



**THE CYPRUS INSTITUTE OF NEUROLOGY & GENETICS**  
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**ESC First Initiative Contact Grant Report**

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Dear Council Members,

I would like to express my sincere thanks to the European Society of Cardiology for their generous award of the ESC First Contact Initiative Grant. With this support I had the opportunity to visit Dr. John Elefteriades and his team at the Aortic Institute at Yale and the DNA Diagnostic Laboratory of Dr. Allen Everett Bale. My training took place from 17th April to 4th May 2018.

Dr. John Elefteriades is the Director of the Aortic Institute at Yale University and Yale New-Haven Hospital and he serves as the Director of the Yale Center for Thoracic Aortic Disease. He is a renowned cardiac surgeon who is an expert in the field of Ascending Aortic Thoracic Aneurysm (AATA). Dr. Allen Everett Bale is the Director of DNA Diagnostic Laboratory and oversees clinical genome sequencing at the Yale Center for Genome analysis.

My research objectives involve the identification of novel genes and variants that could be useful in identifying at risk individuals with aortic aneurysms, in particular non-syndromic familial ascending thoracic aortic aneurysm (ATAA). My visit at Yale provided me with the unique opportunity to acquire the interdisciplinary knowledge within the context of ATAA. The aim of my visit at Yale was to establish a first a contact with Dr. John Elefteriades and his research team and to setup a collaboration between his laboratory and ours in Cyprus, as well as to have the opportunity to observe and work on their whole exome sequencing analysis pipeline.

| During the first days of my stay at Yale, I was introduced -to the team members from Dr. John Elefteriades's and Dr. Bale's laboratories, and also present to me the specific rules

and regulations they have at Yale which I was obliged to respect and to help to orientate myself within their environment.

During my stay I worked closely with the members' of the team of Dr. Allen Everett Bale and I would like to take the opportunity to express my gratitude for all their help with with my training on whole exome sequencing analysis. It was a great experience to work with this team who were all extremely professional, friendly and helpful to me during the whole duration of my stay.

This training introduced me to the field of next generation sequencing data analysis and I had the opportunity to learn how to use different computational tools to identify causative genes in TAA patients. Under the supervision of Dr. Preti Jain, I had the opportunity to learn from their extensive expertise and skills in the host laboratory. I gained insight on how sequencing results are evaluated and interpreted as well as how to classify and report variants according to established guidelines and practices. In addition, I have greatly benefited from the opportunity I had to attend many presentations, journal clubs and lectures related to sequencing analysis as well as the group meetings that included data presentation and discussion for many different clinical cases. Academically, I learnt a lot from the many scientific discussions I actively participated in resulting in an overall very productive training.

The skills and knowledge that I have acquired during this training will be used further in my home Institute for the analysis of whole exome sequencing data in order to identify existing and novel variants that might be associated to ATAA in the Cypriot population, which so far has not been investigated.

Importantly, this visit helped me to strengthen my professional network and laid out the basis for a new and fruitful collaboration between home and host laboratories in order to acquire new knowledge into whole exome sequencing analysis for the identification of variants that are responsible for the onset of ATAA. In conclusion, my visit to Dr. Eleftheriades and Dr. Bale's laboratories has been an enriching experience, not only at the professional but also at the personal level.

Yours sincerely,



Anna Keravnou, PhD