# Course overview

**DAY 1 : KUL 13.10.2023**

**The human genome and the chromosomal basis of heredity** (Prof. Thierry Voet)

- Basic concepts
- Introduction: The chromosome
- The 3D nucleus
- Mitosis
- Meiosis

**The human genome: gene structure and function** (Prof. Jeroen Breckpot)

Gene structure and function
Gene regulation and activity

**Next Generation Sequencing** (Dr. Wouter Bossuyt)

- Different platforms
- Main applications

**Copy Number variation** (Prof. Joris Vermeesch)

- Tools for the study of copy number variation
- Clinical consequences of copy number variation

**DAY 2 : ULB 10.11.2023**

**Aud F.2-104, Campus Erasme, 808 Lennik St, 1070 Brussels**

**Patterns of single gene inheritance** (Marc Abramowicz)

Monofactorial genetic diseases. Penetrance, expressivity. Dominant, recessive, and X-linked inheritance, and why these models are approximations. Incidence and prevalence. Pseudoautosomal inheritance. Lyonisation (partly). Mitochondrial inheritance (partly). Anticipation. New mutations. Germ-line and soma. Somatic mosaics, germ-line mosaics. Pedigrees. Intrafamilial and interfamilial variation. Genetic background.

Consanguinity, chances of homozygosity by descent. Consanguinity as common traditional practice. Inbred communities. Pseudodominance. Founder effect, overdominance. Uniparental disomy.

**Genetic variation in individuals and populations: mutation and polymorphism** (Marc Abramowicz)

The life and death of a highly penetrant mutation (hemophilia). Wild type alleles, mutated alleles. The concept of neutral polymorphisms and minor mutations. Blood groups as an example of polymorphism. SNPs. CNVs. Hardy-Weinberg equilibrium, and factors that disturb it. Metapopulation. Mutations and diversity by change. Meiosis and diversity by assembly. Biological fitness. Positive and negative selection. Divergence and diversity.

The history of DF508. Most recent common ancestors. Coalescence. A schematic overview of the ascent of man. Gene tree, species tree. Trans-species polymorphisms. The "common disease, common variant" hypothesis. Haplotype blocks. HapMap. Effective population size. The special case of the Y chromosome.
Incidental findings.

**Epigenetics** (François Fuks)

1. Chromatin structure
    - 1.a. Notions of chromatin structure.
    - 1.b. Post-translational modifications of histones
    - 1.c. DNA methylation: definition
    - 1.d. Biological significations of epigenetics

2. Epigenetics : mechanistic insights
    - 2.a. Crosstalks between histones modifications et DNA methylation: the histone code hypothesis
    - 2.b. Targeting and regulation of DNA methylation

3. Epigenetics and human diseases
    - 3.a. DNA methylation and cancer
    - 3.b. DNA methylation and neurodevelopmental syndromes (Rett and ICF)
    - 3.c. Deacetylase and DNA methyltransferases inhibitors as anti-cancer epigenetic drugs

Last session = 16:30-17:30

**DAY 3: UG 08.12.2023**

**Human gene mapping and disease gene identification**

Mapping of human genes: principles and applications (Paul Coucke and Andy Willaert)

Practical exercises with lod score calculations (Paul Coucke and Andy Willaert)

**The molecular, biochemical and cellular basis of genetic disease**

General introduction (Paul Coucke)

Examples of monogenic diseases caused by mutations in different classes of proteins:

Inborn errors of metabolism (Olivier Vanakker)

Mutations in developmental genes (Bert Callewaert)

Mutations in extracellular matrix proteins (Sofie Symoens)

Mutations in signaling pathways (Bart Dermaut)

**DAY 4: IPG 19.01.2024**

**09:30 - 11:00** : **Disorders of the autosomes** (cytogenetics/molecular abnormalities and clinical aspects) (Chap. 6, part 1) Damien Lederer and Stéphanie Moortgat

*11:00 - 11:15 coffee break*

**11:15 - 12:45** : **Disorders of gonadal and sexual development** (gonadal embryogenesis, cytogenetics/molecular abnormalities, and clinical aspects) (Chap. 6, part 2) Isabelle Maystadt

*12:45-13:30 lunch*

**13:30 - 15:00 : Developmental Genetics and Birth defects** (Chap. 14) Aude Tessier and Julie Desir

*15:00 - 15:30 coffee break*

**15:30 - 16:30** : **Treatments of metabolic diseases** (Chap. 13, part 1) Dominique Roland

**DAY 5: UCL 09.02.2024**

**Auditoire central C (to be confirmed !)**
**UCL – Medical Faculty (Cliniques universitaires Saint Luc),**
**Avenue Mounier, 51, B-1200 Brussels**
(by metro: line 1 towards Stockel, ALMA station
> Auditoires centraux > Auditoire C)

**08.45-09.00 Registration (signatures of presence)**

**DEVELOPMENTAL GENETICS and BIRTH DEFECTS (relates to Thompson 7th ed chptr 14)**
**09.00-09.05** Introduction: Pr. Miikka Vikkula
**09.05-09.35** Birth defects: approach to syndromology: Pr. Yves Sznajer
**09.35-10.20** Regulators of development: HOXs etc.: Pr. René Rezsohazy
**10.20-10.30** Q & A
**10.30-10.50** ***Coffee & Tea***
**10.50-11.25** Vascular anomalies and overgrowth syndromes: Dr. Pascal Brouillard
**11.25-11.50** Cleft lip and palate: Pr. Nicole Revencu
**11.50-12.00** Q & A

**CANCER GENETICS and GENOMICS (relates to Thompson 7th ed; chptr 16)** Introduction to Cancer
**12.00-12.05** Introduction: Pr. Miikka Vikkula
**12.05-12.30** Molecular basis of somatic oncogenesis: Pr. Nisha Limaye
**12.30-12.55** Hereditary basis of cancer – specifics for genetic counseling: Dr. Anne De Leener
**12.55-13.00** Q & A
**13.00-13.40** ***LUNCH***
Hereditary cancers :
**13.40-14.20** Inherited breast and ovarian cancer: Pr. François Duhoux
**14.20-14.35** Other inherited cancer predispositions Dr. Anne De Leener
**14.35-15.00** Inherited colon cancer: Dr. Anne De Leener
**15.00-15.10** Q & A
**15.10-15.30** ***Coffee & Tea***
Acquired cancers
**15.30-15.50** Hematological malignancies: molecular pathogenesis of AML, as example. Dr. Violaine Havelange
**15.50-16.10** Paragangliomas and pheochromocytomas Pr. Alexandre Persu
**16.10-16.20** Q & A
Cancer treatment
**16.20-16.50** Biomarker-driven clinical trials: Pr. Jean-Pascal Machiels
**16.40-17.00** Q & A
**17.00** ***Closing***

**DAY 6: UA 15.03.2024**

***Genetics of common disorders with complex inheritance***

Elementary concepts of multifactorial diseases (*Bettina Blaumeiser)*

Concepts in complex genetics: from Fisher to GWAS (*Guy Van Camp)*

Beyond GWAS (*Erik Fransen)*

Osteoporosis as paradigm for studies into complex diseases (*Wim Van Hul)*

Of mice and human genetics (*Frank Kooy)*

Web resources and datamining in genetics (*Geert Vandeweyer, Wim Wuyts)*

**DAY 7: ULg 26.04.2024**

***Pharmacogenetics and pharmacogenomics***

Pharmacogenetics (Vincent Bours)
- metabolic enzymes and polymorphisms.
- Polymorphisms and drug transporters and targets.
- Genetic conditions generating unexpected side effects.
- Ethnic differences and clinical consequences.

***Clinical aspects of dysmorphology*** (Saskia Bulk).

***The immune system***

Immune genetics (Stéphanie Humblet (KULeuven), Sabine Franke )
The HLA complex .
inherited immune defects.
Gene therapy in relation with immune defect.
acquired alterations of IGH and TCR loci in lymphoproliferative disorders.

**DAY 8: VUB 17.05.2024**

Auditorium 5,
VUB, Campus Jette,
Faculteit Geneeskunde & Farmacie,
Laarbeeklaan 103**,** 1090 Brussel

**08.45 - 9.15** Registration

**09.15 - 10.00** Recurrence risk in medical genetics Dr. Pieter Verdyck

**10.00 - 10.45** Mt disorders and recurrence risk: Dr. Sara Seneca

**10.45 - 11.05** Coffee break

**11.05 - 13.00** Prenatal diagnosis Clinical introduction: Dr. Kathelijn Keymolen

- Cytogenetic analysis: Dr. Ann Van den Bogaert
- DNA analysis: Dr. Katrien Stouffs
- NIPT: Dr. Ann Van den Bogaert

**13.00 - 14.00** Lunch

**14.00 - 16.00** Preimplantation Genetic Test

- PGT Dr. Martine De Rycke
- PGT for chromosomal abnormalities Dr. Pieter Verdyck
- Psychological issues in reproductive genetics Mevr. Julie Nekkebroeck

**ADDITONAL WORK**

Students must contact the Program Organizing Committee (POC) member at the institution where they registered. Each student must produce a personal work on a genetic subject, under the supervision of a member of the Teaching College from his registration center. The contents should be equivalent to a publication in a peer-reviewed scientific journal as first author, or to a thorough review of a genetic topic presented to an interdisciplinary seminar. The additional must produce a written report. The additional work amounts to 2 ECTS.

**FINAL EXAMINATION**

**07.06.2024**

**06.09.2024**