

KAPITAŁ LUDZKI  
NARODOWA STRATEGIA SPÓJNOŚCI

UNIA EUROPEJSKA  
Europejski Fundusz Społeczny

"Projekt Uniwersytetu Gdańskiego, Uniwersyteckiego Instytutu i Uniwersyteckiego Centrum Badawczego w ramach Europejskiego Funduszu Społecznego"

# Introduction to Human Genetics

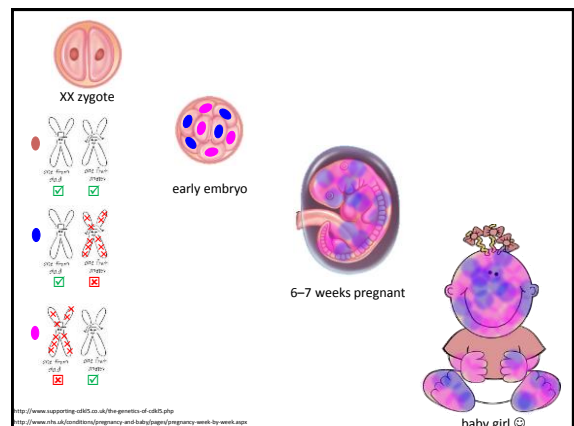
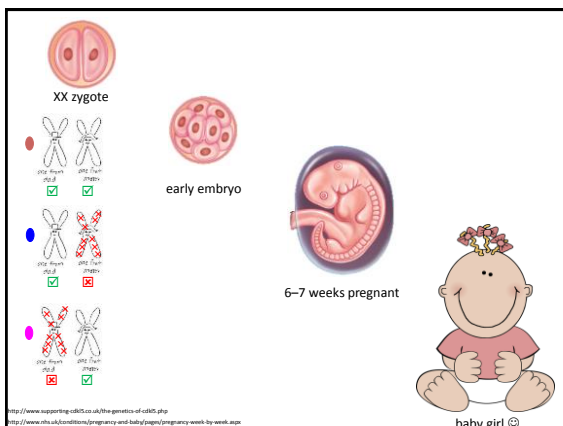
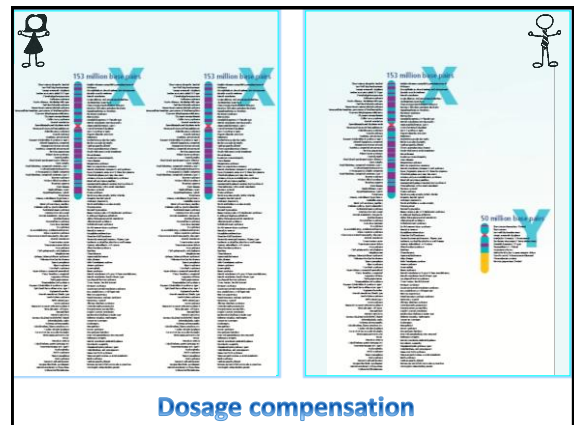
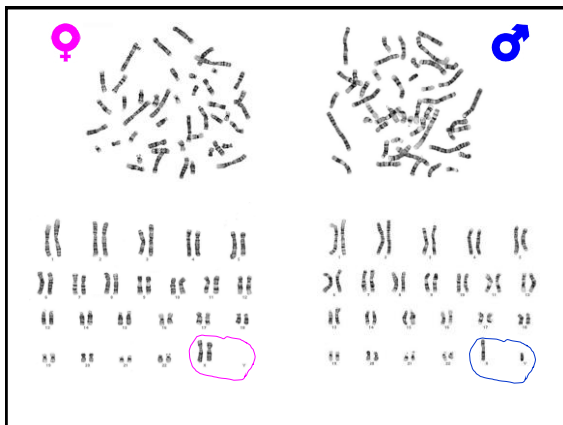
## Module 2 Cytogenetics

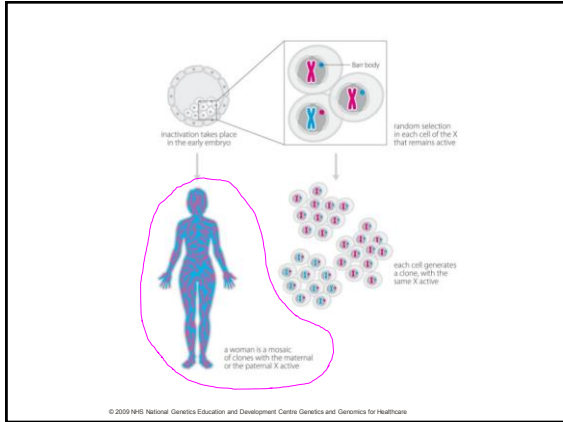
# X chromosome inactivation

Dr Ewa Piotrowska  
Department of Molecular Biology

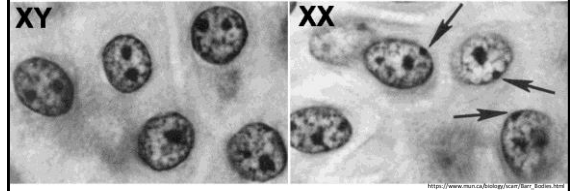
Chromosome	Length (mm)	Base pairs	Variations	Protein-coding genes	Pseudogenes
1	85	248,504,422	12,151,146	2,056	1,220
2	83	242,193,529	12,945,965	1,309	1,023
3	67	198,295,558	10,637,115	1,078	763
4	65	186,214,555	10,165,685	752	727
5	62	181,530,259	9,519,995	876	721
6	58	170,805,879	9,130,476	1,048	801
7	54	159,345,873	8,613,268	989	885
8	50	145,138,336	8,221,520	677	613
9	48	138,394,717	6,939,811	796	661
10	46	133,797,422	7,223,944	733	565
11	46	135,089,222	7,933,370	1,294	821
12	45	133,275,309	7,291,139	1,034	617
13	39	114,364,329	5,082,574	327	372
14	36	107,043,719	4,989,950	830	523
15	35	101,991,189	4,510,706	613	510
16	31	903,383,465	5,101,702	873	465
17	28	83,257,441	4,614,972	1,197	531
18	27	80,373,285	4,035,966	270	247
19	20	58,617,616	3,858,269	1,472	512
20	21	64,444,167	3,439,621	544	249
21	16	46,709,983	2,049,967	234	185
22	17	50,819,698	21,353,111	489	524
X	63	159,040,895	27,538,881	842	874
Y	20	57,227,416	21,194,431	21	388
mtDNA	0.0004	16,569	328	13	0
total		3,082,961,401	155,630,445	20,412	14,600

[https://en.wikipedia.org/wiki/Human\\_genome](https://en.wikipedia.org/wiki/Human_genome)





## Barr bodies



Barr bodies represent the inactive X chromosome and are normally found only in female somatic cells



## Hypohidrotic Ectodermal Dysplasia (HED)



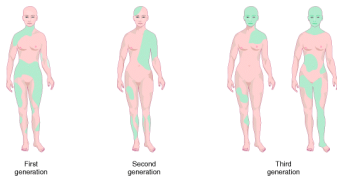
This child with HED has hypodontia with conical teeth, which is often the clinical finding that leads to the diagnosis.

<http://www.orisatlas.com/dermatology/genetic/hypohidrotic-ectodermal-dysplasia/article/20307/>

## Hypohidrotic Ectodermal Dysplasia (HED)

### UNAFFECTED SKIN

- X with the mutated allele was inactivated,
- normal allele present on active X is expressed



### AFFECTED SKIN WITH NO NORMAL SWEAT GLANDS

- X with normal allele was condensed,
- the mutated allele present on active X is being transcribed