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COURSE: **Human and population genetics**

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ACADEMIC YEAR: 2016/2'17

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TYPE OF EDUCATIONAL ACTIVITY: Free choice

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TEACHER: Matilde Valeria Ursini

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e-mail: ursini@igb.cnr.it

website:

phone: +393473053411

mobile (optional):

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Language: italian

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ECTS: 8

n. of hours: 64(lessons e  
tutorials/practice)

Campus: Potenza  
Dept./School: Dept Science  
Program: **BIOTECNOLOGY (L2)**

Semester: First

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#### EDUCATIONAL GOALS AND EXPECTED LEARNING OUTCOMES

Knowledge of the basic elements of population genetics with several examples especially related to pathological conditions. Learning of the basis of human genetics and introduction to the practical applications of human genetics and to the evaluation of future of human specie in a perspective of personalized medicine.

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#### PRE-REQUIREMENTS

Good knowledge of basic genetics and molecular biology. Enough good knowledge of English is requested.

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#### SYLLABUS

Genes and populations. Genetic polymorphisms and calculation of allele and genotype frequencies. Basic elements of population genetics: Hardy-Weinberg equilibrium; selection; mutation; migration; genetic drift; consanguinity The factors of evolution. The mutation. The selection: general concepts and model of heterozygote advantage. Mutation and selection: equilibrium for recessive and dominant mutations. Genetic drift: concepts and examples. Migration. The genetic variability within and inter the populations and the concept of race.

Construction and analysis of pedigrees also in the presence of incomplete penetrance and variable expression. Atypical mode of inheritance, mitochondrial inheritance, multifactorial inheritance.

Abnormalities of the human karyotype and associated diseases. The complex characters: methods of genetic analysis and examples. The hemoglobin genes polymorphisms and malaria: sickle cell anemia and thalassemia. Mutations and trinucleotide repeats, concepts of premutation and anticipation: Huntington's and the X-fragile. Genomic imprinting: the examples of Prader-Willi and Angelman. The assumption of Mary Lyon. Outline of pharmacogenetics and ecogenetics. Human genetics and society. The genetics of cancer: the genes mutated and their functions.

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#### TEACHING METHODS

The course includes 64 hours of teaching between theoretical lessons and classroom tutorials. In particular it is provided 54 hours of theoretical lessons and 10 hours of classroom tutorials.

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#### EVALUATION METHODS

Discussion of a project work and Oral examination,

The assessment will be done through a final exam consisting of a traditional oral examination, which will aim to verify the understanding of the various topics covered in the lessons and knowledge acquired and to highlight the ability of the student to make connections between topics.

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#### TEXTBOOKS AND ON-LINE EDUCATIONAL MATERIAL

Michael R.Cummings, "Eredità". EdiSES -

Deeping for the molecular aspects: T.Strachan, A.P.Read, "Genetica Umana Molecolare". Zanichelli

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#### INTERACTION WITH STUDENTS

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From the beginning of the course, after describing the objectives, program and methods of verification, the teaching materials will be made available to the students (shared folders). Simultaneously, the list of students who intend to enroll in the course will be collected, together with name, serial number and email. Office hours: Tuesdays from 12 am to 14 pm at the Department of science (the study of prof. Lioi). In addition to weekly reception, the professor is available at all times for a contact with the students, through their e-mail or telephone.

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EXAMINATION SESSIONS (FORECAST)<sup>1</sup>

16/02/2017; 09/03/2017; 15/06/ 2017; 13/07/ 2017 ; 14/09/ 2017; 05/10/ 2017.

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SEMINARS BY EXTERNAL EXPERTS    YES X     NO

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FURTHER INFORMATION

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<sup>1</sup> Subject to possible changes: check the web site of the Teacher or the Department/School for updates.