STSM Abstract – Sara Pulido

Using data analysis to understand the molecular pathways involved in ARHL in a murine model

Age related hearing loss (ARHL), also known as presbycusis, is characterized by progressive deterioration of auditory sensitivity, loss of the auditory sensory cells, and central processing functions associated with the aging process. It affects approximately one third of the population over 60 year-old, making it the first neurodegenerative disease and the third most prevalent chronic disease in the elderly. There is no restorative medical treatment for deafness but functional replacement by means of prosthesis. Therefore, prevention and treatment of hearing loss is an unmet medical need. In the context of animal models of genetic ARHL, we have found that among the genes involved in mammalian ageing insulin-like growth factor 1 (IGF-1) is central for the physiology of the auditory system. Indeed, IGF-1 deficiency is a human rare disease (OMIM608747). Accordingly, IGF-1 deficit in mice also causes deafness and IGF-1 haploinsufficiency accelerates the cellular deterioration and the progression of ARHL. Evidences from our group support the hypothesis that IGF-1 levels are a key factor in the cochlear response to ageing and stress damage.

The analysis of gene expression and proteomic data from mice cochlea samples present difficulties and challenges when using standard analysis. This STSM has allowed me to work at the University of Sheffield with Dr. Marta Milo to develop a pipeline to perform high-throughput analysis with programming tools (R and bioconductor) to standardize the data analysis methodologies for this specific and complex tissue. For this learning purpose I have used data from a gene expression array of heterozygous and wild type *Igf1* mice cochleae to obtain more knowledge about the molecular pathways involved in the progression of ARHL. Moreover, I have learnt programming skills that will allow us to modify the pipeline and adjust it to analyze other samples and other experiments. Taken it together, the skills I have acquired have had an immediate impact in my PhD research and they will be a very powerful tool in our research.