Chromosome Analysis

National DNA Day

April 25, 2005

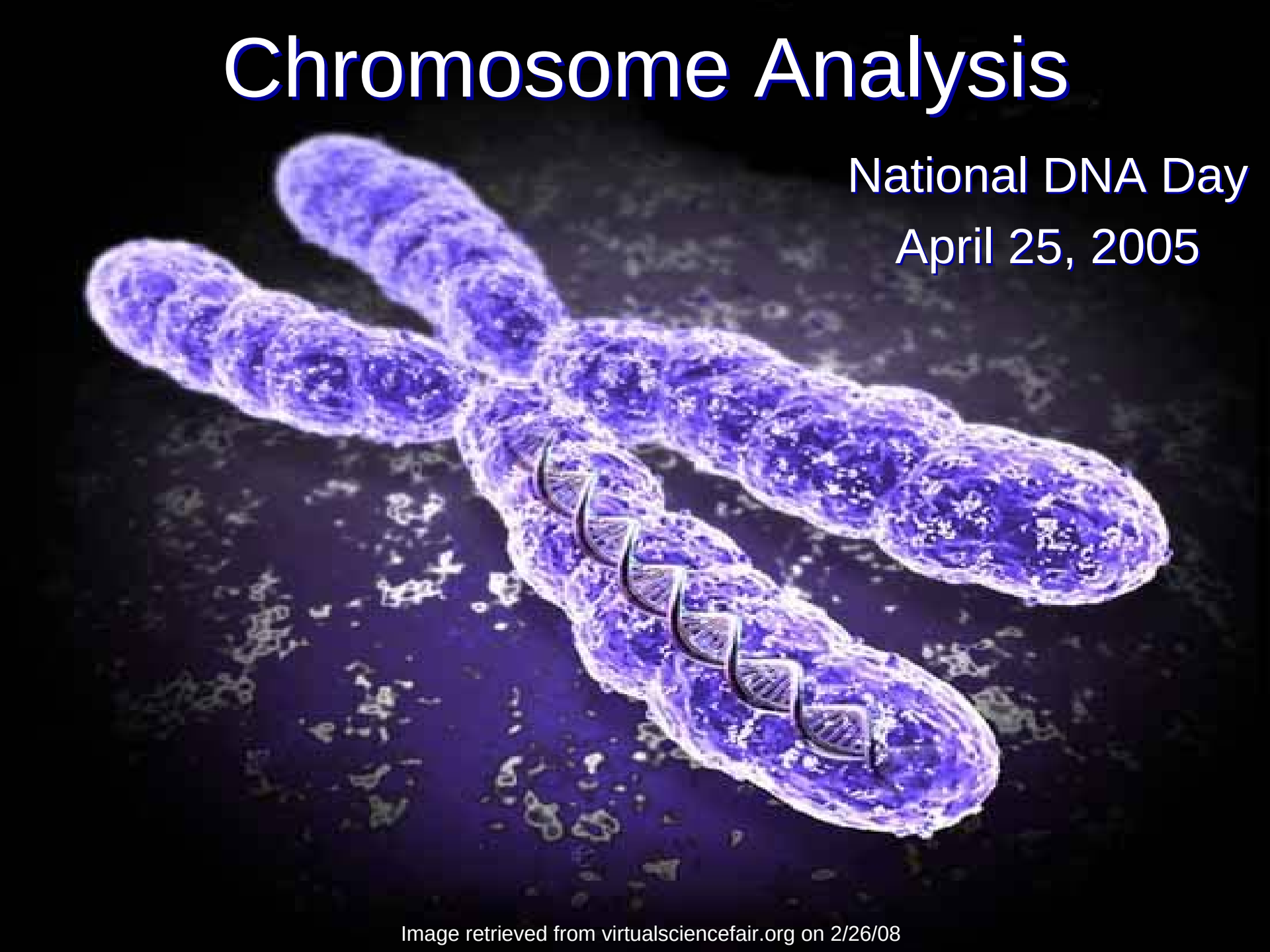


Image retrieved from virtualsciencefair.org on 2/26/08

What are chromosomes?

- Chromosomes are structures found in the nucleus of cells
- Chromosomes carry all of our genes, and therefore all of our genetic information
- Humans have 46 chromosomes, or 23 pairs, to carry our approximately 25,000 genes
  - The first 22 pairs are called *autosomes*
  - The 23rd pair are the sex chromosomes or *gonosomes* – this pair will either be XX or XY

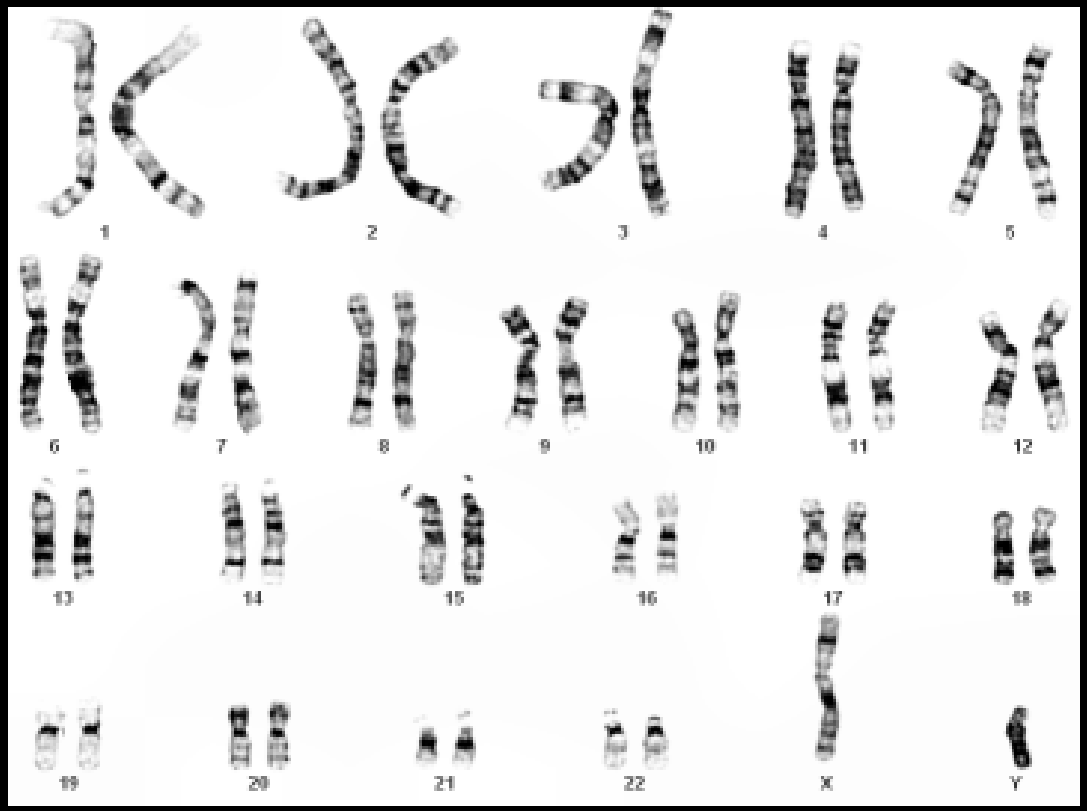
The Karyotype

- A karyotype is a pattern or picture of chromosomes
- The chromosomes are paired and arranged according to size
  - Each chromosome is paired with its *homologous* chromosome – its exact match in size and structure, though the homologous chromosomes may carry different alleles of the same gene
- Then the chromosome pairs are labeled
  - The autosomes are numbered 1 through 22 according to size
- The sex chromosomes are a different story
  - The X and X or the X and Y are paired, then placed at the end, even though they are not necessarily the smallest chromosomes
  - These chromosomes do not receive a number – just XX or XY

Using a Karyotype

- A karyotype allows a cytogeneticist or lab technician to examine the chromosomes and see if there is anything extra or missing, or if the structure of the chromosomes is different than usual
- Sometimes there is an obvious abnormality, but other times it takes very careful observation to spot something

Karyotype Example



- This is an example of a completed karyotype
- Since both an X and a Y chromosome are present, this is a karyotype of a male

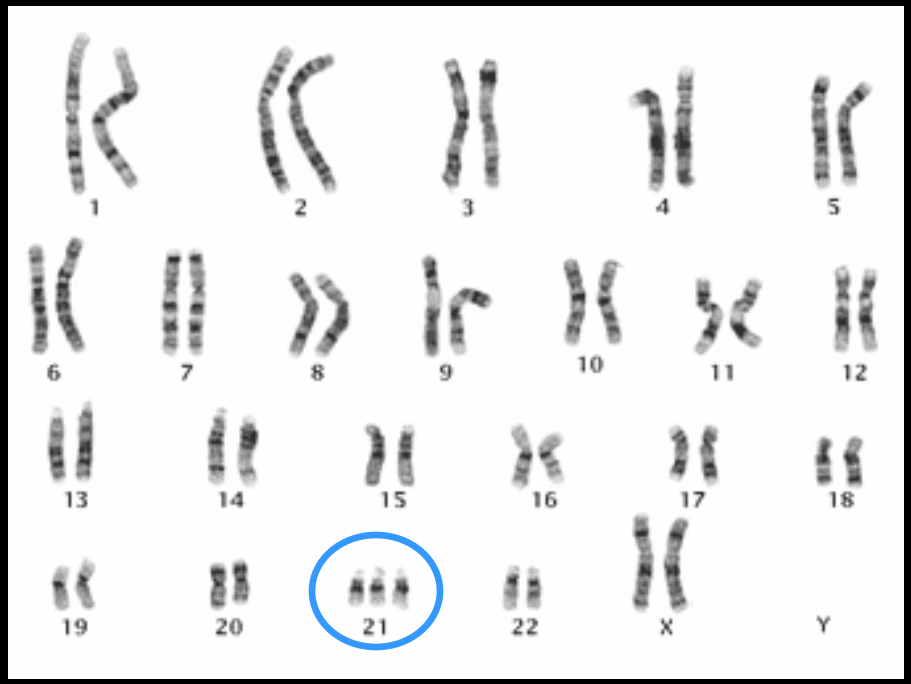
Creating a Karyotype

- Open your envelopes and empty the contents on your desk
- Arrange the chromosomes to create a karyotype
- Chromosomes should all be paired together and arranged from largest to smallest
  - The sex chromosomes should go last, after the other 22 pairs
  - Go ahead and number your chromosome pairs once they are paired and arranged
- Be careful – your karyotype might contain an abnormality!

What kind of results do you see?

Trisomy 21

- Trisomy 21 is the presence of 3 chromosome 21's. Trisomy 21 causes the condition commonly known as Down syndrome
- The extra chromosome leads to the specific characteristics of Down syndrome, some of which are very familiar
  - However, not all individuals with Down syndrome will show the exact same characteristics – there is a great deal of variability

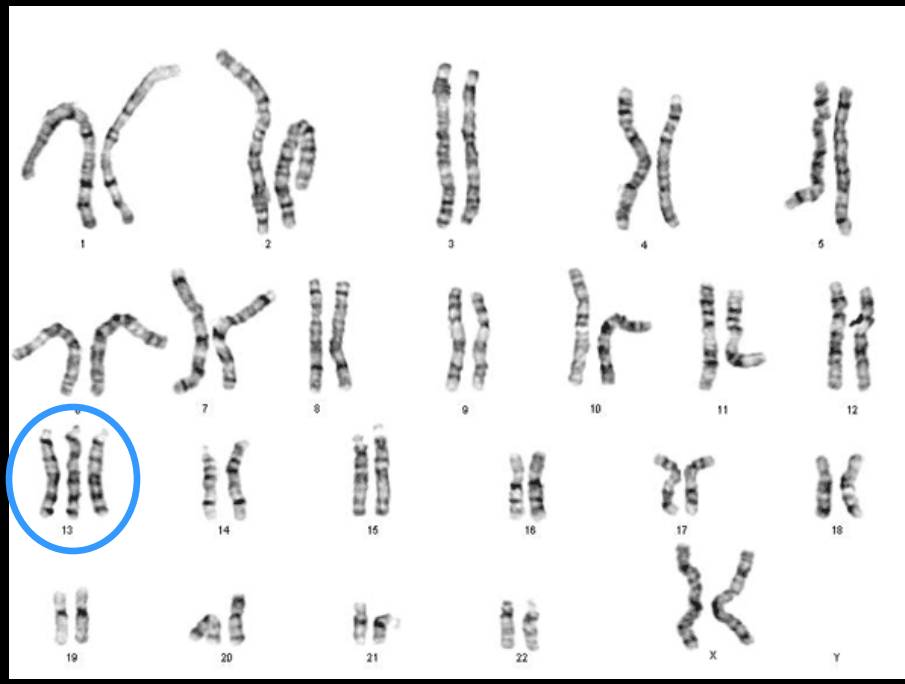


Trisomy 21/Down syndrome

- Individuals with Down syndrome have a typical facial appearance
- All have some degree of mental retardation, but for most it is mild or moderate
  - Can learn to read, write, do some math, and live day-to-day with minimal assistance from others
  - Others require a lot of attention and care
- There are several possible health concerns, including heart problems, hearing loss, feeding concerns, and others
  - Individuals with Down syndrome are now living longer than in the past – into their 50's and 60's
  - We are now discovering that those individuals who survive to this age are at very high risk of developing Alzheimer's disease

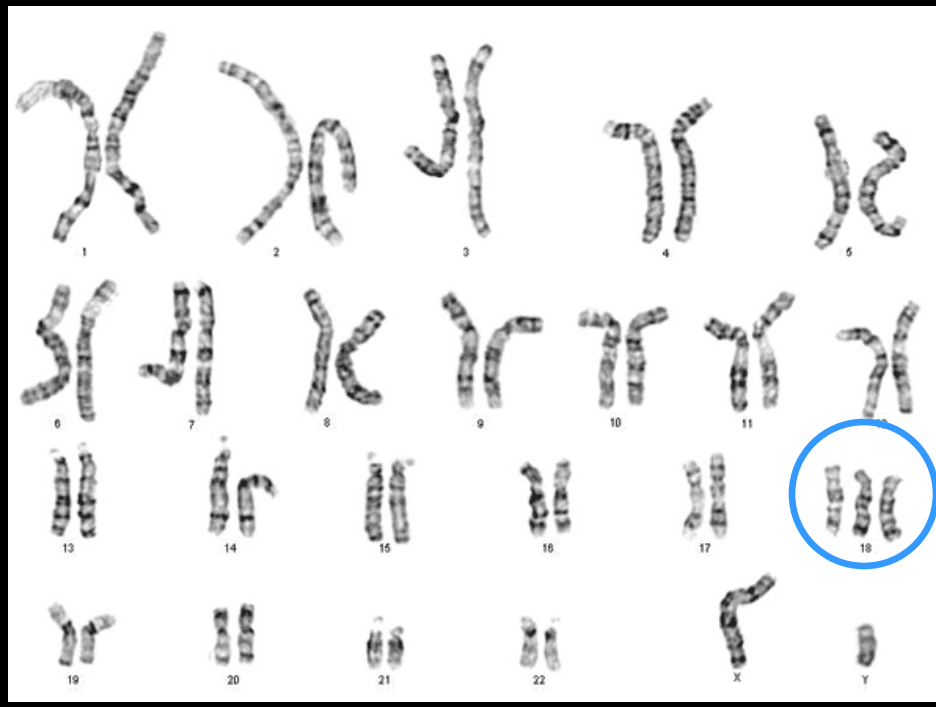
Trisomy 13

- Trisomy 13 is the presence of 3 chromosome 13's. Trisomy 13 causes the condition sometimes known as Patau syndrome
- Trisomy 13 is a very serious condition – only about 5% of babies with the disorder survive past their first year
  - Most pregnancies involving Trisomy 13 end in miscarriage
- Children with Trisomy 13 usually have a lot of trouble breathing, especially when they sleep, and many have seizures. All individuals with Trisomy 13 have severe mental retardation
- There are other characteristics commonly seen in people with Trisomy 13, including a small head, extra fingers and/or toes, and a cleft lip or cleft palate



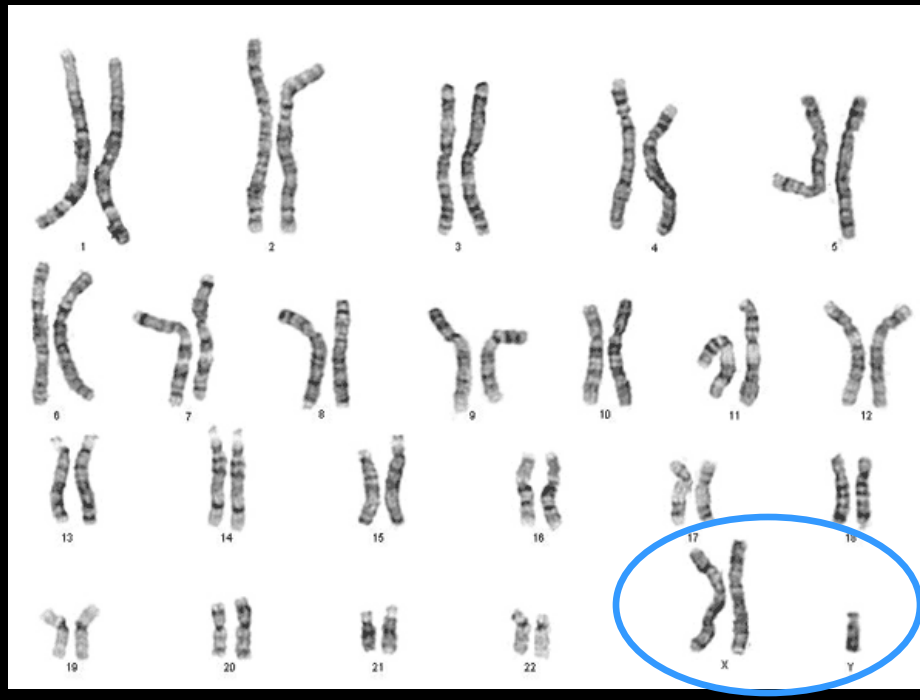
Trisomy 18

- Trisomy 18 is the presence of 3 chromosome 18's. Trisomy 18 causes the condition sometimes known as Edwards syndrome
- Trisomy 18 is another very serious condition – only about 10% of babies with the disorder survive past their first year. A majority of babies who survive are female.
  - Most pregnancies involving Trisomy 18 end in miscarriage
- Children with Trisomy 18 usually have breathing problems, difficulty eating, and many have seizures. Some have serious heart conditions. All individuals with Trisomy 18 have severe mental retardation.
- Most babies with Trisomy 18 are very small and have certain recognizable facial features. They also tend to overlap their fingers in a very distinct pattern



47, XXY

- An individual with the genotype 47, XXY is male
  - The person has 47 chromosomes just like someone with Trisomy 21, 13, or 18, but does not have a typical “trisomy,” as he has two of one chromosome and one of another
  - One could say, though, that such an individual has a sex chromosome trisomy
- The extra X chromosome leads to features of the condition commonly known as Klinefelter syndrome

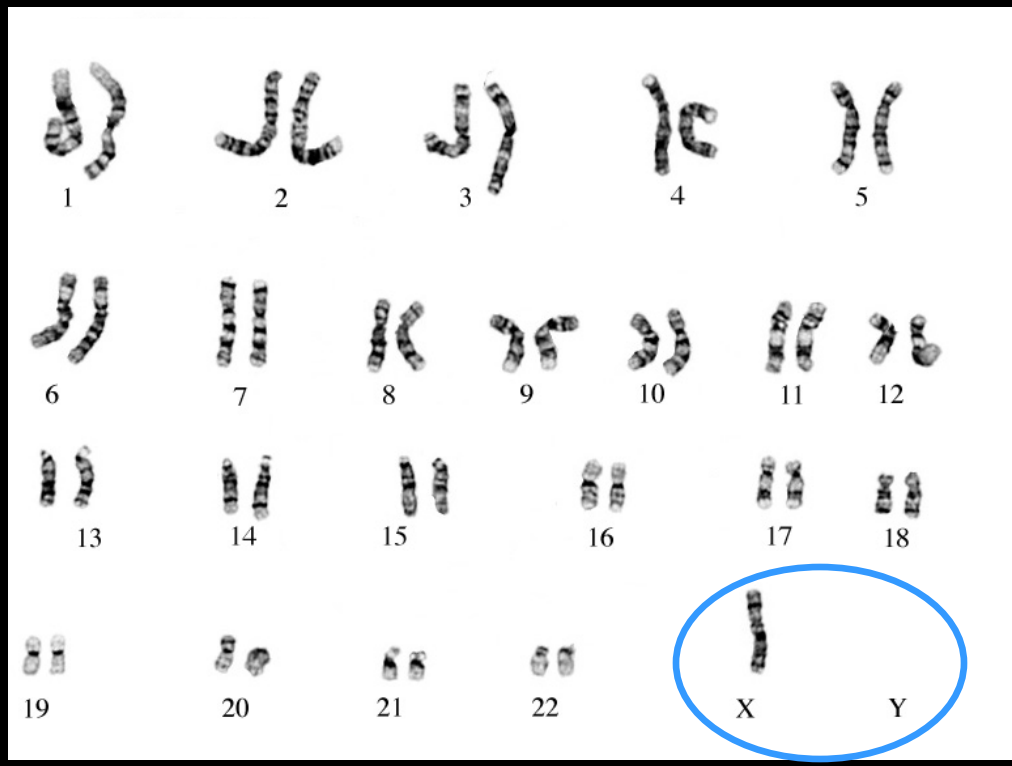


Klinefelter syndrome

- Affects about 1 in 1000 males
- Most males are taller than average and may have a different distribution of body fat (e.g. more than usual in the hips or chest)
  - Also tend to have sparse facial and body hair
- Some have a degree of mental retardation, but many have normal intelligence
- The most common feature of Klinefelter syndrome is infertility
  - It is estimated about 2% of infertile men have Klinefelter's syndrome
- Since the characteristics of the syndrome are not always obvious, many males with Klinefelter will never be diagnosed

45, X

- An individual with the genotype 45, X is phenotypically female
  - The person has 45 chromosomes instead of the usual 46
  - Instead of a trisomy, this would be called a *monosomy*
  - One copy of an X chromosome = monosomy X
  - Monosomy X is the only monosomy known to be compatible with life
- Having only one copy of an X chromosome leads to the features of the condition known as Turner syndrome



Turner syndrome

- Affects about 1 in 5000 newborn females
- Females with Turner syndrome are shorter than average and have other noticeable physical features
  - Swelling of the hands and feet, webbing of the neck, broad chest
  - May also have features which affect their health, including heart disease and a “horseshoe-shaped” kidney
- Individuals with Turner syndrome will not typically have mental retardation, but may have specific learning disabilities

Questions for Discussion

- Why would the presence (or absence) of a chromosome lead to certain characteristics in a person?
- Why wouldn’t the physical, mental, and behavioral features be exactly the same for two people with the same extra (or missing) chromosome?
- Why are babies with trisomy 21 more likely to survive than babies with trisomy 13 or 18?
- Why do you think monosomy X (Turner syndrome) is the only monosomy in which the individual survives?