


**KAPITAŁ LUDZKI**  
 NARODOWA STRATEGIA ROZWOJU

Unia Europejska  
 Unie Europejskie  
 FUNDUSZ SPÓŁCZESKI

"PRO: Uniwersytet Adama Mickiewicza w Poznaniu (poprzez wsparcie z Uniwersytetem Houston-Dowtown)"  
 Projekt współfinansowany ze środków Unii Europejskiej w ramach Europejskiego Funduszu Społecznego

## Introduction to Human Genetics

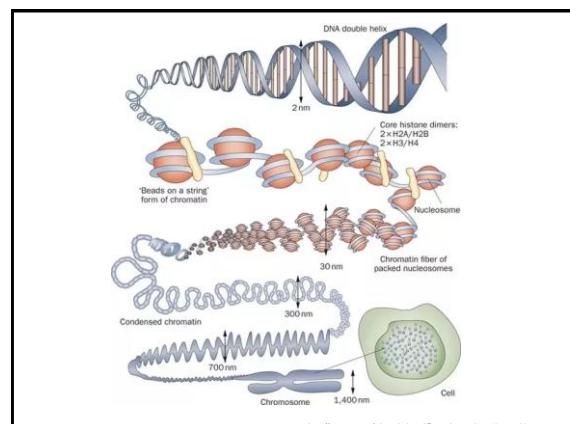
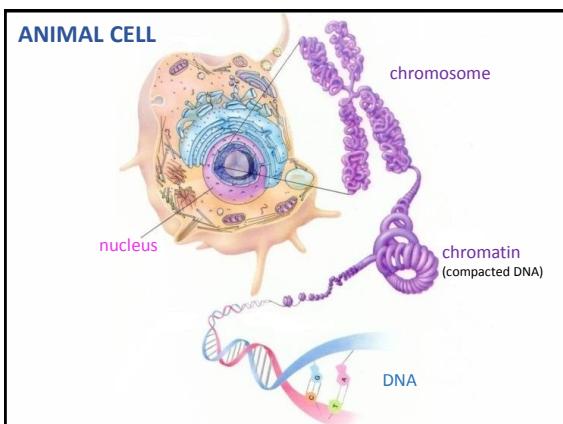
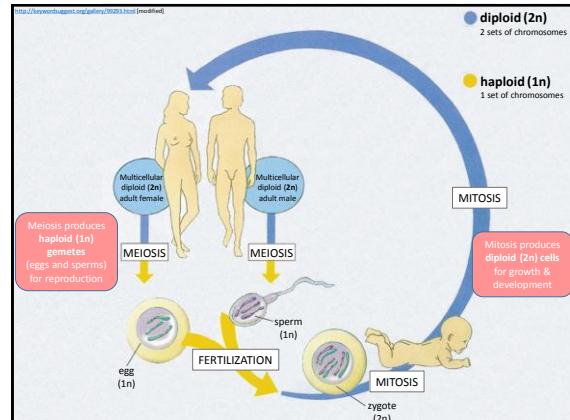
### Module 2 Cytogenetics

## Human chromosome set


**UNIWERSYTET GDAŃSKI**

Dr Ewa Piotrowska  
 Department of Molecular Biology


**KBM**  
 KATEDRA  
 BIOTECHNOLOGII MOLEKULARNEJ



## CYTOGENETICS

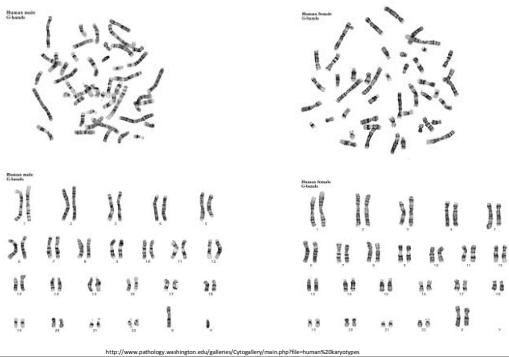
The study of the genetic constitution of cells through the visualisation and analysis of chromosomes

<http://www.well.ox.ac.uk/olympus-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



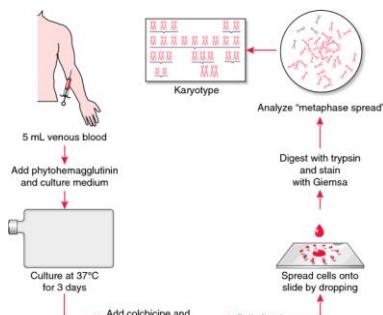
<http://www.pathology.washington.edu/galleries/Category/main.php?file=human%20karyotypes>

## PREPARATION OF KARYOTYPE

### Sources of cells

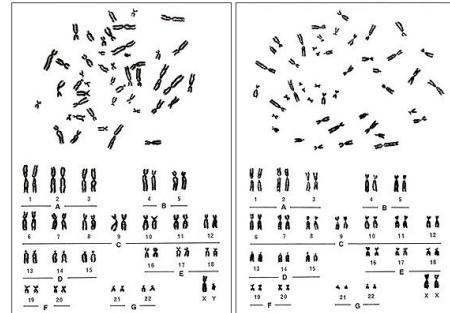
- peripheral blood,
- bone marrow,
- cultured fibroblasts (usually obtained from a skin biopsy)
- amniotic fluid,
- trophoblastic cells from the chorionic villus,

## PREPARATION OF KARYOTYPE



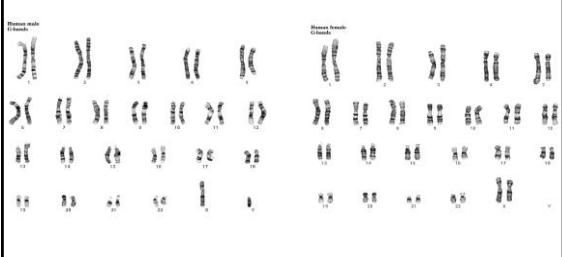
Millan-Kane Encyclopedia and Dictionary of Medicine, Nursing, and Allied Health, Seventh Edition. © 2003 by Saunders, an imprint of Elsevier, Inc. All rights reserved.

### normal male      normal female



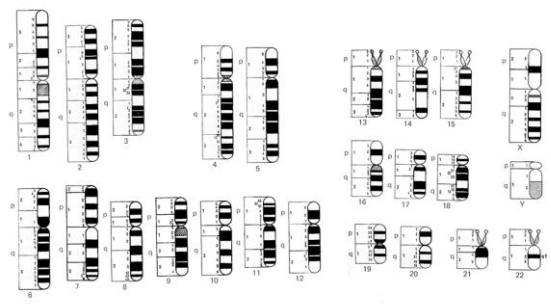
[http://www.ncbi.nlm.nih.gov/entrez/Genomes/geneinfo\\_A.html](http://www.ncbi.nlm.nih.gov/entrez/Genomes/geneinfo_A.html)

## Chromosome-banding pattern



<http://www.pathology.washington.edu/galleries/Category/main.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.

