

## Short-term scientific mission announcement for Grant period 2 (June 2015-May 2016)

**Title: Developing expertise in variants filtering methodology in whole-exome sequencing datasets from patients with tinnitus.**

**Host:** Otology & Neurotology Group CTS495, Centre for Genomics and Oncology Research – Genyo- Pfizer-University of Granada- Andalusian Regional Government, Granada, Spain

<http://www.genyo.es/en/content/view-research-group>



**Supervisor:** Dr. Jose Antonio Lopez-Escamez, MD, PhD

**Goal of the STSM:** This mission would enable the visiting researcher to initiate and conduct the analyses of whole-exome sequencing (WES) data to prioritize and filtering variants from multicase families with tinnitus with and without hearing loss.

**What the STSM host offer to the grantee:** Our research is dedicated to discover the genetic bases of Meniere's disease (MD), a complex disease characterized by episodic vertigo, hearing loss and tinnitus. MD is clinically a heterogeneous condition and our group has recruited >70 multicase families and identified some of the MD-causing genes, segregating with vestibular symptoms or sensorineural hearing loss. Recently, we have described the first genes causing familial MD.

<http://www.ncbi.nlm.nih.gov/pubmed/25305078>

We are planning to search for candidate gene segregating the tinnitus phenotype by using two approaches:

- a) WES in selected multicase families with tinnitus with or without hearing loss.
- b) WES in patients with MD with extreme tinnitus phenotype.

Specific tasks for this STSM will include:

1. Filtering of single nucleotide variants by several prioritizing strategies (PaVar score, Exomizer, VAAST, Phevor).
2. Filtering of insertion and deletions in control datasets.
3. Gene network analyses by IPA.

As Chair of the Genetic working group within TINNET, I am interested in how we can best define the canonical pathways and gene networks involved in tinnitus. For this, a prerequisite is to define tinnitus subtypes with a deep phenotyping of each subtype.

The anticipated output is contribution to a manuscript for publication, co-authored with our research group, strengthening collaboration across TINNET members.



**Requirements of the grantee:**

1. Graduate student or early career researcher in neuroscience or molecular biology who wishes to learn about exome sequencing and data analyses. Clinical researchers in audiology or ETN with strong interest in next generation sequencing willing to learn about data analysis are also welcome.
2. Good level of spoken and written English is essential and some basic level of Spanish is recommended.
3. Background with public genome databases and statistical analysis.

**Duration:** This STSM is available for 4 weeks to 12 weeks from September 2015. An extension in the framework of another mission (up to next 12 weeks) could be discussed, according to the results obtained and the budget available.

**How to apply for the STSM**

Please contact [antonio.lopezescamez@genyo.es](mailto:antonio.lopezescamez@genyo.es) in the first instance. If both parties agree, please request an application form from Susanne Staudinger [Susanne.Staudinger@medbo.de](mailto:Susanne.Staudinger@medbo.de)

After completion of the STSM the grantee is required to submit a scientific report (3-4 pages) on the visit within 30 days after end of the stay. Please submit the report to the STSM coordinator: [malgorzata.wrzosek@amu.edu.pl](mailto:malgorzata.wrzosek@amu.edu.pl)