

# Nondisjunction disorders

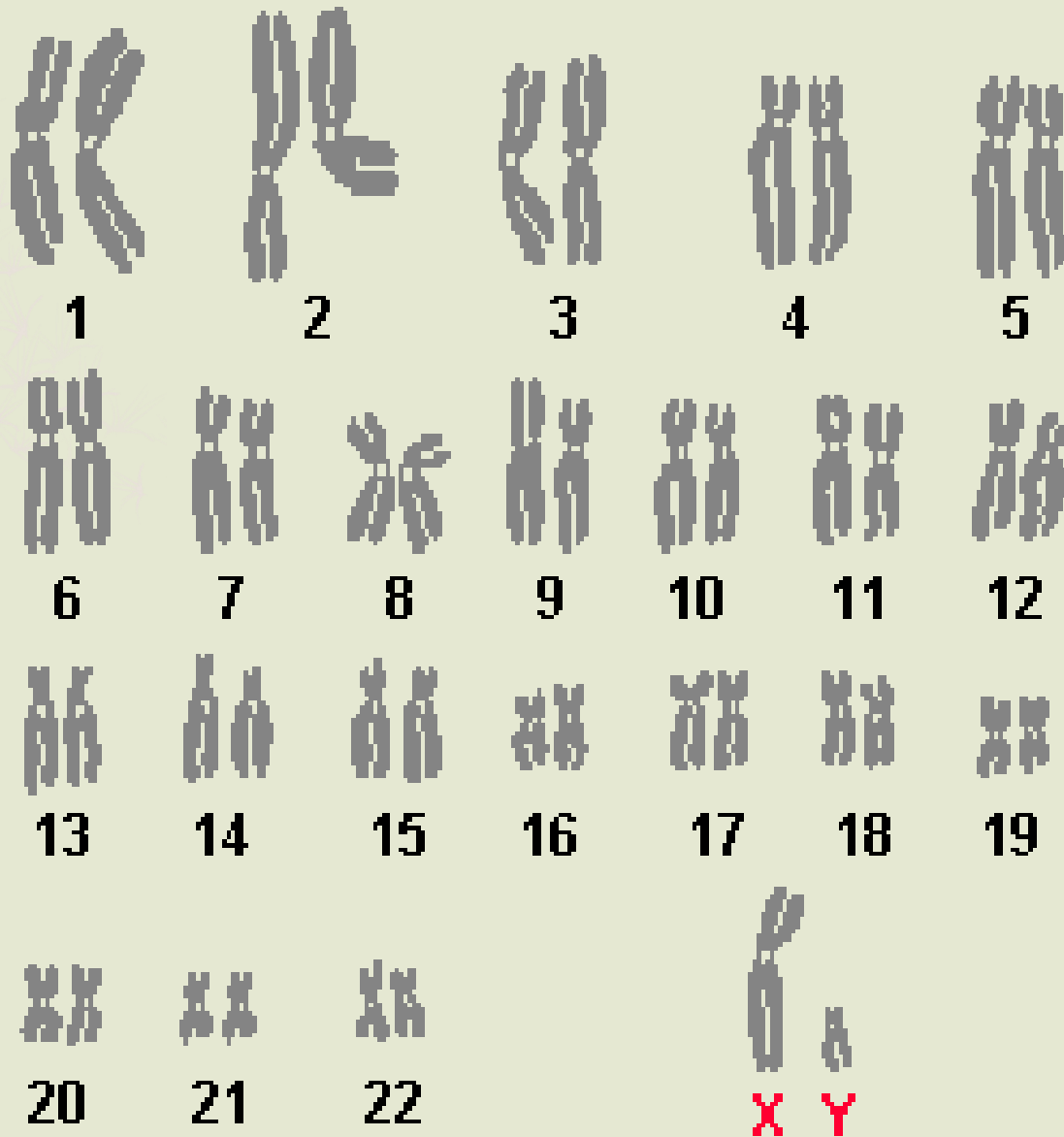
And other abnormal chromosome events

Taken from Mr. Blankenship's website at Rock Hill HS

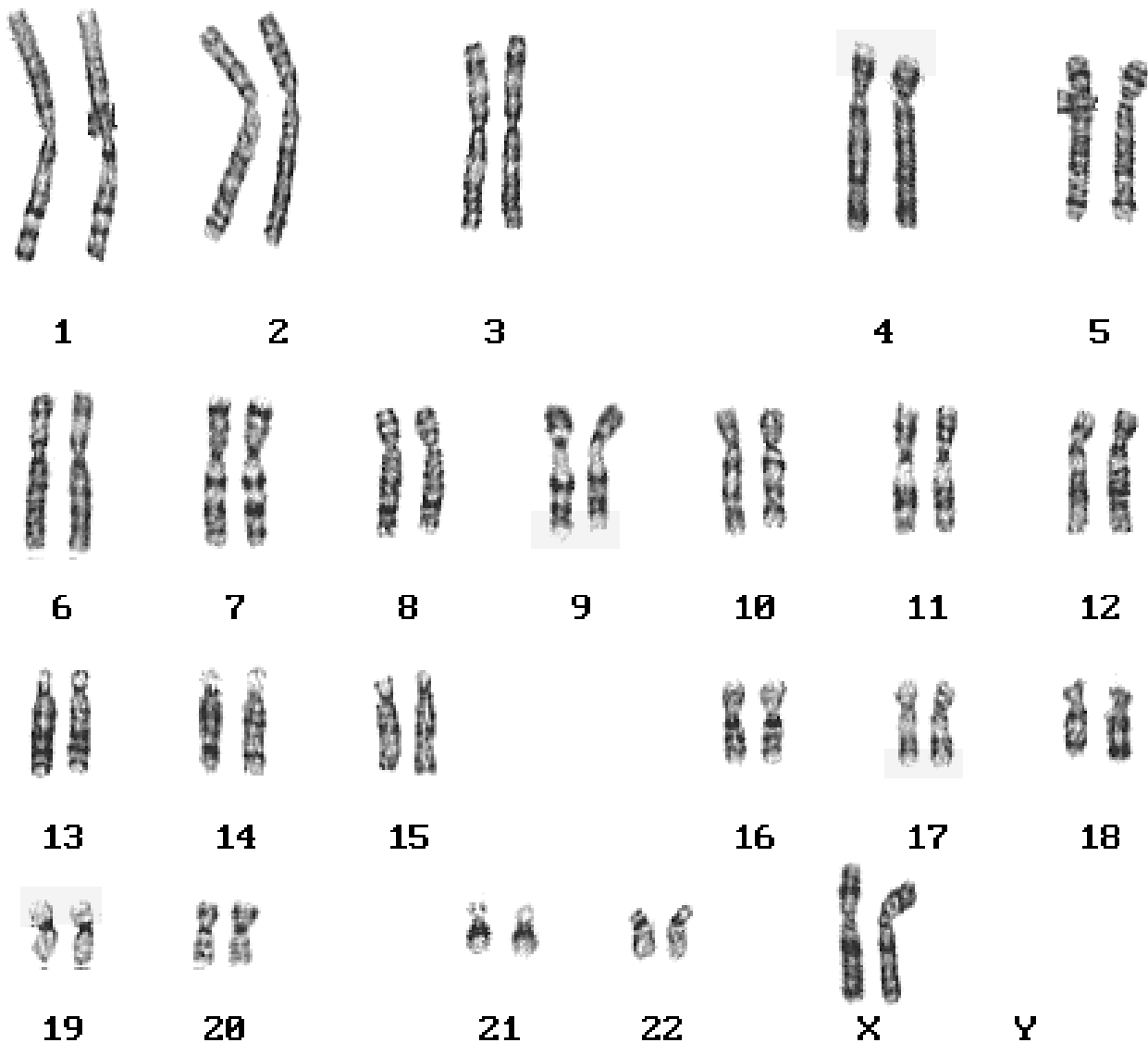
*[www.classhelp.info/Biology/Nondisjunction%20disorders.ppt](http://www.classhelp.info/Biology/Nondisjunction%20disorders.ppt)*



# Human Male Karyotype



Human  
male  
karyotype,  
written as  
**46 XY**



Karyotype: 46,XX

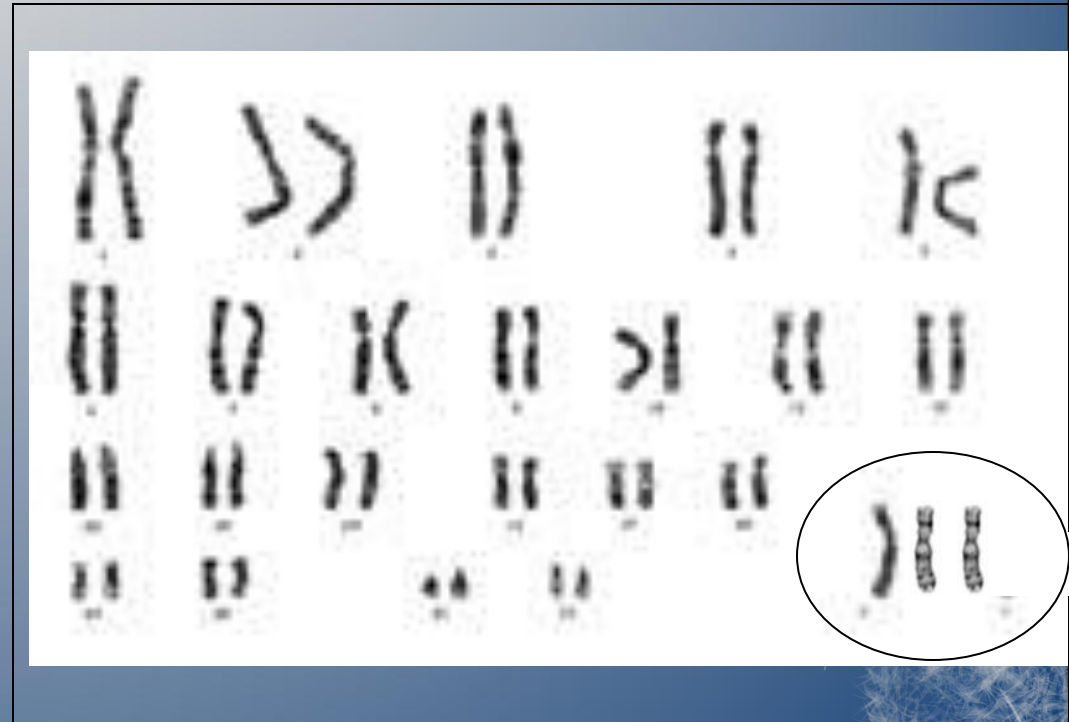
Human  
female  
karyotype,  
written as  
**46 XX**

# Triple X Syndrome

**1 in 2,500 births**

**47 chromosomes  
XXX only**

**#23 Trisomy  
Nondisjunction**



# Triple X Syndrome

Normally physically  
Normal mentally  
Fertile



# Jacob's syndrome

- XYY

- No abnormal appearance/behavior

- fertile



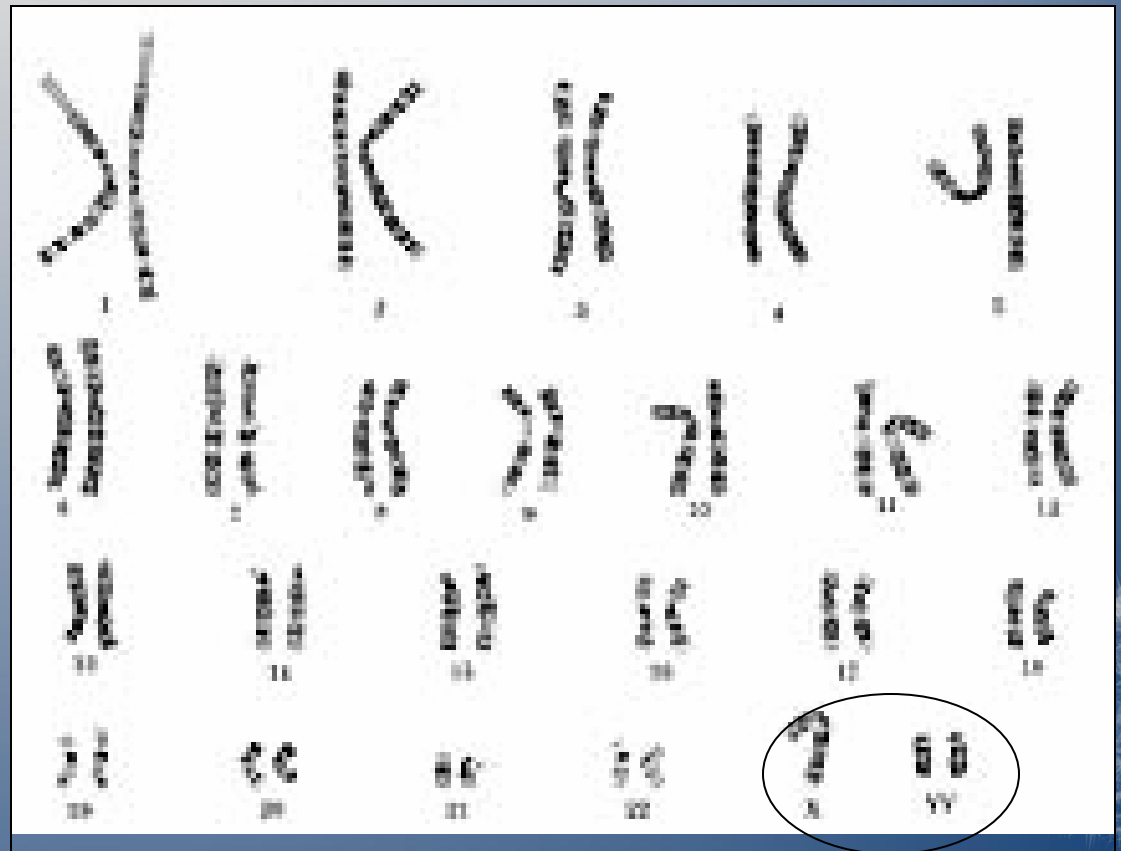
# Jacob's Syndrome

**1 in 1,800 births**

**47 chromosomes  
XYY only**

**47XYY**

**#23 Trisomy  
Nondisjunction**



# Jacob's Syndrome

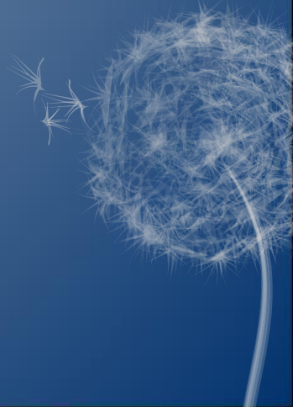
Normal physically

Normal mentally

Increase in testosterone

Perhaps more aggressive

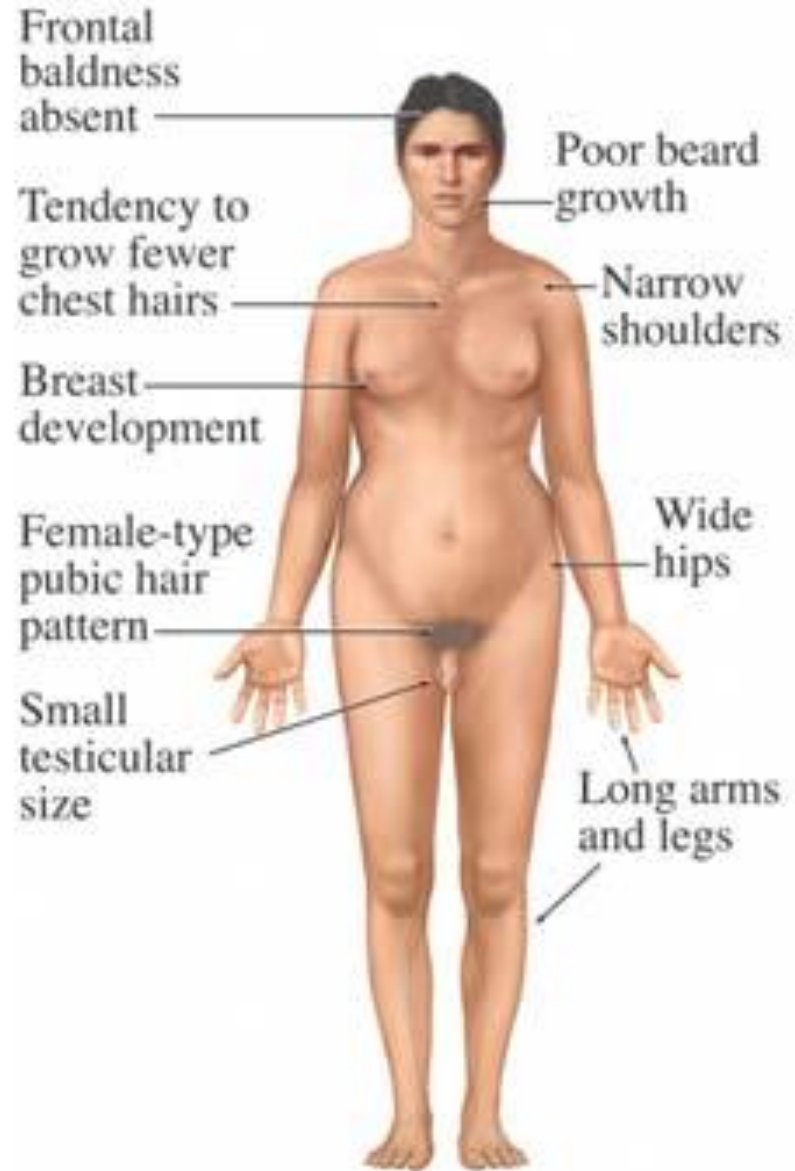
Normal lifespan





# Klinefelter's Syndrome

✱ XXY, male



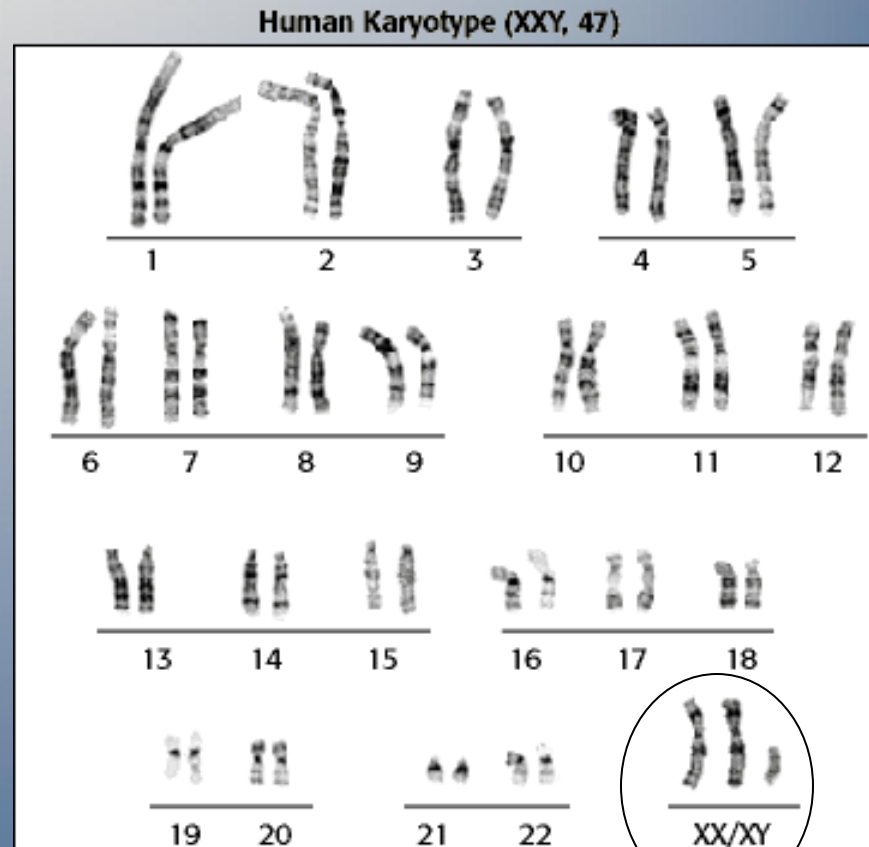
# Klinefelter's Syndrome

1 in 1,100 births

47 chromosomes  
XXY only

47, XXY

#23 Trisomy  
Nondisjunction



# Klinefelter's Syndrome

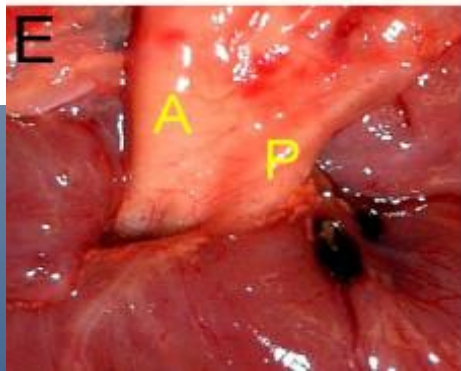
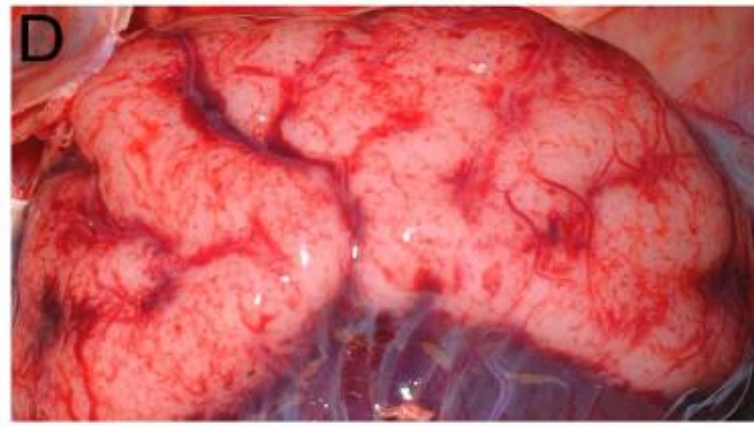
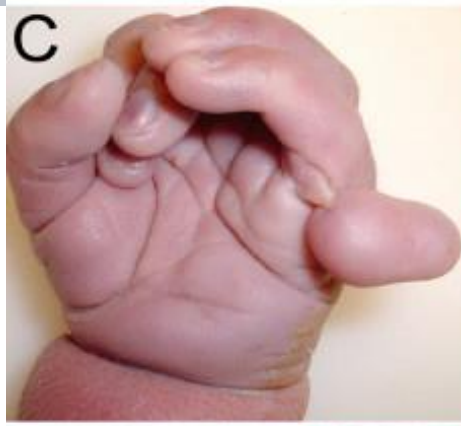
- Scarce beard
- Longer fingers and arms
- Sterile
- Delicate skin
- Low mental ability
- Normal lifespan





# Patau syndrome

★ 47, XX, +13



# Patau's Trisomy Syndrome

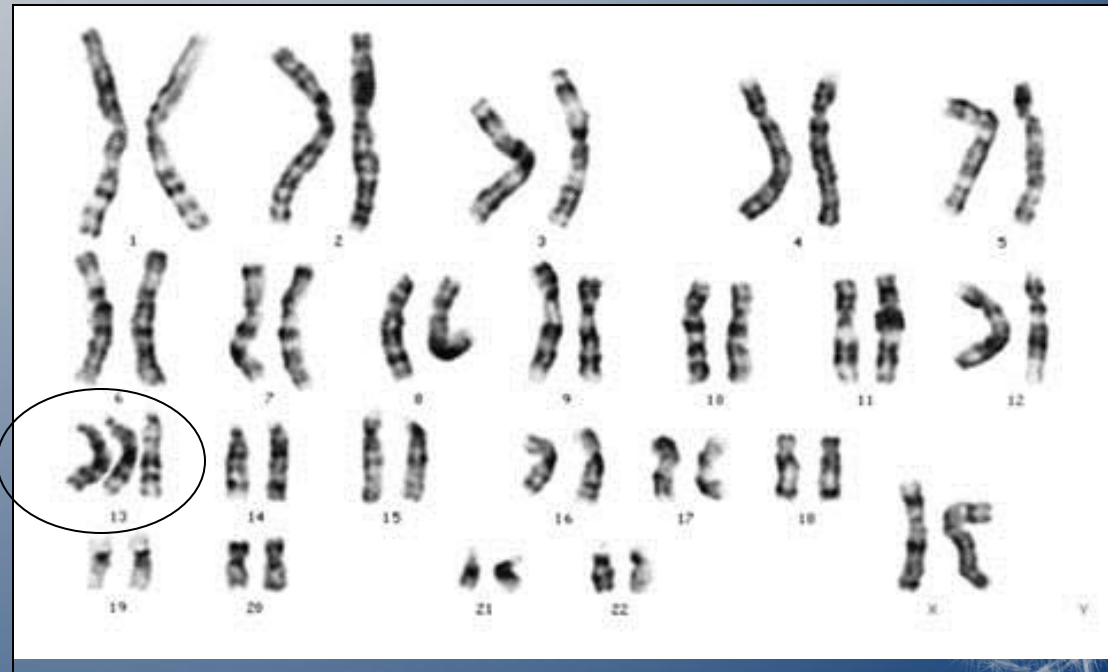
1 in 14,000 births

47 chromosomes

XY or XX

47, +13

#13 Trisomy  
Nondisjunction



# Patau's Trisomy Syndrome

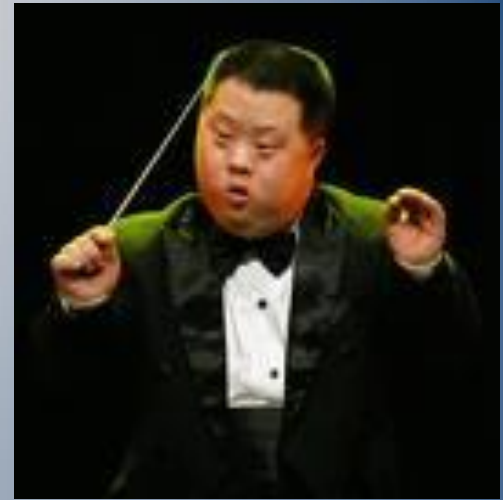
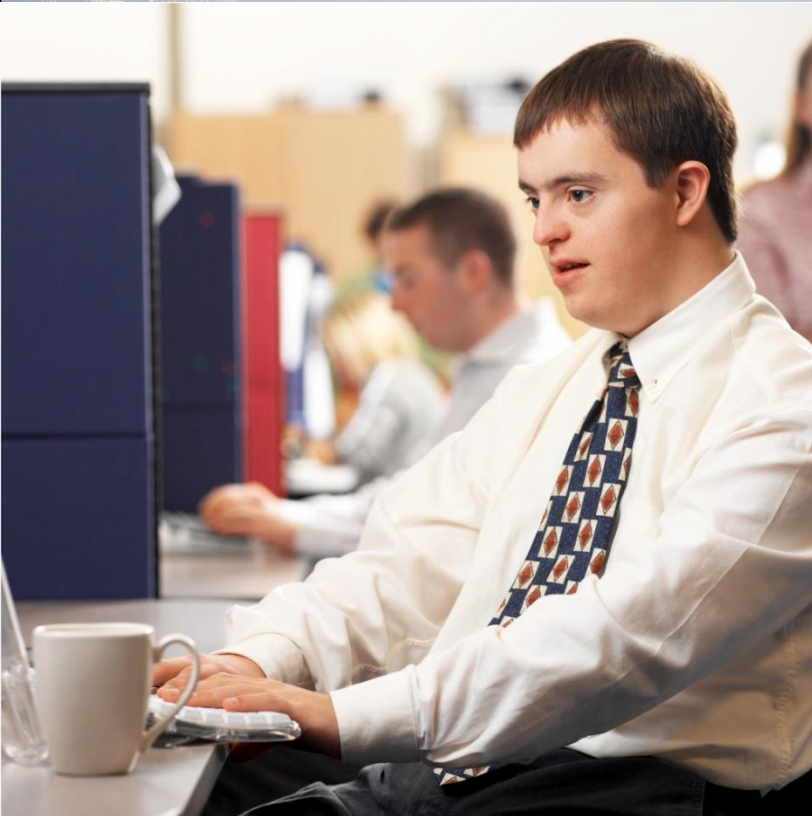
- Small head
- Small or missing eyes
- Heart defects
- Extra fingers
- Abnormal genitalia
- Mentally retarded
- Cleft palate
- Most die a few weeks after birth





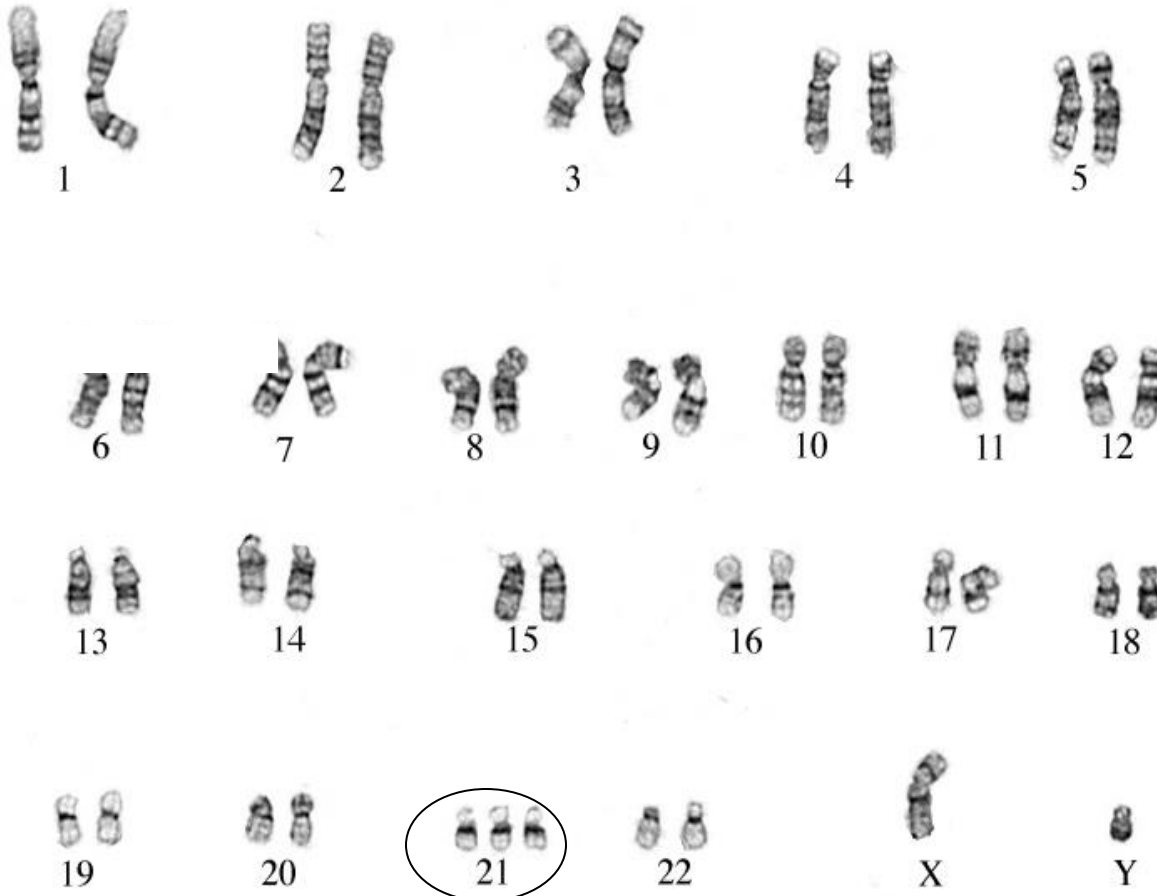
# Down Syndrome

★ 47, XY, +21



# Down Syndrome

ZWK99024 KEY



**1 in 1,250 births**

**47 chromosomes  
XY or XX**

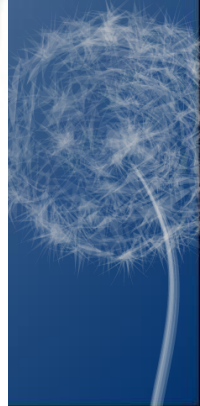
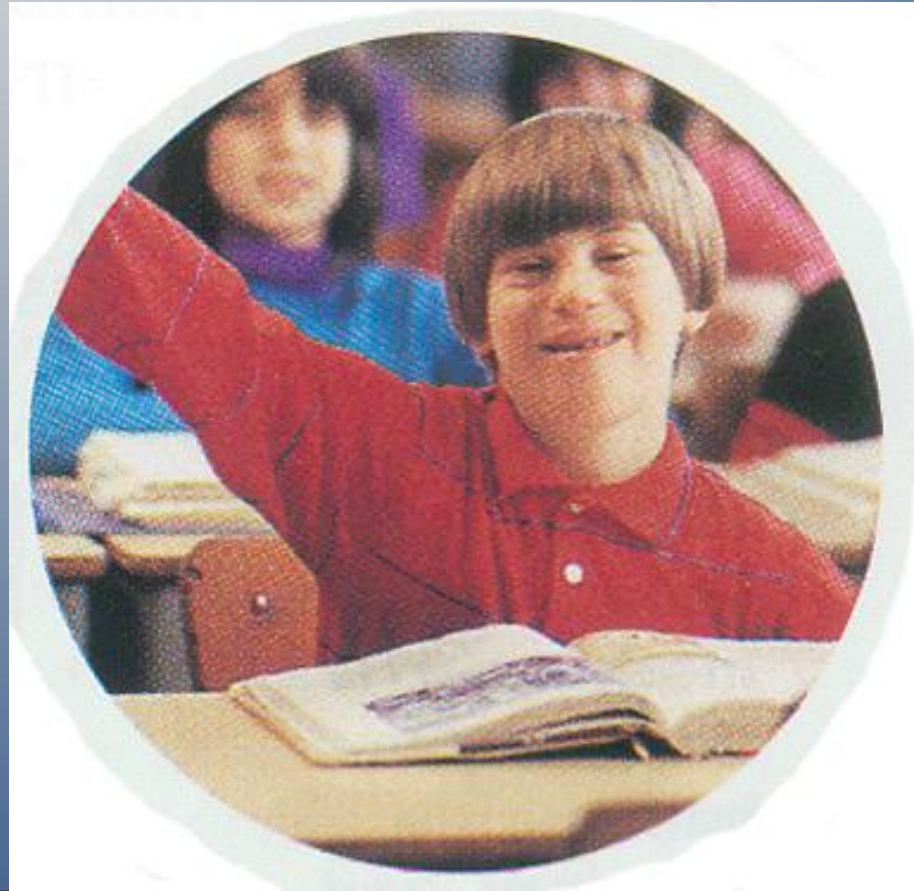
**#21 Trisomy  
Nondisjunction**





# Down Syndrome

Short, broad hands  
Stubby fingers  
Rough skin  
Impotency in males  
Mentally retarded  
Small round face  
Protruding tongue  
Short lifespan



# Edwards Syndrome

★ 47, XY, +18



# Edward's Trisomy Syndrome

**1 in 4,400 births**

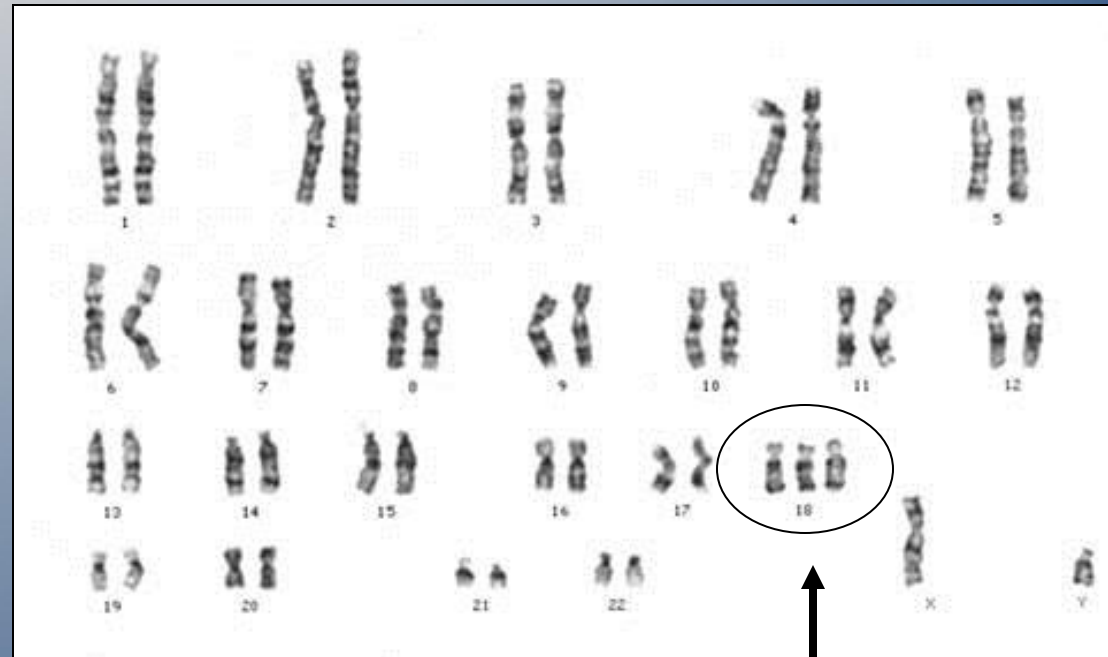
**47 chromosomes**

**XX=80%**

**XY=20%**

**#18 Trisomy**

**Nondisjunction**



# Edward's Trisomy Syndrome

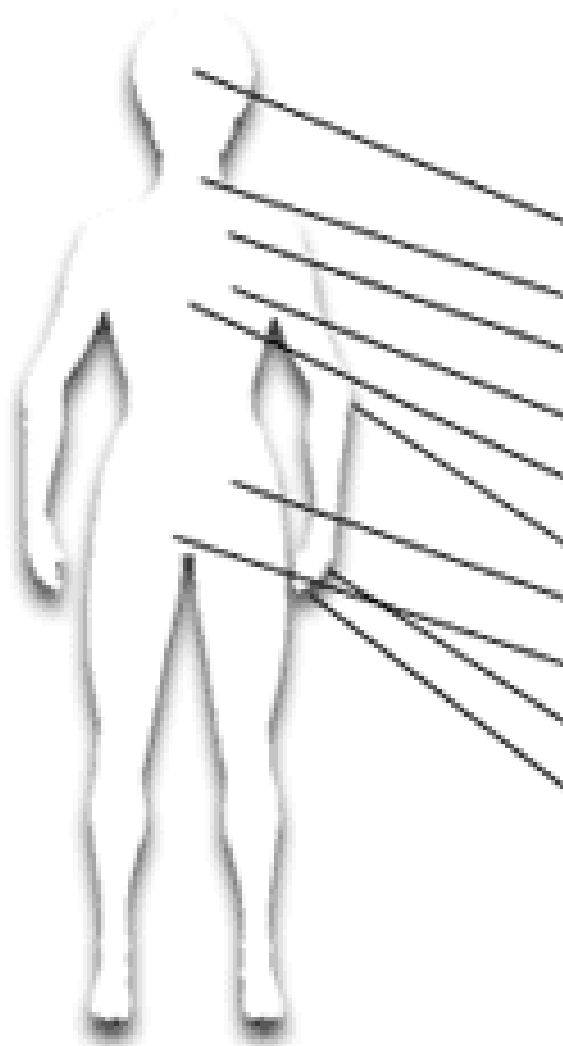
Small head  
Mentally retarded  
Internal organ  
abnormalities  
90% die before  
5 months of age



# Turners Syndrome

★ 45, X

## Common Symptoms of Turner Syndrome



- short stature
- low hairline
- fold of skin on neck
- constriction of aorta
- underdeveloped breasts
- shield-shaped thorax
- elbow deformity
- rudimentary ovaries
- no menstruation
- shortened metacarpal IV
- small finger nails
- brown spots (nevi) on skin



# Turners Syndrome

96-98% do not survive to birth

No menstruation

No breast development

No hips

Broad shoulders and neck



# Trisomy 16, monosomy X

★ 46, X, +16

★ Not viable beyond 1<sup>st</sup> trimester

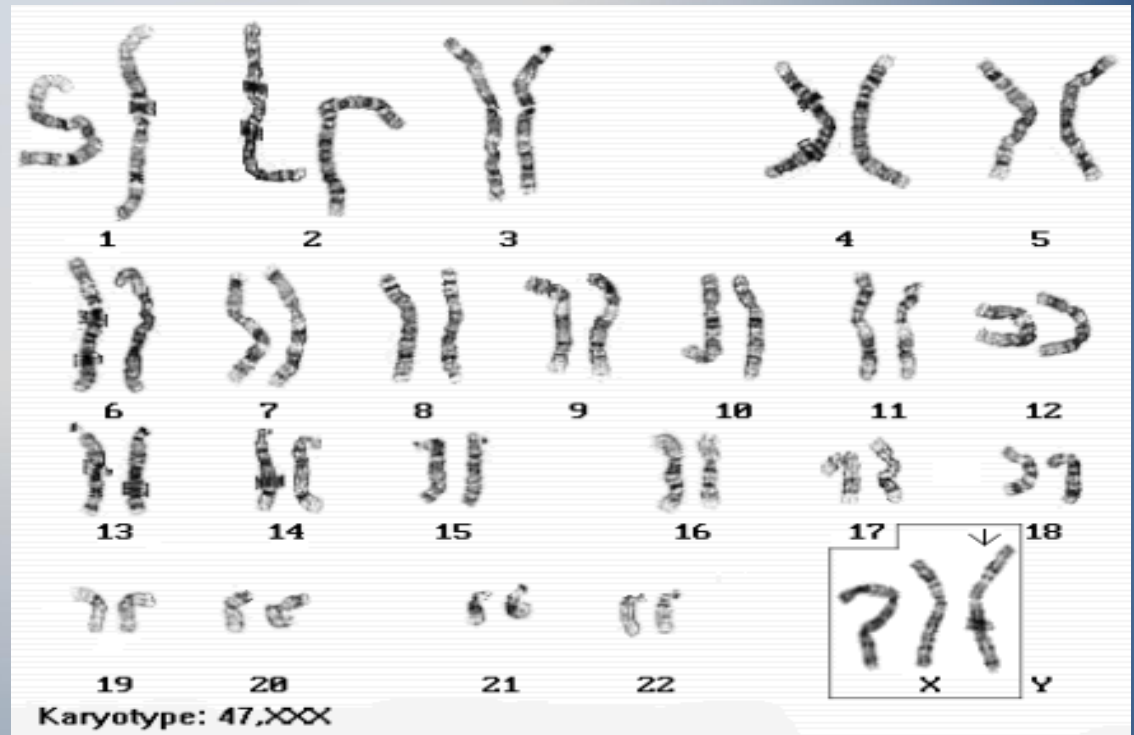
# Trisomy X

★ 47 XXX

symptoms

★ 1/1000

★ healthy and fertile -  
cannot be  
distinguished from  
normal female  
except by karyotype





# Robertsonian Translocation 13-14

- Several types, with varying results.



# Wolf Hirshhorn Syndrome

★ 4p-

- ★ Very rare. Affected children are small, with microcephaly and abnormal facies. There are cardiac, renal, and genital abnormalities. Most are stillborn or die in infancy.

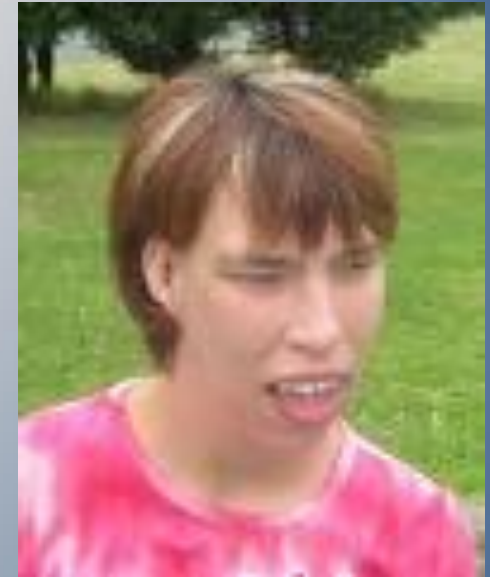
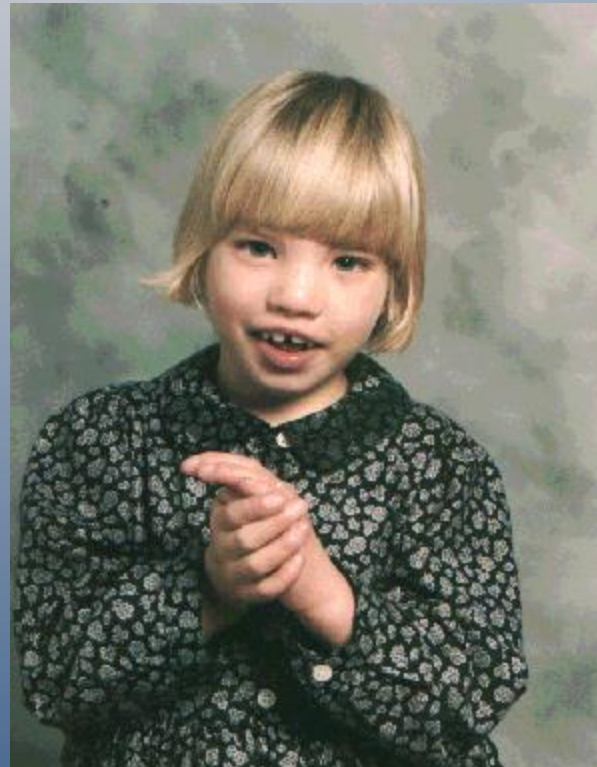


# Cri-du-chat Syndrome

★ 5p-

site

- Moon-shaped face
- Heart disease
- Mentally retarded
- Malformed larynx
- Normal lifespan



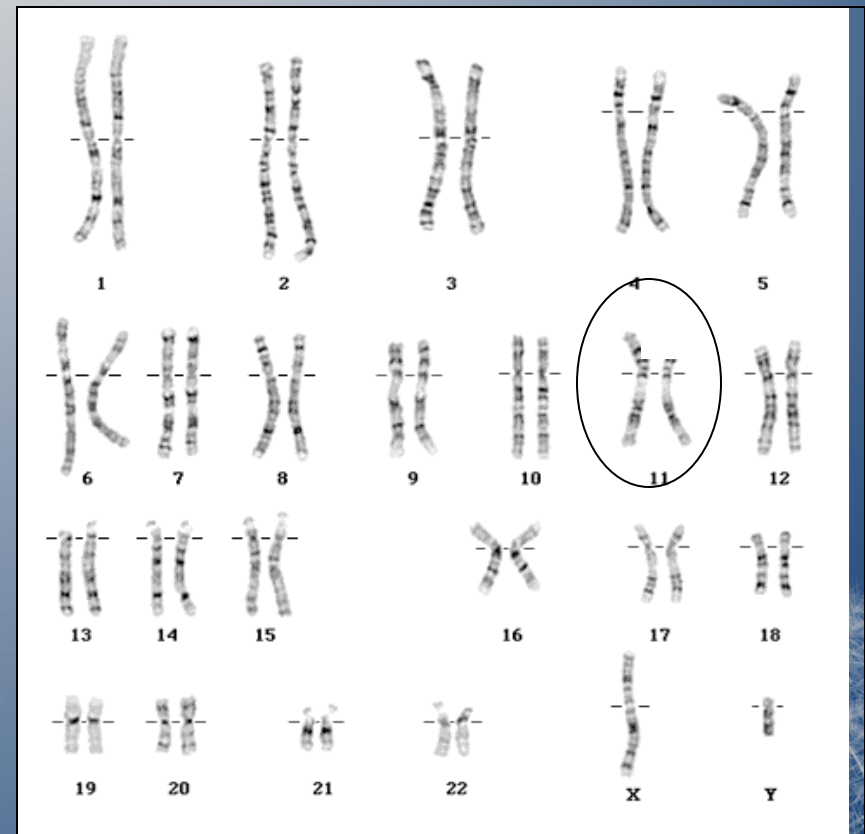
# Aniridia-Wilms Tumor Syndrome

1 in 50,000,000 births

46 chromosomes

XY or XX

#11 Deletion of upper arm





# Aniridia-Wilms Tumor Syndrome

Mentally retarded

Growth retarded

Blindness

Tumors on kidneys

Short lifespan

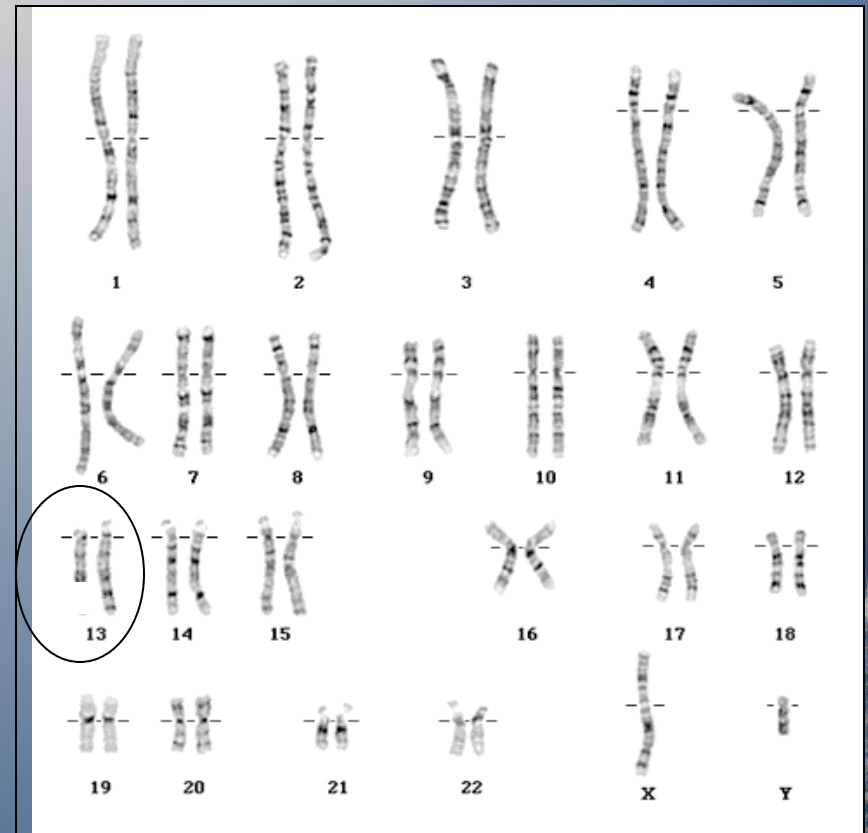


# Thirteen Q Deletion Syndrome

1 in 500,000 births

46 chromosomes  
XY or XX

#13 Deletion of lower arm



# Thirteen Q Deletion Syndrome

Mentally retarded

Deformed face

No thumbs

Heart disease

Short lifespan



# Prader-Willi Syndrome

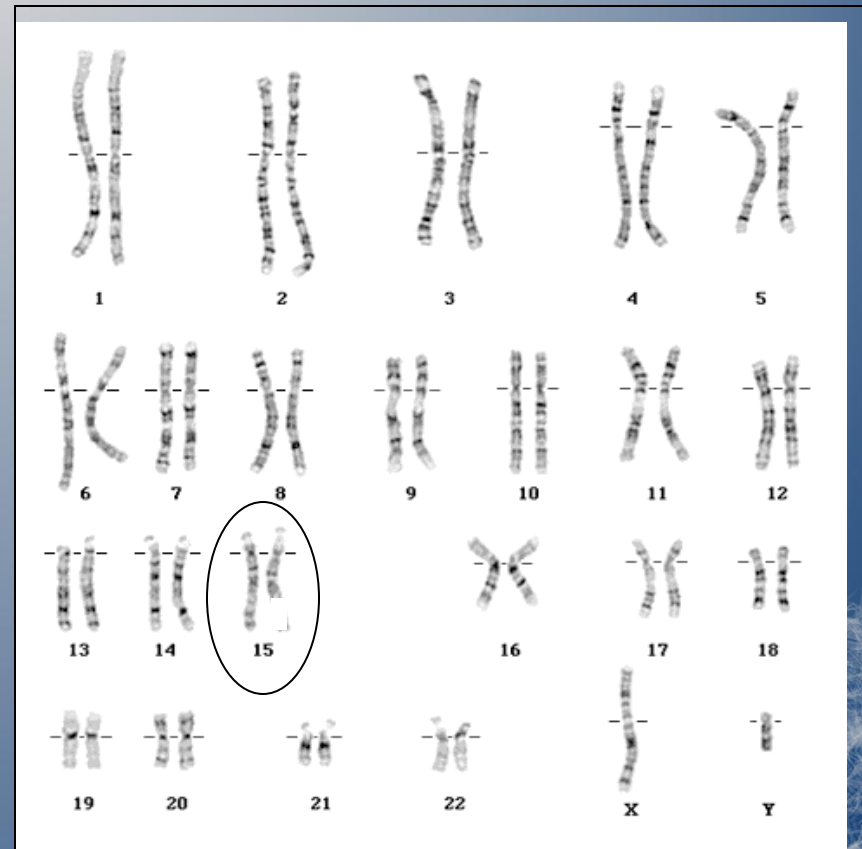
1 in 5,000,000 births

46 chromosomes

XY=97%

XX=3%

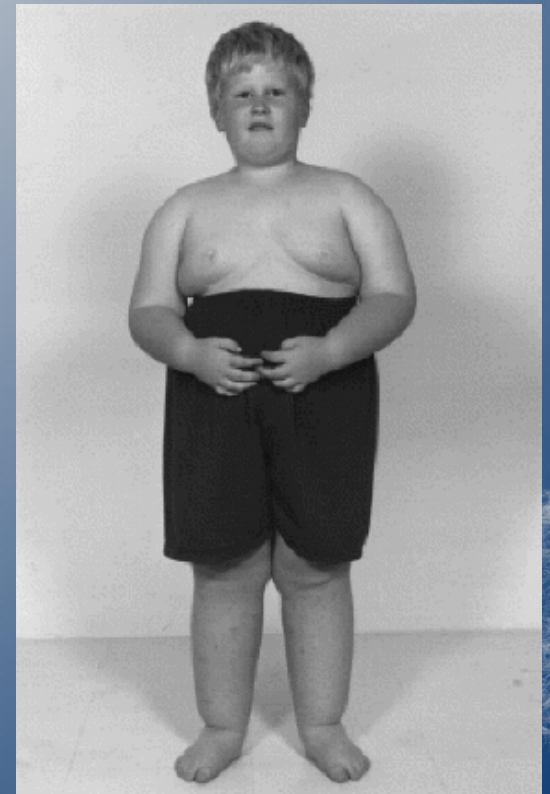
#15 Deletion of lower arm





# Prader-Willi Syndrome

Small bird-like head  
Mentally retarded  
Respiratory problems  
Obesity  
Short lifespan



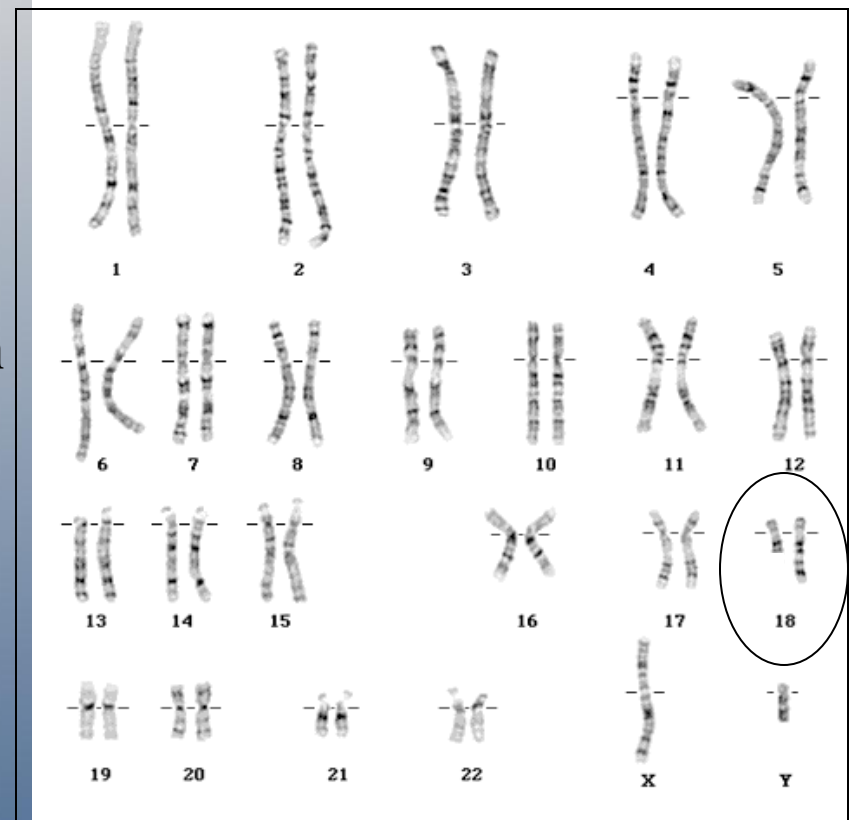
# Eighteen Q Deletion Syndrome

1 in 10,000,000 births

46 chromosomes

XY or XX

#18 Deletion of lower arm



# Eighteen Q Deletion Syndrome

Mentally retarded

Heart disease

Abnormal hands and feet

Large eyes

Large ears

Normal lifespan



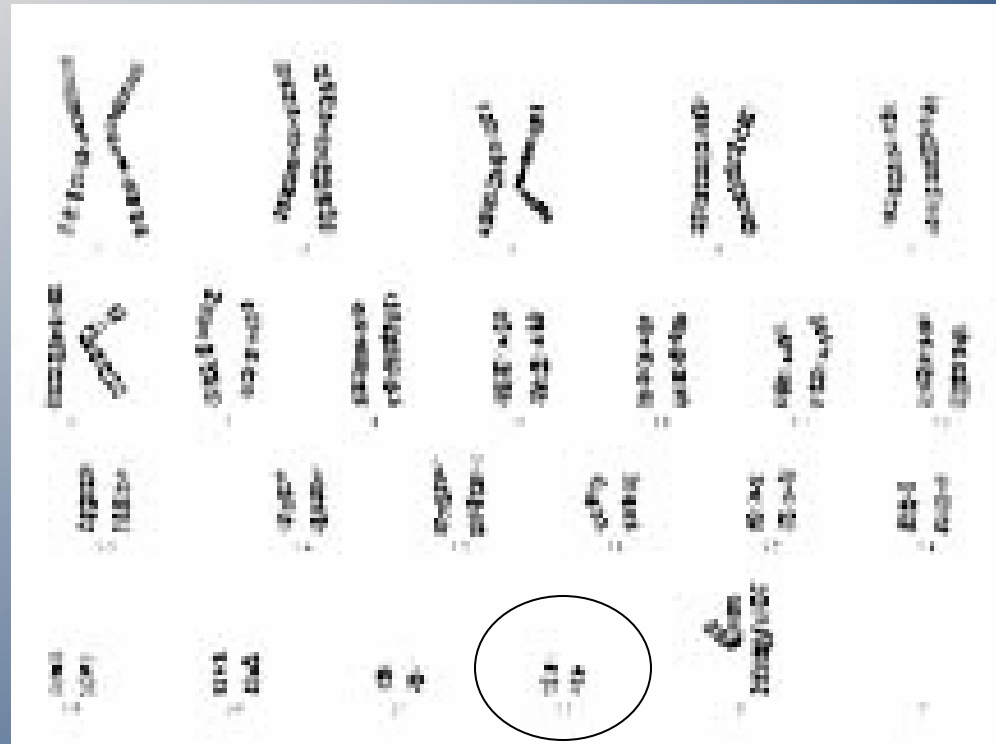
# Cat-Eye Syndrome

**1 in 1,000,000 births**

**46 chromosomes**

**XY or XX**

**#22 Deletion of bottom  
arm**





# Cat-Eye Syndrome

- Fused fingers and toes
- Mentally retarded
- Small jaw
- Heart problems
- Normal lifespan

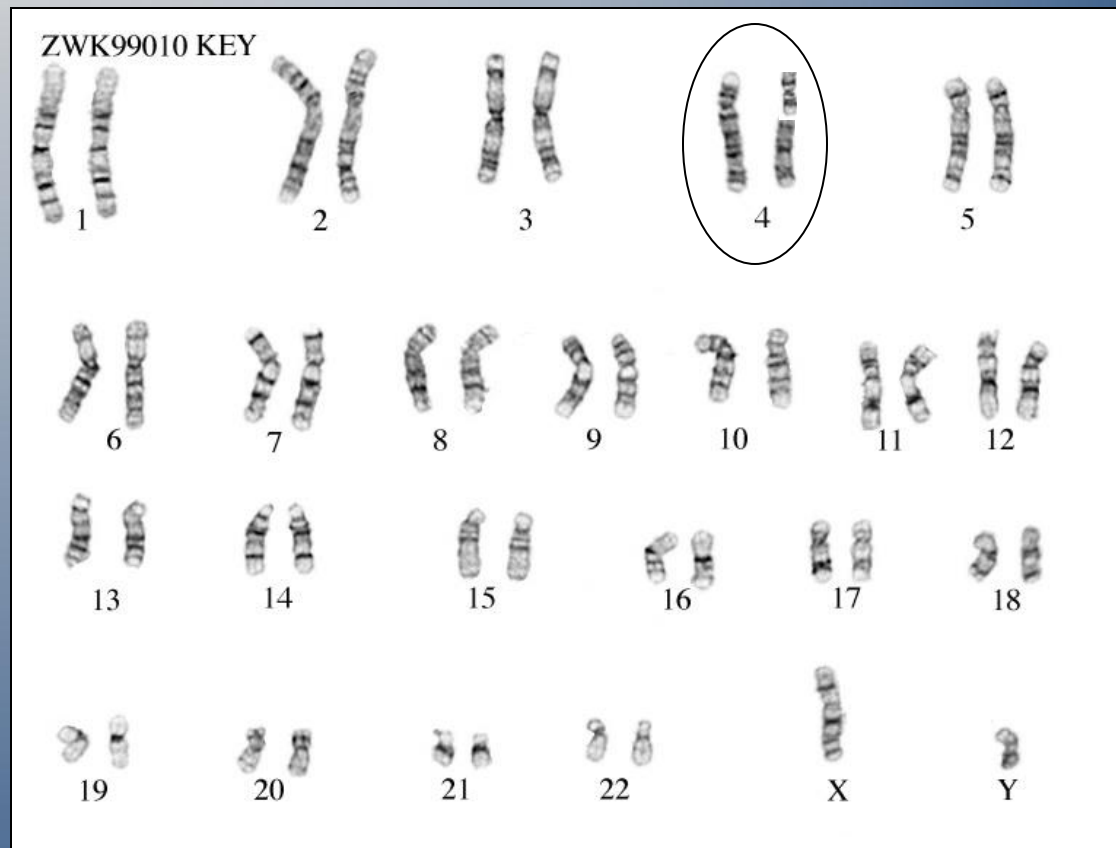


# Four-Ring Syndrome

1 in 10,000,000 births

46 chromosomes  
XY or XX

#4 Inversion



# Four-Ring Syndrome

Cleft palate

Club feet

Testes don't descend

Short lifespan

