	VEXAS SYNDROME; VEXAS		Xp11.3
#301200	AMELOGENESIS IMPERFECTA, TYPE IE; AI1E		Xp22.2
#301220	PIGMENTARY DISORDER, RETICULATE, WITH SYSTEMIC MANIFESTATIONS, X-LINKED; PDR		Xp22.1-p21.3
#301310	ANEMIA, SIDEROBLASTIC, AND SPINOCEREBELLAR ATAXIA; ASAT		Xq13.3
#301500	FABRY DISEASE	FABRY DISEASE, CARDIAC VARIANT, INCLUDED	Xq22.1
#301830	SPINAL MUSCULAR ATROPHY, X-LINKED 2; SMAX2		Xp11.3
#301835	ARTS SYNDROME; ARTS		Xq22.3
#301900	BORJESON-FORSSMAN-LEHMANN SYNDROME; BFLS		Xq26.2
#302045	CARDIOMYOPATHY, DILATED, 3B; CMD3B		Xp21.2-p21.1
#302060	BARTH SYNDROME; BTHS		Xq28
#302200	CATARACT 40; CTRCT40		Xp22.2-p22.1
#302350	NANCE-HORAN SYNDROME; NHS		Xp22.2-p22.1
#302500	SPINOCEREBELLAR ATAXIA, X-LINKED 1; SCAX1		Xq28
#302800	CHARCOT-MARIE-TOOTH DISEASE, X-LINKED DOMINANT, 1; CMTX1		Xq13.1
#302802	CHARCOT-MARIE-TOOTH DISEASE, X-LINKED RECESSIVE, 3; CMTX3		Xq26
#302905	ABRUZZO-ERICKSON SYNDROME; ABERS		Xq21.1
#302950	CHONDRODYSPLASIA PUNCTATA 1, X-LINKED RECESSIVE; CDPX1		Xp22.33
#302960	CHONDRODYSPLASIA PUNCTATA 2, X-LINKED DOMINANT; CDPX2		Xp11.23
#303100	CHOROIDEREMIA; CHM	CHOROIDAL SCLEROSIS, INCLUDED	Xq21.2
#303110	CHOROIDEREMIA, DEAFNESS, AND MENTAL RETARDATION		Xq21
#303350	MASA SYNDROME		Xq28
#303400	CLEFT PALATE WITH OR WITHOUT ANKYLOGLOSSIA, X-LINKED; CPX		Xq21.1
#303600	COFFIN-LOWRY SYNDROME; CLS		Xp22.12
#303700	BLUE CONE MONOCHROMACY; BCM	CONE DYSTROPHY 5, X-LINKED, INCLUDED; COD5, INCLUDED	Xq28, Xq28
#303800	COLORBLINDNESS, PARTIAL, DEUTAN SERIES; CBD	DEUTERANOMALY, INCLUDED	Xq28
#303900	COLORBLINDNESS, PARTIAL, PROTAN SERIES; CBP	PROTANOMALY, INCLUDED	Xq28
#304020	CONE-ROD DYSTROPHY, X-LINKED, 1; CORDX1	CONE DYSTROPHY 1, X-LINKED, INCLUDED; COD1, INCLUDED	Xp11.4
#304100	CORPUS CALLOSUM, PARTIAL AGENESIS OF, X-LINKED		Xq28
#304110	CRANIOFRONTONASAL SYNDROME; CFNS		Xq13.1

#304120	OTOPALATODIGITAL SYNDROME, TYPE II; OPD2		Xq28
#304150	OCCIPITAL HORN SYNDROME; OHS		Xq21.1
#304340	PETTIGREW SYNDROME; PGS		Xp22.2
#304400	DEAFNESS, X-LINKED 2; DFNX2		Xq21.1
#304500	DEAFNESS, X-LINKED 1; DFNX1		Xq22.3
#304700	MOHR-TRANEBJAERG SYNDROME; MTS		Xq22.1
#304790	IMMUNODYSREGULATION, POLYENDOCRINOPATHY, AND ENTEROPATHY, X-LINKED; IPEX	ISLETS OF LANGERHANS, ABSENCE OF, INCLUDED	Xp11.23
#304800	DIABETES INSIPIDUS, NEPHROGENIC, X-LINKED		Xq28
#305000	DYSKERATOSIS CONGENITA, X-LINKED; DKCX	HOYERAAL-HREIDARSSON SYNDROME, INCLUDED; HHS, INCLUDED	Xq28
#305100	ECTODERMAL DYSPLASIA 1, HYPOHIDROTIC, X-LINKED; XHED		Xq13.1
#305390	EXUDATIVE VITREORETINOPATHY 2, X-LINKED; EVR2		Xp11.3
#305400	AARSKOG-SCOTT SYNDROME; AAS	FACIOGENITAL DYSPLASIA WITH ATTENTION DEFICIT-HYPERACTIVITY DISORDER, INCLUDED	Xp11.22
#305450	OPITZ-KAVEGGIA SYNDROME; OKS		Xq13.1
#305600	FOCAL DERMAL HYPOPLASIA; FDH		Xp11.23
#305620	FRONTOMETAPHYSEAL DYSPLASIA 1; FMD1		Xq28
#306000	GLYCOGEN STORAGE DISEASE IXa1; GSD9A1	GLYCOGEN STORAGE DISEASE IXa2, INCLUDED; GSD9A2, INCLUDED	Xp22.13
#306400	GRANULOMATOUS DISEASE, CHRONIC, X-LINKED; CGDX	CYTOCHROME b-POSITIVE GRANULOMATOUS DISEASE, CHRONIC, X-LINKED, INCLUDED	Xp21.1-p11.4
#306700	HEMOPHILIA A; HEMA		Xq28
#306900	HEMOPHILIA B; HEMB	HEMOPHILIA B(M), INCLUDED	Xq27.1
#306955	HETEROTAXY, VISCERAL, 1, X-LINKED; HTX1	CONGENITAL HEART DEFECTS, MULTIPLE TYPES, 1, X-LINKED, INCLUDED; CHTD1, INCLUDED	Xq26.3
#307000	HYDROCEPHALUS DUE TO CONGENITAL STENOSIS OF AQUEDUCT OF SYLVIUS; HSAS	HYDROCEPHALUS, X-LINKED, WITH CONGENITAL IDIOPATHIC INTESTINAL PSEUDOOBSTRUCTION, INCLUDED	Xq28
#307030	GLYCEROL KINASE DEFICIENCY; GKD		Xp21.2
#307150	HYPERTRICHOSIS, CONGENITAL GENERALIZED; HTC2		Xq27.1
#307200	ISOLATED GROWTH HORMONE DEFICIENCY, TYPE III, WITH AGAMMAGLOBULINEMIA; IGHD3		Xq22.1
#307700	HYPOPARATHYROIDISM, X-LINKED; HYPX		Xq27.1

#307800	HYPOPHOSPHATEMIC RICKETS, X-LINKED DOMINANT; XLHR		Xp22.11
#308050	CONGENITAL HEMIDYSPLASIA WITH ICHTHYOSIFORM ERYTHRODERMA AND LIMB DEFECTS		Xq28
#308100	ICHTHYOSIS, X-LINKED; XLI	ICHTHYOSIS, X-LINKED, COMPLICATED, INCLUDED	Xp22.31
#308205	IFAP SYNDROME 1, WITH OR WITHOUT BRESHECK SYNDROME; IFAP1		Xp22.12
#308230	IMMUNODEFICIENCY WITH HYPER-IgM, TYPE 1; HIGM1		Xq26.3
#308240	LYMPHOPROLIFERATIVE SYNDROME, X-LINKED, 1; XLP1		Xq25
#308300	INCONTINENTIA PIGMENTI; IP		Xq28
#308350	DEVELOPMENTAL AND EPILEPTIC ENCEPHALOPATHY 1; DEE1		Xp21.3
#308700	HYPOGONADOTROPIC HYPOGONADISM 1 WITH OR WITHOUT ANOSMIA; HH1		Xp22.31
#308800	KERATOSIS FOLLICULARIS SPINULOSA DECALVANS, X-LINKED; KFSDX		Xp22.12
#308940	LEIOMYOMATOSIS, DIFFUSE, WITH ALPORT SYNDROME; DL-ATS		
#308990	PROTEINURIA, LOW MOLECULAR WEIGHT, WITH HYPERCALCIURIA AND NEPHROCALCINOSIS		Xp11.23
#309000	LOWE OCULOCEREBRORENAL SYNDROME; OCRL		Xq26.1
#309120	SPERMATOGENIC FAILURE, X-LINKED, 2; SPGFX2		Xq13.1
#309300	MEGALOCORNEA; MGC1		Xq23
#309350	MELNICK-NEEDLES SYNDROME; MNS		Xq28
#309400	MENKES DISEASE; MNK		Xq21.1
#309500	RENPENNING SYNDROME 1; RENS1		Xp11.23
#309510	PARTINGTON X-LINKED MENTAL RETARDATION SYNDROME; PRTS		Xp21.3
#309520	INTELLECTUAL DEVELOPMENTAL DISORDER, X-LINKED, SYNDROMIC, LUJAN-FRYNS TYPE; MRXSLF		Xq13.1
#309530	MENTAL RETARDATION, X-LINKED 1; MRX1		Xp11.22
#309541	METHYLMALONIC ACIDEMIA AND HOMOCYSTEINEMIA, cblX TYPE		Xq28
#309548	MENTAL RETARDATION, X-LINKED, ASSOCIATED WITH FRAGILE SITE FRAXE		Xq28
#309549	MENTAL RETARDATION, X-LINKED 9; MRX9		Xp11.23
#309580	MENTAL RETARDATION-HYPOTONIC FACIES SYNDROME, X-LINKED, 1; MRXHF1		Xq21.1

#309583	MENTAL RETARDATION, X-LINKED, SYNDROMIC, SNYDER-ROBINSON TYPE; MRXSSR		Xp22.11
#309585	WILSON-TURNER X-LINKED MENTAL RETARDATION SYNDROME; WTS		Xq12
#309590	MENTAL RETARDATION, X-LINKED, SYNDROMIC, TURNER TYPE; MRXST		Xp11.22
#309630	METACARPAL 4-5 FUSION; MF4		Xq21.1
#309800	MICROPHTHALMIA, SYNDROMIC 1; MCOPS1		Xq28
#309801	LINEAR SKIN DEFECTS WITH MULTIPLE CONGENITAL ANOMALIES 1; LSDMCA1		Xp22.2
#309900	MUCOPOLYSACCHARIDOSIS, TYPE II; MPS2		Xq28
#310200	MUSCULAR DYSTROPHY, DUCHENNE TYPE; DMD		Xp21.2-p21.1
#310300	EMERY-DREIFUSS MUSCULAR DYSTROPHY 1, X-LINKED; EDMD1		Xq28
#310400	MYOPATHY, CENTRONUCLEAR, X-LINKED; CNMX		Xq28
#310440	MYOPATHY, X-LINKED, WITH EXCESSIVE AUTOPHAGY; MEAX		Xq28
#310468	NEPHROLITHIASIS, X-LINKED RECESSIVE, WITH RENAL FAILURE; XRN		Xp11.23
#310490	CHARCOT-MARIE-TOOTH DISEASE, X-LINKED RECESSIVE, 4, WITH OR WITHOUT CEREBELLAR ATAXIA; CMTX4		Xq26.1
#310500	NIGHT BLINDNESS, CONGENITAL STATIONARY, TYPE 1A; CSNB1A	NYCTALOPIA, INCLUDED	Xp11.4
#310600	NORRIE DISEASE; ND		Xp11.3
#310700	NYSTAGMUS 1, CONGENITAL, X-LINKED; NYS1	NYSTAGMUS, INFANTILE PERIODIC ALTERNATING, X-LINKED, INCLUDED; XIPAN, INCLUDED	Xq26.2
#311070	CHARCOT-MARIE-TOOTH DISEASE, X-LINKED RECESSIVE, 5; CMTX5		Xq22.3
#311200	OROFACIODIGITAL SYNDROME I; OFD1		Xp22.2
#311250	ORNITHINE TRANSCARBAMYLASE DEFICIENCY, HYPERAMMONEMIA DUE TO		Xp11.4
#311300	OTOPALATODIGITAL SYNDROME, TYPE I; OPD1	OTOPALATODIGITAL SPECTRUM DISORDER, INCLUDED	Xq28
#311360	PREMATURE OVARIAN FAILURE 1; POF1		Xq27.3
#311510	WAISMAN SYNDROME; WSMN		Xq28
#311900	TARP SYNDROME; TARPS		Xp11.3
#312000	PANHYPOPITUITARISM, X-LINKED; PHPX		Xq27.1
#312060	PROPERDIN DEFICIENCY, X-LINKED; CFPD	PROPERDIN DEFICIENCY, TYPE II, INCLUDED	Xp11.23

#312080	PELIZAEUS-MERZBACHER DISEASE; PMD		Xq22.2
#312170	PYRUVATE DEHYDROGENASE E1-ALPHA DEFICIENCY; PDHAD	LACTIC ACIDEMIA, THIAMINE-RESPONSIVE, INCLUDED	Xp22.12
#312300	ANDROGEN INSENSITIVITY, PARTIAL; PAIS		Xq12
#312600	RETINITIS PIGMENTOSA 2; RP2		Xp11.3
#312700	RETINOSCHISIS 1, X-LINKED, JUVENILE; RS1		Xp22.13
#312750	RETT SYNDROME; RTT	RETT SYNDROME, ZAPPELLA VARIANT, INCLUDED	Xq28
#312863	COMBINED IMMUNODEFICIENCY, X-LINKED; CIDX		Xq13.1
#312870	SIMPSON-GOLABI-BEHMEL SYNDROME, TYPE 1; SGBS1		Xq26.2
#312920	SPASTIC PARAPLEGIA 2, X-LINKED; SPG2		Xq22.2
#313200	SPINAL AND BULBAR MUSCULAR ATROPHY, X-LINKED 1; SMAX1		Xq12
#313400	SPONDYLOEPIPHYSEAL DYSPLASIA TARDA, X-LINKED; SEDT		Xp22.2
#313500	TOOTH AGENESIS, SELECTIVE, X-LINKED, 1; STHAGX1		Xq13.1
#313900	THROMBOCYTOPENIA 1; THC1	THROMBOCYTOPENIA, X-LINKED, INTERMITTENT, INCLUDED	Xp11.23
#314050	THROMBOCYTOPENIA WITH BETA-THALASSEMIA, X-LINKED; XLTT		Xp11.23
#314250	DYSTONIA 3, TORSION, X-LINKED; DYT3		Xq13.1
#314390	VACTERL ASSOCIATION, X-LINKED, WITH OR WITHOUT HYDROCEPHALUS; VACTERLX		Xq26.3
#314400	CARDIAC VALVULAR DYSPLASIA, X-LINKED; CVD1		Xq28
#314580	WIEACKER-WOLFF SYNDROME; WRWF		Xq11.2
#249700	LANGER MESOMELIC DYSPLASIA; LMD		Xp22.33, Yp11.2
#300000	OPITZ GBBB SYNDROME, TYPE I; GBBB1		Xp22.2
#300004	CORPUS CALLOSUM, AGENESIS OF, WITH ABNORMAL GENITALIA		Xp21.3
#611162	MALARIA, SUSCEPTIBILITY TO	MALARIA, RESISTANCE TO, INCLUDED	1q23.2, 1q23.3, 1q23.3, 1q32.2, 2q14.3, 3p21.2, 4q31.21, 4q31.21, 6p21.33, 7q21.11, 11p15.4, 11q24.2,

			17q11.2, 17q21.31, 19p13.2, Xq28
MIM Number	Title	Included Titles	Cytogenetic Location
#127300	LERI-WEILL DYSCHONDROSTEOSIS; LWD	MADELUNG DEFORMITY, INCLUDED	Xp22.33, Yp11.2
#300009	DENT DISEASE 1		Xp11.23
#300018	46,XY SEX REVERSAL 2; SRXY2		Xp21.2
#300029	RETINITIS PIGMENTOSA 3; RP3		Xp11.4
#300048	INTESTINAL PSEUDOOBSTRUCTION, NEURONAL, CHRONIC IDIOPATHIC, X-LINKED	CONGENITAL SHORT BOWEL SYNDROME, X-LINKED, INCLUDED - FLNA	Xq28

[a]Titles as listed verbatim in OMIM.

References:

1. Ben-Shachar, R., Svenson, A., Goldberg, J. D. & Muzzey, D. A data-driven evaluation of the size and content of expanded carrier screening panels. *Genet. Med.* **21**, 1931-1939 (2019).
2. Guo, M. H. & Gregg, A. R. Estimating yields of prenatal carrier screening and implications for design of expanded carrier screening panels. *Genet. Med.* **21**, 1940-1947 (2019).
3. Lek, M. et al. Analysis of protein-coding genetic variation in 60,706 humans. *Nature*. **536**, 285-291 (2016).