

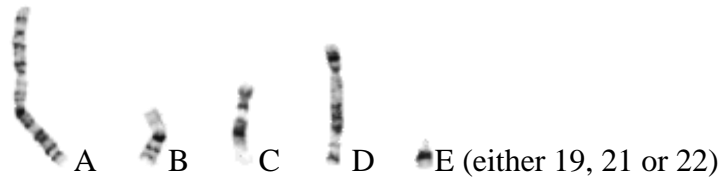
Karyotyping Activity

This exercise is a simulation of human karyotyping using digital images of chromosomes from actual human genetic studies. You will be arranging chromosomes into a completed karyotype, and interpreting your findings just as if you were working in a genetic analysis program at a hospital or clinic.

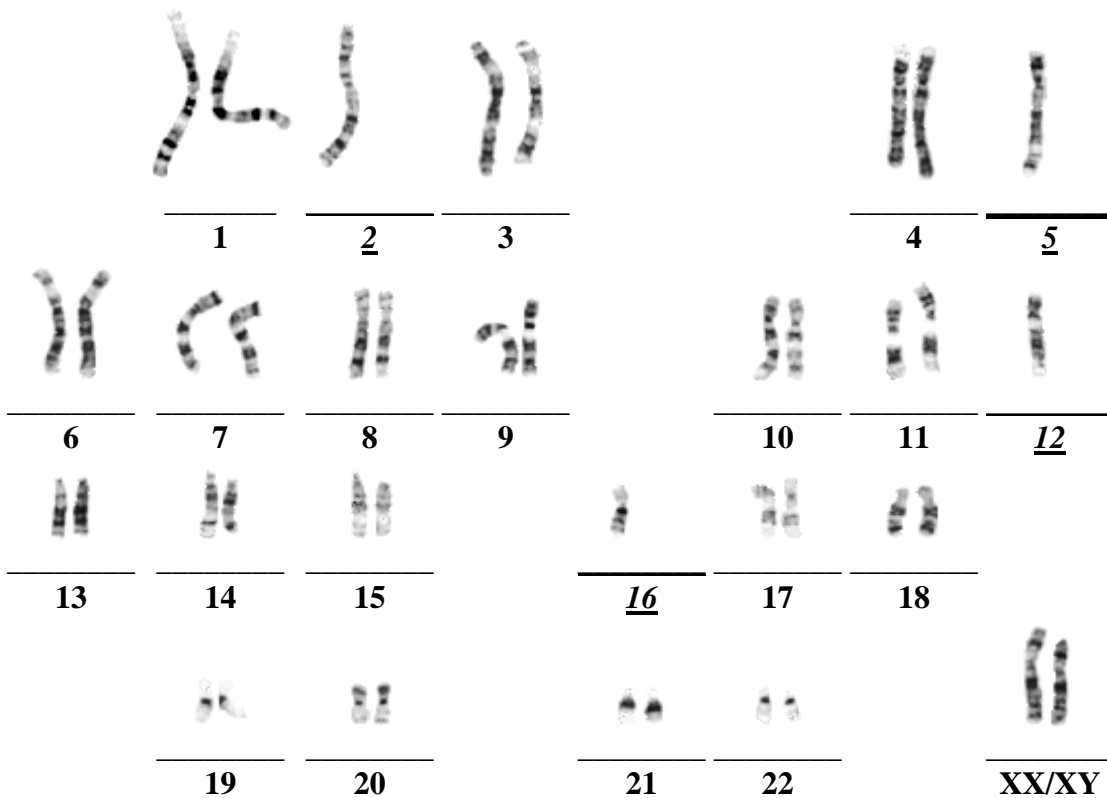
Patient A

Patient A is the nearly-full-term fetus of a forty year old female.

Write the letter of the correct chromosome next to its homologous pair on your own paper for the partially completed karyotype below. (HINT: Some pairs may contain more than 2 chromosomes.)

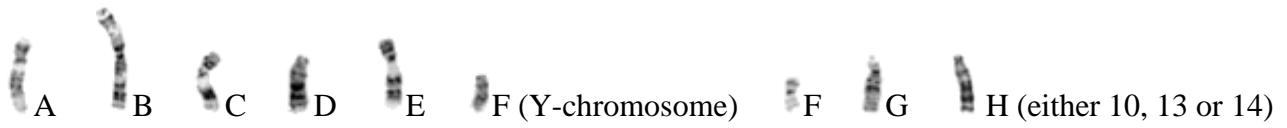


Patient A's Karyotype

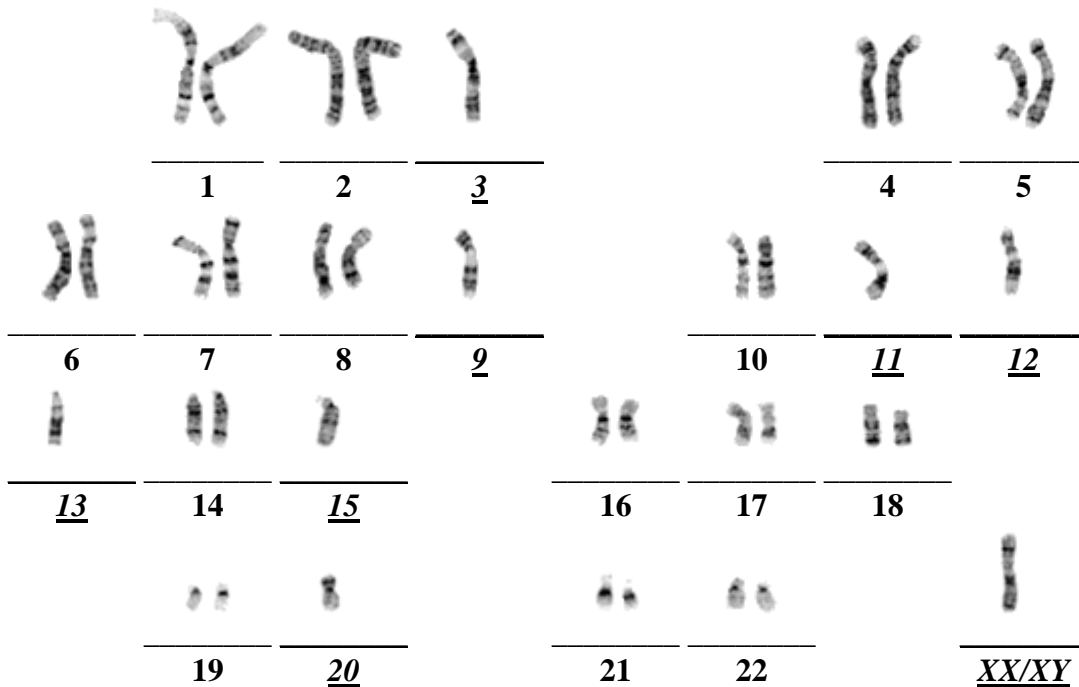


Patient C

Patient C died shortly after birth, with a multitude of anomalies, including polydactyly and a cleft lip. Write the letter of the correct chromosome next to its homologous pair on your own paper for the partially completed karyotype below. (HINT: Some pairs may contain more than 2 chromosomes.)



Patient C's Karyotype



Interpreting the Karyotype

Lab technicians compile karyotypes and then use a specific notation to characterize the karyotype. This notation includes the total number of chromosomes, the sex chromosomes, and any extra or missing autosomal chromosomes. For example, **47, XY, +18** indicates that the patient has 47 chromosomes, is a male, and has an extra autosomal chromosome 18. **46, XX** is a female with a normal number of chromosomes. **47, XXY** is a patient with an extra sex chromosome.

What notation would you use to characterize each patient's karyotype? Draw the chart below on your own paper and complete:

Patient A	Patient B	Patient C

Making a Diagnosis

The next step is to either diagnose or rule out a chromosomal abnormality. In a patient with a normal number of chromosomes, each pair will have only two chromosomes. Having an extra or missing chromosome usually renders a fetus inviable. In cases where the fetus makes it to term, there are unique clinical features depending on which chromosome is affected. Listed below are some syndromes caused by an abnormal number of chromosomes.

Diagnosis	Chromosomal Abnormality
Normal # of chromosomes	Patient's problems are due to something other than an abnormal number of chromosomes.
Klinefelter's Syndrome	One or more extra sex chromosomes (i.e. XXY)
Down Syndrome	Trisomy 21, extra chromosome 21
Patau Syndrome	Trisomy 13, extra chromosome 13

What diagnosis would you give each patient? Draw the chart below on your own paper and complete:

Patient A	Patient B	Patient C