Course: Graduate Program in Health Sciences.

Subject:
Clinical, genetic and pathophysiological aspects of molecular research and diagnosis of albinism.

Summary:
Knowledge about molecular research and diagnosis is of fundamental importance in the context of albinism and also of other disorders commonly seen in school hospitals and reference centers. The main goal of the program is to exchange information and sum clinical and molecular knowledge about albinism. This should improve the molecular research in this field bringing numerous benefits to the patients, as well as the improvement and greater qualification of the members of the graduate student and medical and team.

Goal:
The exchange of information and knowledge between different reputed and referential medical institutions in several areas. Student and medical training, through the exchange of experiences, especially among different countries and the share of different views and approaches in relation to diseases.

Methodology:
Expositive classes and paper discussion.

Programm:
05/06/2018 - 10h às 12h - Dr. Christiano Altenfelder room - Genome maps and genome organization.
06/06/2018 - 10h às 12h - Dr. Christiano Altenfelder room - Techniques for the molecular diagnosis of genetic diseases, genome sequencing.
07/06/2018 - 8h às 10h - Dr. Zeferino Veloso room - Albinism: clinical, genetic and pathophysiological aspects.
08/06/2018 - 10h às 12h - Dr. Christiano Altenfelder room - Therapeutic aspects in genetic diseases.
18/06/2018 – 9h às 12 – room to be determined - Paper discussion.
Distance Learning (oral presentation preparation and study) – 14 hours.

Evaluation:
Students will be evaluated by their presence in the classes and performance in the paper discussion.
Recommended bibliography:

Increasing the complexity: new genes and new types of Albinism.

Albinism in a patient with mutations at both the OA1 and OCA3 loci.

High-resolution array-CGH in patients with oculocutaneous albinism identifies new deletions of the TYR, OCA2, and SLC45A2 genes and a complex rearrangement of the OCA2 gene.

The Cardiovascular Actions of DOPA Mediated by the Gene Product of ocular albinism 1.

Molecular characterization of a series of 990 index patients with albinism.

Oculocutaneous Albinism Type 4.

Mutational Analysis of Oculocutaneous Albinism: A Compact Review.

Identification of a homozygous mutation of SLC24A5 (OCA6) in two patients with oculocutaneous albinism from French Guiana.

Lessons of a day hospital: Comprehensive assessment of patients with albinism in a European setting.

Teaching Staff:

Professor Benoît Arveiler, PhD - Laboratoire Maladies Rares : Génétique Service de Génétique Médicale et Métabolisme – INSERM U1211 – Université de Bordeaux.

Professor Tatiana R. Rosenstock - Department of Physiological Sciences – Santa Casa de Sao Paulo School of Medical Sciences.

Professor Hudson S. Buck - Department of Physiological Sciences – Santa Casa de Sao Paulo School of Medical Sciences.