

ERN Support and Training programme- WP17- EJP RD

Report Workshop “Workshop on functional exploration of genetic variants in cardiac diseases”

Date/Venue/Format

The workshop held on the 14th and 15th June 2022 in Lyon.

11 interactive presentations were presented by European and international speakers.

Numbers (participants, speakers, ERNs represented, patient representatives)

33 participants attended the workshop. Among them there were 11 speakers and 2 patient representatives (from the association Coeur des Mamans). Most of participants attended both days.

Analysis of the workshop satisfaction survey

14 participants answered the satisfaction survey.

The best aspect of the event reported was the high quality of the presentations and the interaction between participants.

The content of the presentations corresponded to the expectations of the participants. The presentations allowed several participants to discover the interest of other models to test the pathogenicity of cardiac variants and to consider new research projects on these themes.

2-3 participants' testimonials

➤ Pr Mohamed Chahine, Institut universitaire en santé mentale de Québec:

“This Workshop on functional exploration of genetic variants in cardiac diseases held in Lyon, France from June 14th to June 15th, 2022 provided a unique forum to discuss comprehensive scientific themes shared across genetics of cardiac disorders. The content and focus of this workshop was carefully planned to include emerging areas of research related to the genetics of cardiac arrhythmic disorders. A distinguishing feature of the Conference is the program design, which promoted an integrated discussion and multidisciplinary approach from the genetic, molecular, cellular including induced pluripotent



stem cells, tissue, and organ levels to clinical translation with novel therapeutic strategies and precision medicine.”

➤ Dr Chris Jopling, Institut de Génomique Fonctionnelle, Montpellier, France:

“I recently attended the ERN « Workshop on functional exploration of genetic variants in cardiac diseases » in June 2022 organised by Philippe Chevalier. I found this to be an excellent workshop which showcased a diverse array of models which can be used to study genetic variants in cardiac diseases. The environment was very relaxed which allowed for a great deal of discussion about the different models which were presented and how they could be used to answer a variety of questions associated with cardiac diseases. I was also able to initiate some interesting collaborations which will hopefully develop in the future.”

➤ Jean-Baptiste Reissq, PhyPedExp (INSERM U1046, CNRS UMR9214, Université de Montpellier), Montpellier, France.

“Concerning the ERN workshop about on *functional exploration of genetic variants in cardiac diseases*, I really appreciated this format, which I find is better than the big scientific congresses.

The time to chat with the other speakers between presentations is really a plus for sharing. I have nothing to complain about the general organization of the workshop which was efficient. I hope that this workshop will be repeated next year.”

If applicable a main result of the workshop /

Thank to this gathering we have been able to set up networks and start a proposal for a european Path finder call (ENEIDE). We have also consolidate a project on functional exploration in drosophilia between France and USA.

An annual meeting would be welcome if an EU grant is still available.

Conclusions, lessons learned, best practice to keep in mind.

Functional exploration of genetic variants is increasingly used. Genome editing yields to better results and application in human cardiac diseases. A multimodal approach in experimental models is shaping the future of drug discovery and prescription. International meetings are vital in a field where research is accelerating and when there is a need of better understanding and effective therapies to combat inherited cardiac diseases.