**Karyotyping Activity**

**Purpose:**

To learn about Karyotypes and how they can be used to identify a variety of chromosomal abnormalities.

You will evaluate 6 patients' Karyotypes and diagnose any missing or extra chromosomes, and describe the characteristics of someone with this Karyotype.

**Procedure:**

Starting at your assigned patient, use the Karyotype given and observe the chromosomes. Fill in your chart for that patient.

If you suspect there is an abnormality, consult the list of syndromes to find which one your patient has. (ALL columns of the chart should be filled out for an abnormal Karyotype.)

If your patient has a normal Karyotype, you only need to fill out the first three columns.

Rotate around to each patient until you have seen all six.

**Conclusion:**

Upon completing your chart, answer the following questions.

1. How many pairs of chromosomes are in a normal Karyotype?
2. What is non-disjunction?
3. What is trisomy?
4. Is it possible for a person to have an abnormal chromosome and still lead a normal life? Explain.
5. In your own words, explain how doctors and geneticists can use Karyotyping to diagnose certain syndromes.

**Patient #1**



**Patient #2**

**Patient #3**

**Patient #4**



**Patient #5**



**Patient #6**

**Diseases and Abnormalities**

**Down Syndrome**

* Caused by an extra copy of the 21st chromosome
* They have moderate to severe intellectual disability
* Physical characteristics: a flat face, a small broad nose, abnormally shaped ears, a large tongue, and upward-slanting eyes with small folds of skin in the corners
* Medical symptoms: increased risk of developing respiratory infections, blockages in the digestive tract, leukemia, heart defects, hearing loss, hypothyroidism, and eye abnormalities

**Klinefelter’s Syndrome**

* Caused by an extra X chromosome
* Affects sexual development
* Physical characteristics: small testes, taller than average, low muscle tone, long arms and legs, narrow shoulders, wider hips, less body hair
* Medical symptoms: More likely to develop osteoporosis, Diabetes II, varicose veins, infertility

**Patau Syndrome**

* Caused by an extra copy of the 13th chromosome
* Most babies born with this syndrome die within weeks or months of birth
* Physical characteristics: very small or poorly developed eyes, extra fingers or toes, cleft lip or palate, and weak muscle tone
* Medical symptoms: Heart defects, brain or spinal cord abnormalities, various other life-threatening medical issues

**Turner’s Syndrome**

* Caused by a missing or incomplete X chromosome
* Affects growth and sexual development
* Ovaries do not properly develop, and therefore most women are infertile
* Physical characteristics: shorter than average height, stocky build, receding lower jaw, short, webbed neck
* Medical symptoms: can have high blood pressure, heart and kidney defects, and swelling of hands and feet

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| **Patient #** | **Normal or Abnormal** | **Male****or Female** | **What chromosome is affected** | **Diagnosis** | **List the characteristics of a person with this chromosomal abnormality** |
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