UCLouvain

wmds1229

2021

Génétique humaine

2.00 credits	20.0 h	Q2

Teacher(s)	Vikkula Miikka ;
Language :	French > English-friendly
Place of the course	Bruxelles Woluwe
Prerequisites	*The prerequisite(s) for this Teaching Unit (Unité d'enseignement – UE) for the programmes/courses that offer this Teaching Unit are specified at the end of this sheet.*
Learning outcomes	
Evaluation methods	The student should show his-her acquired knowledge in following situations : during the exam (multiple choice questions). This assessment is based on a series of multiple-choice questions covering all the material given during the course - 5 proposals / 2 only correct - no negative point. If the student has chosen 0 or only 1 of the correct answers, or more than 2 answers, no point is awarded. Scores are rounded up to the next highest grade if > or equal to 0.5 (e.g. 12,5 becomes 13), except for grades between 9.5 and <10 which are rounded to 9. The book Pasternak conteins questions and the book Thompson & Thompson questions and answers for self-learning after each chapter.
Teaching methods	Teaching is based on lectures (total 20 hours). It relies on the development of theoretical concepts, but also on the description of concrete examples of genetic diseases and genetic analyses.
Content	**1. DNA - carrier of genetic information** **2. Cytogenetics: low-resolution genome analysis** 2.1. Normal karyotype 2.2. Abnormal karyotype **3. Types of polymorphisms** 3.1. Vocabulary: polymorphism, gene, allele, homozygous, heterozygous 3.2. Genome in a single (<10) copy (haploid genome) 3.3. Genome moderetly repetitive 3.4. Genome strongly repetitive 3.5. Other polymorphisms 3.6. Counting of heterozygosity **4. Methods to detect genetic markers** (polymorphisms) 4.1. Southern blot 4.2. PCR amplification 4.3. Molecular karyotyping (DNA arrays) 4.4. Sequencing 4.5. Next Generation Sequencing (NGS) **5. Transmission of hereditary characters (Laws of Mendel)** 5.1. How to draw a genealogic tree in genetics - symbols 5.2. Types of heredity 5.3. Independent and non-independent segregation **6. Use of polymorphisms** 6.1. Genetic maps 6.2. Identification of an individual 6.3. Linkage analysis 6.4. Autozygosity analysis 6.5. Association studies 6.6. Loss-of-heterozygosity analyses **7. Other parameters of heredity** 7.1. *de novo* mutations 7.2. Factors to modify the phenotype 7.3. Law of Hardy-Weinberg 7.4. eQTL

	7.5. Personalised medecine
Bibliography	Dian-Donnai-Genetique-medicale; De la biologie à la pratique clinique; de Boeck, 2009. New Clinical Genetics 4 (Reed & Donnai) Robert L. Nussbaum, Roderick R. McInnes, Huntington F. Willard. Genetics in Medicine Editeur: Thompson & Thompson, 8e édition, Elsevier, (2016) - Syllabus (Notes de cours vérifié)(iCampus) - Dias du cours (iCampus) et Jack Pasternak : Génétique moléculaire humaine : une introduction aux mécanismes des maladies héréditaires Editeur: De Boek (2003) **Chapitres: 1-3(p.3-80), 5.1, 5.4, 5.6, 5.7, 6 (p.161-183)**
Faculty or entity in charge	MED

Programmes containing this learning unit (UE)

Program title	Acronym	Credits	Prerequisite	Learning outcomes
Bachelor in Biomedicine	SBIM1BA	2	WMD1120 AND WMD1106 AND WFARM1221S AND WSBIM1226 AND WMDS1230 AND WFARM1247	🔍
Bachelor in Medecine	MD1BA	2	WMEDE1112 AND WMDS1109 AND WMDS1111	🔍