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HST.161 Molecular Biology and Genetics in Modern Medicine Fall 2007

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Assignment (part 1): Identify the chromosome abnormalities

Instructions: Each of the karyograms below has a chromosome abnormality. Identify it and answer each question for each karyogram.

- A) What is the chromosome abnormality. Try to write it in correct cytogenetic nomenclature (no points off if you get the nomenclature wrong).
- B) Would you expect the patient to be able to live with this abnormality?
- C) Is this abnormality consistent with a known syndrome? If so, what is it?
- D) In a sentence or two, describe the clinical phenotype (if any) you would expect this patient to have.

Note: ✓ have been placed where the chromosomes are NORMAL but may look funny because of cross-overs or unusual bending. Ignore these spots – they are NOT the abnormality you are looking for.

Hint: None of these abnormalities are subtle.

Slides removed due to copyright restrictions. Normal and abnormal karyotypes.

Assignment (part 2): Cutting chromosomes

Use the template on the next page to place chromosomes.

Use the examples of normal karyograms on previous pages to help you identify chromosome number and orientation.

The next few pages have normal metaphase spreads from a single cell each.

Pick a metaphase to: 1) print out, 2) cut out the chromosomes,3) tape or glue the cut chromosomes onto the template in the proper order and orientation. Label your karyogram with it's ID# (found in the lower left hand corner)

If two chromosomes overlap in a metaphase, print that page twice and cut out each over-lapped chromosome separately.

N.B. Ignore the big dark blobs.

You are only required to do one cell, but feel free to do more than one!

Slides removed due to copyright restrictions. Images of the chromosomes of a single cell during metaphase.