GENEVA Aneuploidy Reference Table: Version 9-4-2009

Chromosome Abnormalities Reference: http://www.ncbi.nlm.nih.gov/omim/

	Aneuploidy	Disease	Chromosome(s)	Gene map position	Gene(s)	Characteristics	Population Rate	Gender Prevalence
	Tetrasomy	Trisomy 22/Cat eye syndrome	22	22q11.2	multiple	coloboma of the iris and anal atresia with fistula, downslanting palpebral fissures, preauricular tags and/or pits, frequent occurrence of heart and renal malformations, and normal or near-normal mental development. An extra small marker chromosome is present representing tetrasomy (or occasionally trisomy) of 22q11.2.	between 1:50,000 and 1:150,000 based on patients observed in Northeastern Switzerland	affects females and males equally
		Pallister-Killian	12	12p	multiple	Tetrasomy 12p secondary to an extra isochromosome of 12 short arm is associated with Pallister-Killian syndrome. This syndrome is an example of tissue limited mosaicism. The isochromosome 12p is typically found in fibroblasts, but not in the lymphocytes. (See Molecular Cytogenetics update) Phenotype includes profound MR, seizures, streaky pigmentation, sparse hair (especially in temporal areas), coarse facial features in older patients.		
	Trisomies	Warkany syndrome 2	8			moderate to severe mental retardation, retarded psychomoto development, abnormally short/tall stature, expressionless face		
		Mosaic Trisomy 8	8			Although full trisomy 8 is often lethal, many patients have been reported with mosaic trisomy 8. The phenotype includes a variable degree of MR (mild to moderate) with a tendency to poor coordination, prominent forehead, deep set eyes, strabismus, hypertelorism, cleft palate, joint contractures and other skeletal abnormalities, deep creases on palms and soles.		
		Trisomy 9	9			dysmorphisms in the skull, nervous system, and mental retardation, small face, wide fontanelle, prominent occiput, low set ears, webbed neck, rocker bottom feet		
		Trisomy 9 Mosaicism	9			Phenotype includes severe MR, and growth deficiency, sloping forehead, deeply set eyes, joint contractures, and heart defects.		
		Patau Syndrome	13			These patients typically demonstrate growth retardation, macrocephaly, sloped forehead, scalp defects, cleft lip/palate, micro or anopthalmia, post axial polydactyly, heart defects, renal anomalies, severe CNS malformation and MR. microencephaly, low-set ears, eye defects (microphthalmia), kidney defects, prominent heel, rocker-bottom feet	1 in 3000 live births	appears to affect females more than males
		Trisomy 16	16			intrauterine growth retardation, congenital heart defects	occurs in more than 1% of pregnancies	
		Edwards Syndrome/Trisomy 18	18			Patients typically demonstrate MR, failure to thrive, heart malformations, hypertonia, clenched fists, "rocker-bottom feet" with prominent heel bone, prominent occiput, micrognathia, low-set malformed ears, short palpebral fissures, kidney malformations, intestines protruding outside the body, and growth deficiency	1 in 3,000 to 1 in 8,000	Females 3x more likely to be affected than males
		Down Syndrome	21			These patients have MR, hypotonia, short stature, brachycephaly with a flat occiput, short neck, flat nasal bridge, low-set abnormally folded ears, open mouth with protruding tongue, upslanting palpebral fissures and epicanthal folds, oblique eye fissures, white spots on iris, congenital heart defects, broad head, round face	1 in 650 to 1,000 live births	Male > Female (1.3:1)
	Duplications		3	3q26.3		A small number of patients have been observed with rearrangements in this band and phenotypic overlap with Cornelia de Lange syndrome. These patients have characteristic facies (low anterior hairline, synophrys, anteverted nares, maxillary prognathism, long philtrum, 'carp' mouth), prenatal and postnatal growth retardation, mental retardation and, in many cases, upper limb anomalies.		
			7	7q11.23q11.23		Patients with microduplications complementary to the microdeletions associated with Williams syndrome demonstrate mild to moderate MR, significant speech delay, and behaviors associated with autism spectrum disorder. No strongly reproducible physical features have been identified.		
			8	8p23.1p23.1		A number of similar appearing duplications have been reported in the literature and existing data suggests that they may fall into one of two cytogenetically indistinguishable categories. The first category of duplications is thought to represent benign variants. The molecular basis for this duplication variant appears to be an extreme of normal copy number variation involving the beta-defensin gene. Recent data suggests that the second category of patients may carry a clinically significant duplication involving a different group of genes. This second group of patients often has developmental delays and/or heart defects.		
		Prader-Willi/ Angelman duplication	15	15q11-q13	multiple	Duplications if this region in the maternally inherited 15 are associated with autism and mental retardation. These duplications involve molecular duplications of one or more of the probes that have been demonstrated to be deleted in PSW/AS. A cytogenetically indistinguishable benign variant duplication also exists. The molecular basis for this benign 15q11.2 duplication variant is the presence of multiple copies of a gene cluster that maps proximal to the Prader-Willi/Angelman region		
		CMT1A (Charcot-Marie-Tooth)	17	17p12	PMP22	(Generally submicroscopic) duplications of this region are associated with Charcot-Marie Tooth disease (CMTIA), a peripheral neuropathy associated with muscle weakness.		

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	Potocki-Lupski syndrome	17	17p11.2	RAI1	Several patients with Potocki-Lupski syndrome have now been reported with visible duplication of the same region deleted in Smith-Magenis syndrome. These patients have developmental delay without distinctive physical features		
	22q11.2 microduplication	22	22q11.2	multiple	This small interstitial duplication involves the same region deleted in DiGeorge syndrome. Duplications range from 3-4 Mb to 6 Mb. Some phenotypic overlap with the DiGeorge deletion (cleft palate/insufficiency, thymic aplasia). Facial features include superior placement of eyebrows, widely spaced eyes, downslanting palpebral fissures, mild micro-/retrognathia, minor ear anomalies. May have hearing loss, malformations of the urogenital tract, poor growth, and cognitive deficits.		
	TAR syndrome	1	1q21.1		A 200kb microdeletion in 1q21.1 has been demonstrated in 30/30 patients with TAR syndrome. TAR is thrombocytopenia-absent radius syndrome and is characterized by thrombocytopenia and bilateral radial aplasia, without absence of thumbs. Some patients also have heart defects and/or cow's milk intolerance. Only 25% of these deletions were de novo, the remaining were inherited from a phenotypically normal carrier parent. This raises the question of reduced penetrance for this deletion phenotype. A contiguous gene deletion syndrome		
	Monosomy 1p36	1	1p36	multiple	Several patients have been reported with very small deletions in distal 1p. Although the phenotype is variable, some of these patients have significant phenotypic overlap with Prader-Willi syndrome.	estimated the frequency to be 1 in 5,000 births.	
	Nephronophthisis 1	2	2q13	NPHP1	Clinical features of familial juvenile nephronophthisis include anemia, polyuria, polydipsia, isosthenuria, and death in uremia. Autosomal recessive inheritance has been observed.		
	Albright hereditary osteodystrophy	2	2q37.3		Several patients have been described with small distal deletions of 2q and Albright hereditary osteodystrophy.		
	3q29 microdeletion	3	3q29	multiple	The clinical phenotype was variable despite an almost identical deletion size. The phenotype included mild to moderate mental retardation, with only slightly dysmorphic facial features that were similar in most patients: long and narrow face, short philtrum, and high nasal bridge. Autism, gait ataxia, chest wall deformity, and long and tapering fingers were noted in at least 2 of the 6 patients. Additional features, including microcephaly, cleft lip and palate, horseshoe kidney and hypospadias, ligamentous laxity, recurrent middle ear infections, and abnormal pigmentation, were observed, each in a single patient.		
	Wolf-Hirschhorn Syndrome	4	4p16.3	multiple	A classic chromosome deletion syndrome. Wolf-Hirschhorn syndrome. Phenotype includes severe growth and mental retardation, microcephaly, "Greek Warrior helmet" face, cleft lip and/or palate, seizures, poor muscle tone, strabismus, severe growth. Although the critical region for the syndrome is in 4p16.3, deletions can be large or small. A milder phenotype associated with similar deletions is seen in patients with Pitt-Rogers-Danks syndrome.	1 in 50,000 births	2:1 female/male ratio
	Cri du chat/Chromosome 5q deletion syndrome	5	5p15.2	multiple	A classic chromosome deletion syndrome. Cri-du-chat syndrome named for the characteristic high pitched cry in infants with 5p deletions that include the p15.31 and/or distal p15.2 region. Phenotype includes microcephaly, round face, hypertelorism, micrognathia, epicanthal folds, low-set ears, hypotonia, severe psychomotor and mental retardation.	1 in 20,000 to 50,000 newborns	slightly more common in females
	Sotos	5	5q35	NSD1	A classic chromosome deletion syndrome. The phenotype includes overgrowth, macrocephaly, mental retardation, hypotonia and poor coordination, advanced bone age and a typical facies (prominent forehead with recessed hairline, long narrow face, pointed chin, hypertelorism and large ears).	Deletions are more common in the Japanese population (~ 50 of patients) than the non-Japanese (~10% of patients).	
	Greig cephalopolysyndactyly	7	7p13	GLI3	polysyndactyly with peculiar skull shape		
	Saethre-Chotzen	7	7p21	TWIST1	mild acrocephaly, asymmetry of the skull, and partial soft tissue syndactyly of fingers 2 and 3 and toes 3 and 4, asymmetry of the skull and orbits (plagiocephaly), strabismus, and a thin, long, pointed nose, bifid terminal phalanges of digits 2 and 3 and absence of the first metatarsal. Cleft palate, hydrophthalmos, cardiac malformation, and contractures of elbows and knees were present in some.		
	Williams-Beuren syndrome	7	7q11.23	ELN	supravalvular aortic stenosis, elfin face, mental and statural deficiency, dental malformation, infantile hypercalcemia, attention deficit disorder, musical talent, overfriendliness. Patients with often have remarkable musical and verbal abilities. This is a submicroscopic deletion detectable by FISH. Genes within the deleted region thought to contribute to the phenotype include elastin (SVAS, aortic stenosis); LIM Kinase 1 (cognitive impairment); RFC 2 gene (reduced efficiency of DNA replication - growth and developmental delay).	1 in 7500 live births	affects both sexes equally
	Holoprosencephaly 3	7	7q36.3	SHH	distal deletions in 7q the location of the sonic hedgehog gene		
	CHARGE	8	8q12.2	CHD7	coloboma of the eye; heart anomaly; atresia, choanal; retardation of mental and somatic development; microphallus; ear abnormalities and/or deafness; Facial palsy, cleft palate, and dysphagia are commonly associated.	estimated birth incidence of 1 in 12,000	

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Autosomal	Deletions	TRPS1 (Trichorhinophalangeal syndrome, Type 1)	8	8q23.3-q24.1	TRPS1	a malformation syndrome characterized by distinctive craniofacial and skeletal abnormalities and is inherited as an autosomal dominant ; patients have sparse scalp hair, bulbous tip of the nose, long flat philtrum, thin upper vermilion border, and protruding ears. Skeletal abnormalities include cone-shaped epiphyses at the phalanges, hip malformations, and short stature.		
		Langer-Giedion	8	8q23.3-q24.1, 8q24.11-24.13	EXT1, TRPS1	This is probably a true contiguous syndrome with symptoms caused by disturbances of both the TRPS1 gene and EXT1. Phenotype includes mental retardation, microcephaly, multiple exostoses, redundant skin, and sparse hair.		
			9	9p24		Partial monosomy 9p is associated with male to female sex reversal suggesting the presence of a gene important for normal male development. Patients with 9p deletions also have MR, motor development delay, trigonocephaly, wide nasal bridge, long fingers and toes, etc.		
		DiGeorge 2	10	10p13-p14	multiple	A small number of patients with the DiGeorge phenotype have been reported with deletions of 10p. May present many clinical problems, including cardiac defects, hypoparathyroidism, T-cell immunodeficiency, and facial dysmorphism.		
		Beckwith-Wiedemann	11	11p15.5	IGF2 p57(KIP2) (CDKN1C; 600856), H19 (103280), and LIT1 (604115).	Caused by mutation or deletion of imprinted genes within the chromosome 11p15.5 region. Patients with duplications in this region of 11p have been reported with Beckwith-Wiedemann syndrome. The primary features associated with this syndrome include macroglossia (enlarged tongue), omphalocele (protrusion of abdominal contents through umbilicus, macrosomia (gigantism) and ear creases.	Thorburn et al. (1970) described 6 cases in Jamaican blacks and estimated an incidence of 1 in 13,700 births.	
		Potocki-Shaffer	11	11p11.2	EXT2, ALX4	patients typically have multiple exostoses, an enlarged parietal foramina, craniofacial dysostosis and MR.		
		WAGR (Wilms tumor, anirida, genitourinary, mental retardation syndrome)	11	11p13	PAX6, WT1	a contiguous gene syndrome due to deletion, either microscopic or submicroscopic, at chromosome 11p13 in a region containing the WT1 (607102) and PAX6 (607108) genes.		
		Jacobsen syndrome	11	11q23	multiple	growth retardation, psychomotor retardation, trigonocephaly, divergent intermittent strabismus, epicanthus, telecanthus, broad nasal bridge, short nose with anteverted nostrils, carp-shaped upper lip, retrognathia, low-set dysmorphic ears, bilateral camptodactyly, hammertoes, and isoimmune thrombocytopenia. This deletion may be caused by an inherited fragile site at 11q23.3 in some patients.	1 in 100,000 live births	Female/male ratio = 2:1
			13	13q14.2		Deletions of 13q that overlap this region are associated with a high risk of developing retinoblastoma (tumor of the retina).		
		Angelman Syndrome	15	Xq28, approx 70% of cases result from de novo maternal deletions in 15q11.2-q13 critical region	UBE3A	Small deletions on proximal 15q(maternal homologue) in this region are associated with Angelman syndrome. Evidence from molecular studies suggests that AS is caused by mutations or deletions of UBE3A (the E6-associated protein ubiquitin-protein ligase gene) in this region. This gene is imprinted and because only a single gene has been implicated, AS is not considered a contiguous gene syndrome. Approximately 60 - 70% of AS patients have deletions detectable by high resolution banding or FISH. UPD pat is the cause of ~2 - 3% of AS and ~6% have imprinting mutations. The rest may be caused by mutations in the UBE3A gene. The phenotype includes severe motor and mental retardation, ataxia, hypotonia, epilepsy, absence of speech, and large mandible, frequent laughter/smiling, apparent happy demeanor.	1 in 12,000 to 20,000 people	affects both sexes equally
		Prader-Willi syndrome	15	15q12, 15q11-q13	SNRPN	diminished fetal activity, obesity, muscular hypotonia, mental retardation, short stature, hypogonadotropic hypogonadism, small hands and feet. Deletions of the paternally inherited chromosome 15 that overlap 15q11.2. Between 70 - 80% of PWS is caused by deletions (most but not all are cytogenetically visible by high resolution analysis). 20% of PWS is due to UPD mat, which there is no paternal copy of 15. 2 - 4% of patients have a small deletion in the imprinting center (IC) resulting in an imprinting defect. PWS is a true contiguous gene syndrome. Genes within the critical region that contribute to the phenotype are SNRPN (small nuclear ribonucleoprotein polypeptide N) believed to have its effect on the development of the nervous system, and necdin, another gene important in brain development. Abnormal behaviors include scratching and skin picking. Individuals with deletion-type PWS are often hypopigmented as compared to relatives.	1:10,000 to 30,000 births	affects both sexes equally
			16	16p11p11.2		A submicroscopic deletion of approximately 600 kb has been found to be associated with a predisposition to autism. Several patients with the complementary duplication and autism have also been reported		
		Alpha-Thalassemia/Mental Retardation	16	16p13.3	HBA1, HBA2	Patients with small deletions in 16p have been observed with alpha thalassemia, MR, and a broad spectrum of associated anomalies.		
		Rubinstein-Taybi	16	16p13.3	CREBBP	Phenotype includes broad thumbs and great toes, mental retardation, small mouth, beaked nose, micrognathia, low hair line, heart defects, skeletal abnormalities.		

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	Neurofibromatosis 1	17	17q11.2	NF1	Neurofibromatosis is an autosomal dominant disorder characterized particularly by cafe-au-lait spots and fibromatous tumors of the skin. Other features are variably present		
	Smith-Magenis syndrome	17	17p11.2	RAI1	Phenotype includes brachycephaly, midface hypoplasia, prognathism, hoarse voice, MR, growth retardation, behavior problems (including self-destructive behavior and sleep disturbances). hypotonia, speech delay, small ears, conductive hearing loss, esotropia. More distally placed deletions within the 17p11.2 band are associated with HNPP	1 in 25,000 births maybe closer to 1/15,000 (identified worldwide in all ethnic groups)	
	HNPP (Hereditary neuropathy with liability to pressure palsies)	17	17p12	PMP22	(Generally submicroscopic) Deletion of the same region duplicated in CMTIA is associated with Hereditary Neuropathy with liability to Pressure Palsies (HNPP). More proximally placed deletions involving the 17p11.2 band are associated with Smith Magenis syndrome.		
	Miller-Dieker Lissencephaly syndrome	17	17p13.3	LIS1	microencephaly and a thickened cortex with 4 rather than 6 layers, "smooth brain"; 90% of patients with MDS have either a visible or submicroscopic deletion in this region. Phenotype: unusual facial appearance, and malformations of the heart and kidneys, polydactyly.	11.7 per million births to 40 per million	
	Isolated lissencephaly	17	17p13.3	LIS1	40% of patients with isolated lissencephaly (no associated malformations) have a submicroscopic deletion in this region detectable by LIS1 probes.		
	17q21.31 microdeletion	17	17q21.31	multiple (CRHR1, MAPT)	A recurring submicroscopic deletion associated with developmental delays, hypotonia, feeding difficulties, seizures, growth retardation, and an abnormal facies (long face, ptosis, bulbous nasal tip, hypoplastic nostrils with a long columella, broad chin, large low set ears, short philtrum, thin upper lip, and a protruding tongue).		
	18p deletion	18	18p		Deletion of all or a large portion of 18p is a classic chromosome deletion syndrome. The associated phenotype includes moderate growth deficiency, MR, hypotonia, microcephaly, holoprosencephaly, micrognathia, large ears.		
	18q deletion	18	18q		Another classic chromosome deletion syndrome. These are relatively large deletions, some interstitial, some apparently terminal. The phenotype includes MR, short stature, hypotonia, hearing impairment and foot deformities.		
	Alagille	20	20p12.3	JAG1	Deletions in this region are associated with Alagille syndrome, although fewer than 7% of individuals with Alagille have visible deletions. Alagille syndrome is caused by mutations of JAG1. Phenotype includes paucity of intrahepatic bile ducts, posterior embryotoxin and other anomalies.		
		21	21q22.3	EHOC1	Several patients have been reported with distal deletions in 21q and holoprosencephaly. One candidate is the Epilepsy Holoprocencephaly Candidate 1 (EHOC1) gene.		
	Di George's syndrome/VCF	22	22q11.2	HIRA, TBX1	Cardiac Abnormality, Abnormal facies, Thymic aplasia, Cleft palate, Hypocalcemia; This small deletion in proximal 22 is associated with DiGeorge/VCF (velocardiofacial AKA Shprintzen syndrome. DiGeorge syndrome is the infantile presentation including neonatal hypocalcemia, susceptibility to infection, and conotruncal heart anomalies. In older children, VCF syndrome includes bulbous nose with square nasal tip, hypernasal speech with (generally) milder cardiac anomalies. Short stature and mild-to-moderate learning disabilities are often seen, as well as psychiatric disorders in adults. The acronym CATCH 22 [cardiac abnormality, abnormal facies, T-cell deficit due to thymic hypoplasia, cleft palate, hypocalcemia, resulting from del(22)(q11)] has been proposed to describe the variable phenotype. Although the majority are de novo, approximately 10% are inherited deletions from an affected (often mildly affected) parent.	1 in 4000-5,950 births overall, 1 in 6000-6500 among whites, blacks and Asians, and 1 in 3,800 among Hispanics	
	22q13.3 microdeletion (Phelan-McDermid)	22	22q13.3	multiple	The “other 22q deletion syndrome" or Phelan-McDermid syndrome. The majority of these (distal) deletions are terminal, although some are the result of segregation out of a familial translocation. The clinical presentation includes infantile hypotonia, normal growth, global developmental delay, absent or delayed speech, autistic-like behavior, and minor dysmorphic features. The probe for the arylsulfatase gene (the “control probe” in the DiGeorge FISH assay), ARSA, has often been used to detect this deletion, although some patients have very small deletions in which ARSA is not deleted.		
		2	2p11.2q13		A recurring pericentric inversion. There appears to be no increased risk for liveborn unbalanced recombinant offspring among these inversion carriers although the risk for spontaneous abortions and stillbirths appears to be increased approximately 2-fold		

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Inversions		8	8p23.1q22.1		This pericentric inversion is typically seen in individuals of Hispanic descent with ancestry from the San Luis Valley of southern Colorado and northern New Mexico. These inversion carriers have an ~6% chance of having a child with recombinant 8 syndrome. The recombinant chromosome 8 contains a duplication of q arm material (q22.1 qter) and a deletion of p arm material (p23.1 pter). Children with this recombinant chromosome typically have MR, heart defects, seizures and a characteristic facies (hypertelorism, thin upper lip, anteverted nares, wide face, abnormal hair whorl, low set ears, downturned mouth, low posterior hairline, etc).		
		9	9p11q13		A recurring pericentric inversion that is not associated with an increased risk for liveborn unbalanced recombinant offspring, spontaneous abortions or stillbirths.		
		11	11q21q23		A recurring paracentric inversion common in the Netherlands and among the Canadian Hutterites. There is no increase in the rate of spontaneous abortions among carriers of the inversion or their partners and recombinant chromosomes arising from the inversion are thought to be rare.		
		15	+inv dup(15) or psu idic(15)(q)		These small, often dicentric chromosomes are composed of 2 copies of chromosome 15 short arm, (usually) 2 centromeres and 2 copies of varying amounts of proximal 15q material. If the "marker" contains the probe loci that are usually deleted in PWS/AS, the patient will have MR and often autism. If these loci are not present in the marker, the phenotype can be normal. These are the most common identifiable markers or ESAC's seen in the human population.		
Translocations		4, 8	4;8(p16;p23)		One of only 2 recurring constitutional reciprocal translocations; mediated by olfactory receptor gene clusters on chromosomes 4 and 8. Patients with unbalanced karyotypes and the der(4) have features associated with Wolf-Hirschhorn syndrome		
		11, 22	t(11;22)(q23.3;q11.2)		One of only 2 recurring constitutional reciprocal translocations; mediated by palindromic repeat sequences on both chromosomes. Patients with unbalanced karyotypes almost always have a supernumerary der(22)t(11;22) and Emanuel syndrome.		
	Emanuel syndrome	11, 22	+der(22)t(11;22)(q23.3;q11.2)		This karyotype is seen in the viable offspring of individuals who carry the recurring t(11;22) translocation and results in a double trisomy involving proximal chromosome 22 and distal chromosome 11 long arm material. Emanuel syndrome is associated with mental retardation, congenital heart disease, malformed ears with preauricular skin tags and/or pits, a high arched or cleft palate, micrognathia, anal stenosis or atresia, renal aplasia or hypoplasia and genital abnormalities in males.		
Monosomy or Deletion	Turner syndrome	XO or mosaic XX/X0			short stature, lymphedema, broad chest, low hairline, low-set ears, amenorrhea, small fingernails, webbed neck, horseshoe kidney	1 in 1500- 2,500 newborn girls	Females only
	AHC (Adrenal Hypoplasia Congenita)	X	Xp21.2-21.3	NR0B1	Congenital adrenal hypoplasia (AHC) is a rare disorder that can be inherited in an X-linked or autosomal recessive (see 240200) pattern. In X-linked AHC, primary adrenocortical failure occurs because the adrenal glands lack the permanent adult cortical zone. The remaining cells are termed 'cytomegalic' because they are larger than typical fetal adrenal cells (Hay et al., 1981; Reutens et al., 1999).		
	CGKD (Complex Glycerol Kinase Deficiency)	X	Xp21.2-21.3	NR0B1, DMD, GK	Francke et al. (1987) noted that there are 3 clinically distinct forms of glycerol kinase deficiency: infantile, juvenile, and adult. The infantile form is associated with severe developmental delay, and those with the adult form have no symptoms and are often detected fortuitously. Walker et al. (1996) noted that an infantile form of GK deficiency, or the 'complex,' results from the Xp21 contiguous gene deletion syndrome with congenital adrenal hypoplasia (300200) and/or Duchenne muscular dystrophy (DMD; 310200), whereas the juvenile and adult forms have isolated GK deficiency.		
	Duchenne Muscular Dystrophy	X	Xp21.2	DMD	Dystrophin-associated muscular dystrophies range from the severe Duchenne muscular dystrophy (DMD) to the milder Becker muscular dystrophy (BMD; 300376). Mapping and molecular genetic studies indicate that both are the result of mutations in the huge gene that encodes dystrophin, also symbolized DMD. Approximately two-thirds of the mutations in both forms are deletions of one or many exons in the dystrophin gene. Although there is no clear correlation found between the extent of the deletion and the severity of the disorder, DMD deletions usually result in frameshift.		
	Kallmann 1	X	Xp22.3	KAL1	Males with Kallmann syndrome typically carry a submicroscopic deletion involving the Kallmann syndrome 1 (KAL1) gene and demonstrate hypogonadotropic hypogonadism and anosmia (inability to smell). Carrier females typically have milder manifestations. These deletions lie proximal to those associated with X-linked ichthyosis		

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X/Y Linked		Steroid Sulfatase Deficiency (Ichthyosis, X-linked)	X	Xp22.3	STS	Males who carry a submicroscopic deletion of the steroid sulfatase (STS) gene have a skin disorder referred to as X-linked ichthyosis. In contrast, females who are heterozygous for the deletion are not typically affected, although mild manifestations are sometimes present. These deletions lie distal to those associated with Kallmann syndrome		
	Duplication	DSS (Dosage sensitive sex-reversal)	X	Xp21.2-21.3	NR0B1	Sex-reversed XY males result from duplication of the DAX1 gene. In the presence of a DAX1 duplication, the male determining function of SRY is suppressed and ovarian development results.		
		Pelizaeus-Merzbacher	X	Xq22.2	PLP1	A large percentage of patients with Pelizaeus-Merzbacher disease contain duplications involving the proteolipid protein 1 (PLP1) gene and flanking sequences. Typical patients demonstrate nystagmus, spasticity, ataxia, and MR.		
	Trisomy/tetrasomy, etc	Klinefelter's syndrome	47, XXY			infertile, language learning impairment, lanky, youthful build and facial appearance, rounded body type, gynecomastia, hypogonadism, microorchidism	1 in 500 to 1,000 males, but most variants much rarer	Males only
			48, XXXY			tall stature, gynecomastia, truncal obesity, skin ulcers, craniofacial dysmorphism	1 in 50,000 male births	Males only
			49, XXXXY			cleft palate, club feet, respiratory conditions, short/broad neck, low birth weight, short stature, narrow shoulders, hypertelorism, prognathism, congenital heart defects	1 in 85,000 males	Males only
		Triple X syndrome	47, XXX			menstrual irregularities, increased risk of learning disabilities, delayed speech, deficient language skills, delayed development of motor skills	1 in 1,000 newborn girls	Females only
			48, XXXX			epicanthal folds, flat nasal bridges, upslanting palpebral fissures, midface hypoplasia, small mouths, cleft palates, absent teeth, muscle tone abnormalities	1 in 1,000 newborn girls	Females only
			49, XXXXX			microencephaly, micrognathia, round face, low-set ears, eyes show palpebral fissures, hypertelorism, strabismus	1 in 1,000 newborn girls	Females only
			47, XYY			increased growth velocity (early childhood), severe acne	1 in 1,000 newborn boys	Males only
	Inversion		Y	inv(Y)(p11;q11)		This pericentric inversion typically represents a benign variant, however some inversions have been reported in males with infertility due to a small accompanying deletion.	The incidence of this inversion is estimated to be 0.6/1000 males, but a much higher incidence (30.5%) has been reported in the Gujerati Muslim Indian population.	