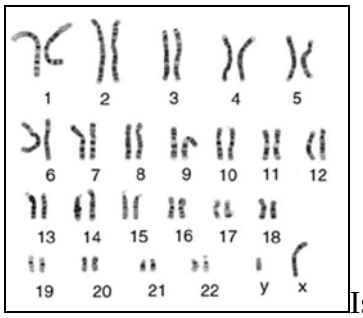
Class Notes ***Genetic Disorders*** **Questions/Main Idea:**	Name: _____ Period:_____ Date: _____ **Notes:**
Types of genetic disorders	• Autosomal disorders: found on chromosome pairs 1-22 (autosomes) • Sex-linked (recessive) disorders: found on chromosome pair 23 (sex chromosomes) • Chromosomal disorders: too few or too many of a chromosome
Sex-linked disorders 	Found on X chromosome, generally affects men -- • **Colorblindness**: can't distinguish colors, commonly red/green (1:10 males, 1:100 females) • **Hemophilia**: blood fails to clot after injury (1:10,000 males) • **Duchenne Muscular Dystrophy**: rapid weakening and loss of skeletal tissue (1:3,500 boys)
Autosomal recessive disorders	• **Cystic Fibrosis (CF)**: Mucus clogs airways and ducts in lungs and other organs; digestive problems (1:3,500) • **Albinism:** lack of pigment (melanin) in skin, hair, eyes, extremes case deafness (1:17,000) • **Sickle Cell Disease:** Abnormal hemoglobin is rigid and sickle-shape, can't transport oxygen well and get stuck in capillaries tissues (1:500 African-American births, 1:1,200 Hispanic births); heterozygous = malaria resistant • **Phenylketonuria (PKU):** Destroys the nervous system and causes mental retardation (1:15,000), easily treated • **Tay-Sachs Disease:** Causes mental retardation, blindness, muscle weakness (1:5,000); 1:27 eastern European Jews is a carrier
Autosomal dominant disorders	• **Huntington's Disease (HD):** Wasting away of brain tissue, causes uncontrolled movements, emotional disturbances, mental deterioration, fatal (8:100,000) • **Acondroplasia:** Bone disorder causing dwarfism (1:30,000)
Chromosomal disorders	• Having more or less than 46 chromosomes (in humans) • Generally **NOT** inherited • Happens when homologous chromosomes fail to separate during meiosis → **nondisjunction**

What is a karyotype?	• **Karyotype** → an organized picture of a person's chromosomes  Is this male or female?
Down Syndrome (DS)	• Extra chromosome 21 (aka Trisomy 21) • Birth defects, mild to severe mental retardation, deformed facial features (1:800)
Turner's Syndrome	• Missing an X chromosome • Person is sterile, sex organs do not develop at puberty (1:2,500 females)
Klinefelter's Syndrome	• An extra X chromosome (XXY) • Sterile, tends to have both stunted male and feminine features (1:750 males)

Summary: