

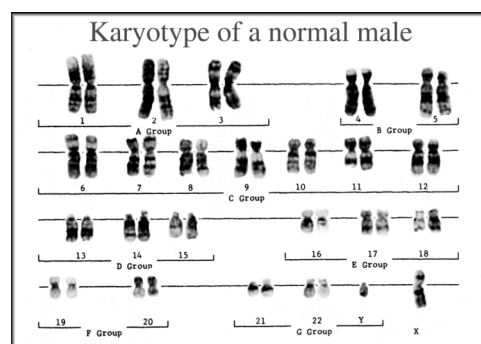
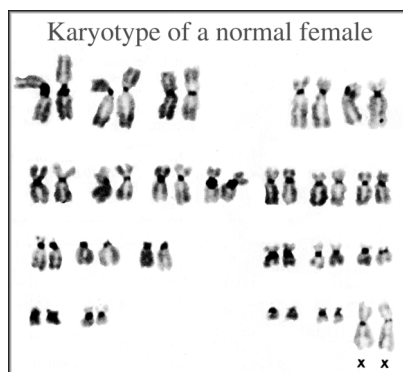
Human Chromosomal Disorders

Resulting from Variations of Chromosomal Number



Normal Human Karyotypes

(metaphase chromosomes from a diploid cell)



**Note that there are a total of 46 chromosomes for each individual and that the female has two X chromosomes in position 23 whereas the male has an X and Y in position 23*

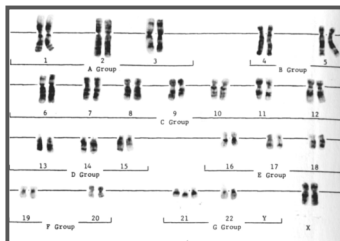
Examples of Chromosomal Variations

I. Extra Chromosomes

A. Down Syndrome = one extra chromosome 21

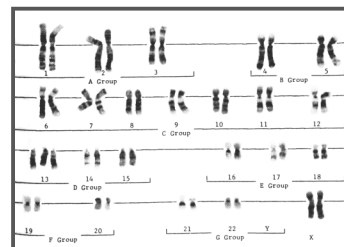
↳ written as (47,+21)

1. 1/700 births of individuals of European decent
2. 1/6 die within the first year
3. average life expectancy is 16 years
4. lower than average mentality
5. many physical deviations such as shorter than average height, broad forehead, round head, open mouth, etc.



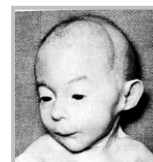
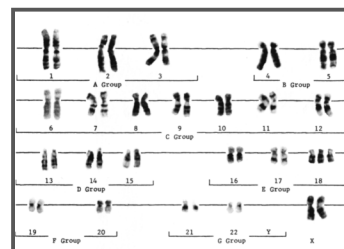
B. Trisomy 13 (47, +13)

1. 1/20,000 newborns
2. longest life = 5 years; most die within 3 months of birth
3. symptoms include small brain, deafness, and many other internal and external abnormalities

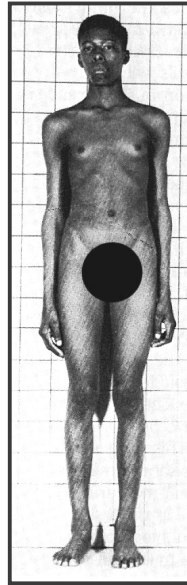


C. Trisomy 18 (47, +18)

1. 1/8,000 newborns
2. longest life = 13 years old; 90% die within 6 months
3. small mouth and nose, receding lower jaw, and a general elfin appearance



- D. Klinefelter Syndrome = males with extra X-chromosomes**
- ↘ can be **XXY, XXXY, OR XXXXY**
1. **1/500 births**
 2. **phenotypically male with some female tendencies - particularly physical characteristics**
 3. **enlarged breasts, low body hair count, small testes, long legs, and thin**
 4. **sterile**
 5. **mental retardation when there are more than 2 X-chromosomes**



- E. Triple X and Quad X Syndrome**
- ↘ females with 1 or 2 extra X-chromosomes (**XXX, or XXXX**)
1. **1/700 female births**
 2. **sometimes there is no visible difference from XX females**
 3. **limited fertility**
 4. **XXXX females are always mentally retarded**

II. Fewer Chromosomes

A. Turner Syndrome

- ↘ females with only one X-chromosome
1. **1/2500 live female births**
 2. **more than 90% self abort**
 3. **no ovaries, short, shield-like chest**
low set ears, webbed neck
 4. **no mental deficiencies**

