Karyotypes and Nondisjunction

Date ______________

Karyotypes
• A picture of all of the chromosomes in a cell.
• Where can I get a karyotype?
  – Genetic counseling centers, such as the Greenwood Genetic Center
• How do they do it?
  – They use a chemical to halt the cell cycle during metaphase, when the chromosomes are two sister chromatids held together at the centromere.
  – They extract the genetic material from the nucleus, stain it, and arrange each chromosome by size.

Why should people get karyotypes?
• They can show large-scale chromosomal abnormalities, like deletions or extra copies of chromosomes.
• Downside – only shows large changes, and small mutations (like sickle-cell anemia, color-blindness, or progeria) will not show up.

Expressing Karyotypes
• Normal karyotypes are expressed as follows:
  – Females: 46,XX
  – Males: 46,XY
• Can you figure out how we've expressed these?

Chromosomal Disorders
• Usually arise from a nondisjunction during meiosis
  – Nondisjunction: incorrect separation of chromosomes; leads to too many copies of a chromosome or not enough.
  – Remember: there should be TWO of every chromosome!
• Includes trisomy and monosomy
  – Trisomy: three copies of a chromosome
  – Monosomy: one copy of a chromosome
Disorders

- Down Syndrome: 47,XX(+21) or 47,XY(+21) (1 in 800 births)
- Cri-du-chat Syndrome: 46,XX(del5) or 46,XY(del5) (1 in 50,000 births)
- Turner Syndrome: 45,XO
- Klinefelter Syndrome: 47,XXY
- Edward Syndrome: 47,XX(+18) or 47,XY(+18) (1 in 8,000 births)
- Patau Syndrome: 47,XX(+13) or 47,XY(+13) (1 in 20,000 births)
- XXX syndrome: 47,XXX (1 in 1200 births)
- XYY syndrome: 47,XYY

Focus Questions

1. What is a karyotype?
2. How do we express a normal karyotype for a male and female?
3. What is nondisjunction?
4. What is the difference between trisomy and monosomy?
5. Be able to express karyotypes of various disorders, based on descriptions!