Karyotyping ELE 482, Biomedical Engineering Seminar, February 9, 2004 Tanyn Boulay University of Rhode Island Kingston, RI 02881

Karyotyping is the classification the chromosome complement of an organism or a species according to the arrangement, number, size, shape, or other characteristics of the chromosomes. The chromosome is strand of DNA that carries the genes and functions in the transmission of hereditary information. Karyotyping is useful for detecting certain genetic disorders and for studying evolutionary relationships and mechanisms.

Traditional karyotyping allows scientists to view the full set of human chromosomes in black and white, a technique that is useful for observing the number, size and shape of the chromosomes. This requires an expert, who might need hours to examine a single chromosome.

The chromosomes are obtained by having blood drawn. Preparation for the drawing depends on the person's age and previous experience.

Other kinds of karyotyping:

- 1. Spectral Karyotyping (SKY)
- 2. Fluorescence In Situ Hybridization (FISH)
- 3. Color Changing Karyotyping (CCK)
- 4. Comparative Genomic Hybridization (CGH)

SKY is a laboratory technique that allows scientists to visualize all of the human chromosomes at one time by "painting" each pair of chromosomes in a different fluorescent color.

FISH provides researchers with a way to visualize and map the genetic

material in an individual's cells, including specific genes or portions of genes. Unlike most other techniques used to study chromosomes, FISH does not have to be performed on cells that are actively dividing.

Multicolor karyotyping procedures, such as multiplex FISH (M-FISH), spectral karyotyping (SKY) or color changing karyotyping (CCK) allow identification of all chromosomes in a metaphase preparation (46 chromosomes in humans) based on color differences. They are possible because of the combinatorial labeling procedure.

Comparative genomic hybridization (CGH) is a fluorescent molecular cytogenetic technique that identifies DNA gains, losses, and amplifications, mapping these variations to normal metaphase chromosomes.

SKY Analysis \$100 - \$500. The \$100 rate is only available for staff whose work is deemed a joint project with the SKY/Fish Facility. \$500 - All other service requests.

FISH Analysis (FISH-1) - \$100 - \$500 this rate is only available for staff whose work can be deemed a joint project with the SKY/FISH Facility. Probes of your interest (genes cloned in cosmid, plasmid, YAC, BAC, PAC, etc) have to be prepared by the user. \$500 - All other service requests.

Abnormal results could mean Down Syndrome, Klinefelter syndrome, Turner's syndrome, Trisomy 18, Philadelphia chromosome, and other abnormalities