

KAPITAŁ LUDZKI
NARODOWA STRATEGIA SPÓJNOŚCI

UNIA EUROPEJSKA
EUROPEJSKI FUNDUSZ ROZWOJOWY

"PSP - Uniwersytet Józefo - unijogermosomomem kullaterra w Uniwersytecie Góralim proiec wogólny z Uniwersytelem Wrocław Dowlent"

Projekt współfinansowany ze środków Unii Europejskiej w ramach Europejskiego Funduszu Społecznego

Introduction to Human Genetics

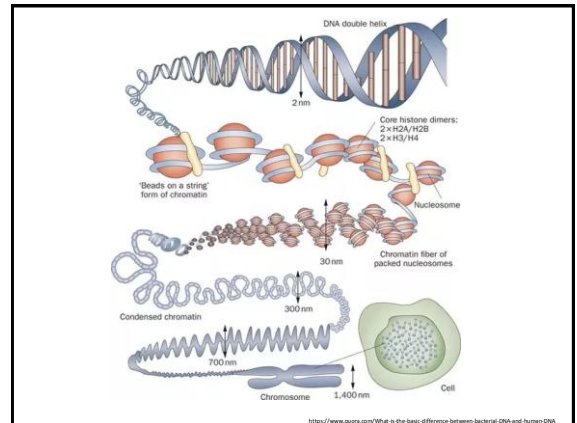
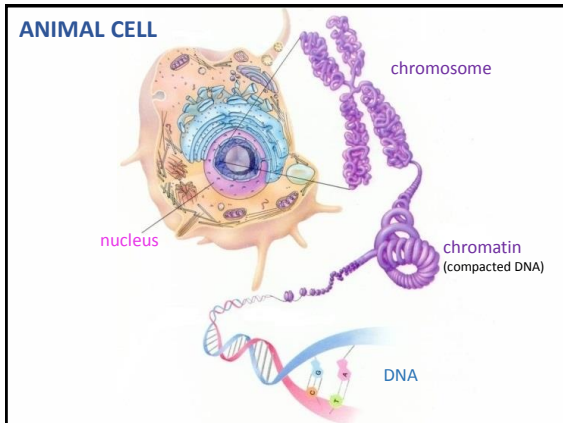
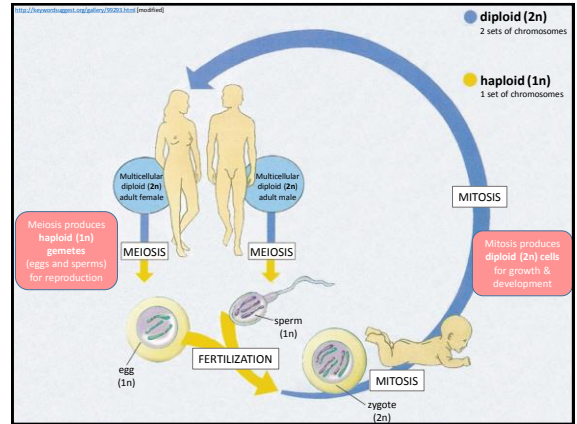
Module 2 Cytogenetics

Human chromosome set

UNIWERSYTET GDAŃSKI

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Katedra Biologii Molekularnej



CYTOGENETICS

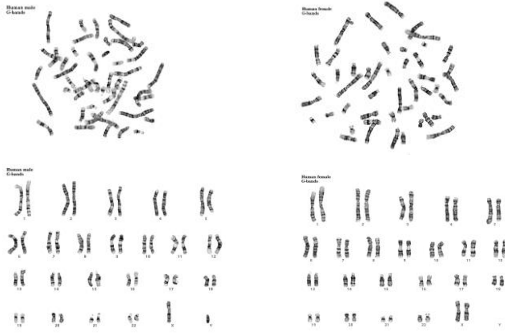
The study of the genetic constitution of cells through the visualisation and analysis of chromosomes

<http://www.well-on.de/cytogenetics-fish-workstation>

INDICATIONS FOR CYTOGENETIC ANALYSIS

- clinical features of a particular syndrome or aberration
- serious mental or physical developmental problems
- ambiguous genitalia/indeterminate gender
- delayed puberty or inappropriate secondary sexual development
- clinically significant abnormal growth
- family history of chromosome abnormality
- infertility of unknown etiology
- couples with a history of multiple spontaneous abortions of unknown cause
- malignant or premalignant disease
- prenatal diagnosis

Normal Human Karyotypes



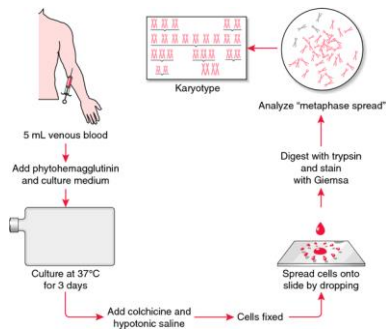
<http://www.pathology.washington.edu/genetics/Cytogenetics/normal.php?file=human%20karyotypes>

PREPARATION OF KARYOTYPE

Sources of cells

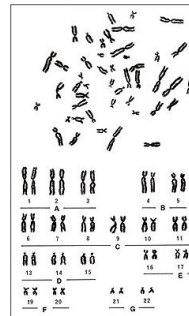
- peripheral blood,
- bone marrow,
- cultured fibroblasts (usually obtained from a skin biopsy)
- amniotic fluid,
- trophoblastic cells from the chorionic villus,

PREPARATION OF KARYOTYPE

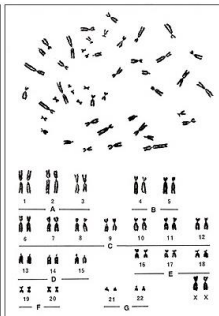


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normal male

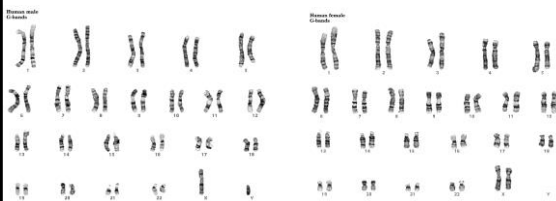


normal female



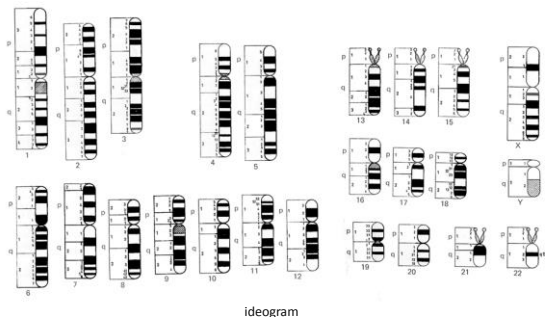
<http://www.ncbi.nlm.nih.gov/pmc/articles/PMC444444/>

Chromosome-banding pattern



<http://www.pathology.washington.edu/genetics/Cytogenetics/normal.php?file=human%20karyotypes>

Chromosome-banding pattern



ideogram

MODULE 2
Lecture 5.2 WEB RESOURCES

Activity 2

Online Resource: NCBI Map Viewer

- Find the hereditary hemochromatosis gene (*HFE*) on a chromosome map.

What is a locus?

The locus for a particular gene describes the region of a chromosome where that gene can be found. For the **6p21.3** locus: **6** is the chromosome number; **p** indicates the short arm of the chromosome, and **21.3** is the number assigned to a particular band or region on a chromosome. When chromosomes are stained in the lab, light and dark bands appear, and each band is numbered. The higher the number, the farther away the band is from the centromere. A locus containing **q** is found on the long arm of a chromosome.

Short and Long Arms of a Chromosome

