

LIST OF CONGENITAL ABNORMALITIES AND HEREDITARY CONDITIONS
(EXCLUSIVE OF MONGOLISM AND SEXUAL ANOMALIES) SHOWING
AN APPARENTLY NORMAL KARYOTYPE (FROM COMPILATIONS)
BY FORD AND HARNDEN, JACOBS AND BUCKTON

Achondroplasia	Gargoylism (Hurler's syndrome)
Acrocephalosyndactyly (Apert's disease)	Gaucher's disease
Albright's syndrome	Hemophilia (chromatin-negative "girl")
Amaurotic idiocy	Huntington's chorea
Amyotonia	Hydatidiform mole**
Anencephaly	Hydrocephalus
Arachnodactyly (Marfan's syndrome)*	Laurence-Moon-Biedl syndrome
Arthrogyphosis	Little's disease
Chondrodystrophy	Mental defective:
Conjoined twins	With hemophilia
Crouzon's disease (dysostosis craniofacialis)	With malformed ears; webbed toes; absent fifth metacarpal
Cyclops deformity	Microcephaly
Dwarfism:	Muscular dystrophy
Primordial	Neurofibromatosis
Hypopituitary	Osteogenesis imperfecta
Ehlers-Danlos syndrome	Phenylketonuria
Epiloia	Sympodia with eventration and renal agenesis
Frohlich's syndrome	

*Abnormal satellites have been found in familial cases.

** Abnormal numbers of chromosomes have been found in the cells of the villi of hydatidiform moles.

ABNORMAL KARYOTYPES REPORTED IN MULTIPLE CONGENITAL ABNORMALITIES
(WITHOUT MONGOLISM OR DOMINATING SEXUAL ANOMALIES)*

Sex	Chromosome Abnormability	Karyotype	Clinical Condition
F	Trisomy in group 16-18	(47) XX	Cardiac defect, webbed neck, shield-like chest, mental retardation
F	Trisomy in group 13-15	(47) XX	Apparent anophthalmia, cleft palate, hare lip, simian creases, polydactyly, cardiac defect, cerebral defect, capillary hemangiomas, mental retardation
M	Trisomy for no. 22	(47) XY	Sturge-Weber syndrome, mental retardation
M	Trisomy in group 6-12 (apparently for nos. 8 and 11) and of the sex chromosomes	(49) XXY	Cardiac and renal defects, peculiar facies, malformed external genitalia and testis, mental retardation
M	Complete trisomy (triploidy)	(69) XXY	Porencephaly, micrognathia, syndactyly, mental retardation
M	Translocation of no. 22 to one of group 13-15	(45) XY	Malformations of vertebral column, small head contour, abnormal sella turcica, mental retardation (polydysspondylism)
M	Enlarged satellite on no. 21	(46) XY	Marfan's syndrome (familial)
M	Enlarged satellite on no. 18	(46) XY	Marfan's syndrome (familial)
F	Enlarged satellite on no. 21	(46) XX	Cleft palate, fused vertebrae, cardiac defect, abnormal ears, short neck
F	Elongated satellite on no. 21	(46) XX	Hare lip, rudimentary nose and eyes, cerebral defects, adrenal hypoplasia

*Taken from Arthur R. Schval, M.D., Recent Progress in Human Chromosome Analysis and Its Relation to the Sex Chromatin, *The American Journal of Medicine*, XXXI:397-441, September, 1961.

SUMMARY OF CHROMOSOME FINDINGS
REPORTED IN VARIOUS CLINICAL STATES

Clinical State	Sex Chromatin	Chromosome Constitution
Normal male	-	(46) XY
Normal female	+	(46) XX
Pseudohermaphroditism		
Male	-	(46) XY
Female	+	(46) XX
True hermaphroditism	+	(46) XX
	-	(46) XY
Klinefelter's syndrome	+	(47) XXY, (47/46) XXY/XX, (48) XXYY
	-	(46) XY, (47/46) XXY/XY
Turner's syndrome	-	(45) XO, (45/46) XO/XX,
	+	(45) XO, (45/46) XO/XX, (46) XX
"Pure" gonadal dysgenesis	-	(46) XY
"Atypical" gonadal dysgenesis	+*	(46) Xx
	=	(45) XO
Poly-X conditions		
Female phenotype		
Fertility or premature menopause	+	(47) XXX
Primary amenorrhea	+	(47/45) XXX/XO
Male phenotype		
Microorchidism	+	(48) XXXY, (48/49) XXXY/XXXXY
Mongolism		(47) with trisomy for no. 21 (46) with trisomy and monosomy or translocation
Multiple congenital anomalies**		(47) with trisomy for various autosomes (49) XXY with triple trisomy (69) XXY, complete trisomy (45) with centric fusion Abnormal satellites
Leukemia**		Abnormal numbers and forms
Radiation effects		Abnormal forms and numbers

*Sex chromatin bodies (small) in 7 per cent of buccal smear nuclei.

**Chromosome abnormalities in some but not all instances.

HUMAN CYTOGENETICS

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