

CoPGr CURRICULAR CHAMBER

SUBJECTS PRESENTATION FORM

SUBJECT'S ACRONYM: **RNP5739**

SUBJECT'S NAME: Hereditary Neuropathies, Spinocerebellar Ataxias and Hereditary Parapareses: Clinical, Genetic and Molecular Bases

CURRICULUM/AREA: Neurology/17140

FOCAL AREA: Neurology

INITIAL VALIDITY (Year/Semester):

N. OF CREDITS: 04

Theoretical Classes: 02 Practical Classes, Seminars and Others: 09 Hours of Study: 04

DURATION IN WEEKS: 4

PROFESSOR(S) IN CHARGE:

USP Professor, n. 93273 – Wilson Marques Junior

External Professor, n. USP 5865279 – Charles Marques Lourenço

ACTUAL COSTS OF THE SUBJECT: BRL

(Presenting, if applicable, the budget foreseen for the year, as an attachment)

PROGRAM

OBJECTIVES:

Studying the clinical aspects and the molecular genetics of the hereditary neuropathies, spinocerebellar ataxias and hereditary spastics parapareses.

JUSTIFICATION:

The study of the neurodegenerative diseases has been propelled with the introduction of molecular biology techniques. The objective of the course is introducing to technician students the investigation in molecular biology and methods of the correlation between genotype - phenotype.

CONTENT (SYLLABUS):

Basic program:

a) Theoretical Classes:

- Principles of genetics
- Techniques in molecular biology
- Studies of generic mapping
- Identifying the genes responsible by the disease

b) Seminars:

- The classification of the hereditary neuropathies
- Motor and sensory hereditary neuropathy IA-type
- Chromosomes duplication 17p11.2
- PMP22 Mutation
- Motor and sensory hereditary neuropathy 1B-type
- Motor and sensory hereditary neuropathy II-type
- Hereditary neuropathy with sensitivity to compression

- Dejerine-Sottas disease
- Recessive autosomal motor and sensory hereditary neuropathies
- Other motor and sensory hereditary neuropathies
- Motor and sensory hereditary neuropathies
- Family amyotrophic lateral sclerosis
- Autosomal dominant cerebellar ataxias
- SNC polyglutamines and degenerative diseases
- Friedreich's ataxias
- Family spastic parapareses

Practical classes:

- Detection of 17p11.2 duplication
- Genetic sequencing
- Detection repeated nucleotides triad

EVALUATION METHOD:

Seminar and report