Name _____ Date _____

# Web Quest: Karyotyping Activity

*In this activity, you will use a computer model to look at chromosomes and prepare a karyotype. You will diagnose patients for abnormalities and learn the correct notation for characterizing karyotypes.*

## Site 1 → http://www.biology.arizona.edu/

Click on "**Karyotyping**" under "**Human Biology**" and read the Introduction page:

1. What causes a dark band on the chromosome?

2. What is a centromere?

*Patient Histories: Click on **Patient Histories** button. You will be completing a karyotype for Patient A, B and C.*

**Patient A (Click on the link to "Complete Patient A's Karyotype")**

*Match the chromosome to its homolog. After all the matches are complete you'll analyze your patient. (Scroll down to view your completed karyotype).

1. What is patient A's history? (Summarize it)

2. The 1st chromosome pictures is the homolog to which chromosome? _____
3. The 2nd chromosome pictures is the homolog to which chromosome? _____
4. The 3rd chromosome pictures is the homolog to which chromosome? _____
5. The 4th chromosome pictures is the homolog to which chromosome? _____
6. The 5th chromosome pictures is the homolog to which chromosome? _____
7. How many total chromosomes are in your karyotype? (Count them.) _____
   The last set of chromosomes is the **sex chromosomes**, if you have two large chromosomes, your patient is **XX** (female), while one large and one small indicates a **XY** (male) .

8. What sex chromosomes does your patient have? _____
9. Which chromosome set has an extra **+** _____
10 . What diagnosis would you give this patient? (What disease?) _____

## 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's Syndrome | Trisomy 21, extra chromosome 21 |
| Trisomy 13 Syndrome | extra chromosome 13 |

**Patient B (Click on the link to "Complete Patient B's Karyotype" and repeat the above process.)**

1. What is patient B's history? (Summarize it)

2. The 1st chromosome pictures is the homolog to which chromosome? _____
3. The 2nd chromosome pictures is the homolog to which chromosome? _____
4. The 3rd chromosome pictures is the homolog to which chromosome? _____
5. The 4th chromosome pictures is the homolog to which chromosome? _____
6. The 5th chromosome pictures is the homolog to which chromosome? _____
7. The 6th chromosome pictures is the homolog to which chromosome? _____
8. The 7th chromosome pictures is the homolog to which chromosome? _____
9. The 8th chromosome pictured is chromosome? _____
10. How many total chromosomes are in your karyotype? (Count them.) _____
11. What sex chromosomes does your patient have? _____
12. Which chromosome set has an extra + _____
13 . Finish the notation for this patient's karyotype: **47 X** _____
14. What is the diagnosis? _____

## 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.

**Patient C (Click on the link to "Complete Patient C's Karyotype" and repeat the above process.)**

1. What is patient C's history? (Summarize it)

2. The 1st chromosome pictured is the homolog to which chromosome? _____
3. The 2nd chromosome pictured is the homolog to which chromosome? _____
4. The 3rd chromosome pictured is the homolog to which chromosome? _____
5. The 4th chromosome pictured is the homolog to which chromosome? _____
6. The 5th chromosome pictured is the homolog to which chromosome? _____
7. The 6th chromosome pictured is the homolog to which chromosome? _____
8. The 7th chromosome pictured is the homolog to which chromosome? _____
9. The 8th chromosome pictured is the homolog to which chromosome? _____
10. The 9th chromosome pictured is chromosome? _____
11. How many total chromosomes are in your karyotype? (Count them.) _____
12. What sex chromosomes does your patient have? _____
13. Which chromosome set has an extra + _____
14 . Finish the notation for this patient's karyotype: _____
15. What is the diagnosis? _____

## Site 2 → https://learn.genetics.utah.edu/content/basics/
**(Genetic Science Learning Center)**

Go to "**Basic Genetics**" → then scroll down and click "**How Do Scientists Read Chromosomes**"
Find the answers to the following questions in this area. Browse all sections

1. List and describe the **three** (3) key features used to read chromosomes?

2. Sketch and label the following: **metacentric**, **submetacentric**, **acrocentric**

Click the "**Back/Return"** button when you finished sketching.

3. Go to **MAKE A KARYOTYPE** - Try it yourself - Create your own karyotype - Turning on **hints** is okay. Check this box ☐ when your karyotype is complete. **Have Dr Legaspi initial this activity**: _____

What did you find difficult about matching the chromosomes? _____

4. **Site 3** → https://rarediseases.info.nih.gov/guides/pages/73/faqs-about-chromosome-disorders

What is trisomy?

What is monosomy?

What is a terminal deletion?

5. **Site 4** → https://rarediseases.info.nih.gov/diseases **(Type each into the SEARCH box.)**
For each of the **Disorders**, describe the chromosome abnormality and the symptoms.

| Genetic Condition | Cause | Chromosome # | Phenotypic effects |
|---|---|---|---|
| **Cri Du Chat** | | | |
| **Turner Syndrome** | | | |
| **Klinefelter Syndrome** | | | |
| **Williams Syndrome** | | | |