

RECRUITING AND TRAINING PHYSICIANS-SCIENTISTS TO EMPOWER TRANSLATIONAL RESEARCH  
A MULTILEVEL TRANSDISCIPLINARY APPROACH FOCUSED ON METHODOLOGY, ETHICS AND INTEGRITY IN  
BIOMEDICAL RESEARCH - 2018-2023



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## RESEARCH TRAINING PROGRAM

### I. General Information

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**Title of the research project:**

GENETICS OF PARKINSON'S DISEASE: TOWARD A DEFINITION OF PECULIAR CLINICAL SUBTYPES

**Name and address of the department:**

Laboratory of Molecular Genetics and Cytogenetics/ Center of Parkinson Disease and Movement Disorders, IRCCS Mondino Foundation

**Student's supervisor:**

Prof. Enza Maria Valente

*Tutor in clinical activity: dr. M. Avenali*

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### II. Description of the project

(max 1500 characters, spaces included)

**Background**

Parkinson's disease (PD) is the second most frequent neurodegenerative disorder, affecting 1% of population over 65 years of age worldwide. In the last 20 years several genes have been associated with PD. The discovery of genetic contributors has given an insight into molecular mechanisms underlying the pathogenesis, providing novel molecular biomarkers for early diagnosis and targets for innovative therapeutic approaches.

**What is the aim of the project?**

To identify a clinical and biochemical profile characterizing patient with PD carrying specific genetic mutations. The definition of a genotype-phenotype correlation will lead to the identification of prognostic factors and will help to select subgroup of patients suitable for novel targeted therapeutic interventions.

**What techniques and methods are used?**

Clinical examination of patients, blood sample collection, patients' clinical data collection, data entry into web-based database.

DNA extraction, genetic analysis with traditional sequencing and next generation sequencing (NGS).

**When did the department start working on this project? (year)**

2016

**Type of research project:**

Basic science       Clinical research without lab work       Clinical research with lab work

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**FMUP** FACULDADE DE MEDICINA  
UNIVERSIDADE DO PORTO



FONDAZIONE  
MONDINO  
Istituto Neurologico Nazionale  
& Clinica Scientifica IRECCS

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### III. Student's involvement

- The student will mainly observe  YES x NO
- The student will observe the experiments but will be involved in data analysis  YES x NO
- The student will take active part in experiments ("lab work")  YES x NO
- The student will take active part in clinical examination (clinical research) x YES  NO
- The student will be allowed to work with patients x YES  NO

*What are the tasks expected to be accomplished by the student?*

(max 500 characters, spaces included) → 305

The student will be involved in clinical practice, taking active part in clinical examination of PD patients, collaborating in collecting and recording patients' clinical data. S/he will learn how to visit a patient with movement disorders and interpret accurately clinical signs. S/he will also learn to analyse and interpret data and formulate his/her first scientific report.

*What is expected from/what will be the general outcome of the student?*

- x To prepare a poster / presentation / scientific report / abstract
- x The student's name will be mentioned in a future publication
- Opportunity to present together with the supervisor the results on a conference
- No specific outcome is expected

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### IV. Requirements

*What skills are required from the student?*

(max 500 characters, spaces included)

Ability to work in team, collaboration and communication skills, knowledge of Scientific English.

*Is there any special knowledge or a certain level of studies needed?*

x Subjects passed:  
Neurology (required) Genetics (preferred) Biochemistry (preferred)

x Previous experience with:  
\_\_\_\_\_ patients \_\_\_\_\_

Certificate of:  
\_\_\_\_\_

None

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Are there any legal limitations in the student's involvement in the project?  YES  NO  
If yes, what are the limitations?

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For the use of students considering participating in the project, further information can be found from the following references:  
(please add specific references, max 3)

### V. Schedule

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Duration of the project:

1 month  2 months  3 months

There are approximately 4 hours of work per day.

Available months:

x January    x February    x March    x April    x May    x June  
x July    x August    x September    x October    x November    x December

How many students can you accept to the project at the same time? 1

Special remarks:

(e.g., students should bring a white coat, any vaccinations required, etc.)

students should bring a stethoscope and a white coat. Vaccination against tetanus and HCV are strongly recommended.

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**NOTE: a scientific report is required at the end of the program**