GENETIC DISORDERS

Disorder	Abnormality / Karyotype	Physical features	Clinical manifestations
AUTOSOMAL ANEUPLOIDIES [numerical]			
Down syndrome	trisomy 21 47,[xx or xy]+21	Low nasal bridge, low-set ears, epicanthal folds in the eye, up slanting palpebral fissures, protruding tongue, gap between the first and second toes, Simian line	CHD [VSD, AV], muscle hypotonia, intestinal problems, increased risk for leukemia, mental retardation
Edward syndrome	trisomy 18 47,[xx or xy]+18	prominent occipital bone, low-set, malformed ears, short sternum, clenched fists, rocker-bottom feet	Mental retardation, CHD, failure to thrive, intestinal problems, hypertonia,
Patau syndrome	trisomy 13 47,[xx or xy]+13	scalp defects, microcephaly, small eyes, low-set malformed ears, cleft lip/palate, polydactyly & syndactyly, rocker-bottom feet	CHD, mental retardation, hypo- or hypertonic, polycystic kidneys
SEX CHROMOSOME ANEUPLOIDIES [mumerical]			
Klinefelter syndrome	extra X chromosome 47, xxy	a MALE with secondary female characteristics [fat distribution, breast development], underdeveloped male sexual organs, coarse/reduced body hair, taller than average	infertility, evidence of mental retardation, evidence of osteoporosis
Turner syndrome	absent chromosome y [only viable monosomy] 45, xo	FEMALE, brown skin spots [nevi], underdeveloped breasts with wide distance between the nipples, shorter than average, abnormal elbow position, webbed neck, small nails, shortened metacarpals	sterility, infertility, no menstrual cycle

Structural abnromalities			
Cri-du-chat	deletion in the p arm of chromosome 5	small head, small chin, small nasal bridge, unusually round face, eyes far from each other	heart defects, hearing or sight problems, motor problems, hyperactivity or aggression in some cases, severe mental retardation
Chronic myelogenous leukemia	translocation [reciprocal] mutation t(9;22)		