Institut Marseille Maladies rares
Aix*Marseille Université

Marseille Rare Diseases Institute – MarMaRa

Symposium

December 10th and 11th, 2020

Education – Research
Genetics – Physics- Bioinformatics - Ethics
Common diseases – Rare diseases
Patients groups - Therapy
Interdisciplinary - Mediterranean

PROGRAM

Thursday 10th December 2020

09:00-09:30 **MarMaRa presentation & news**
Thierry Brue, Director (RST)
Frédérique Magdinier, Deputy Director for Research
Denis Puthier, Deputy Director for Education

09:30-10:00 **Scientific and Training Advisory Board (STAB) presentation**
Jeanne Amiel
José Luis De La Pompa
Alberto M. Pereira
Maria Popa-Roch
Ridha M'Rad
Xenia Proton de la Chapelle

SESSION I : GENOMICS OF RARE DISEASE

10:00-10:20 ***Genetic findings in epileptic encephalopathies***, L. Villard, MMG

10:20-10:40 ***Rare pathogenic variants in mitochondrial and inflammation-associated genes in Chagas disease cardiomyopathy***, C. Chevillard, TAGC

10:40-11:00 ***Heterogeneous data integration,*** A. Baudot, MMG

11:00-11:20 Questions & answers, moderator: V. Delague, MMG

11:20-11:35 *Coffee break*

SESSION II : GENOMIC VARIABILITY FROM RARE TO COMMON DISEASES

11:35-11:55 ***Genetics of valvular heart disease***, S. Zaffran, MMG

11:55-12:15 ***Identification of ATP2B4 regulatory element containing functional genetic variants associated with severe malaria,*** S. Marquet, TAGC

12:15-12:30 Questions & answers, moderator: D. Puthier, TAGC

PRESENTATION FROM A HUMAN AND SOCIAL SCIENCES ACTOR

12:30-12:50 ***Human and social sciences and rare diseases: Schools of thoughts or the keys of a dual relationship***, P. Auquier, CEReSS

12:50-13:00 Questions & answers

13:00-14:15 *Lunch break*

SESSION III : SCIENTIFIC PLATFORM AND RESOURCES

14:15-14:30	***IPS/Reprogrammation: Modelling rare to common diseases through pluripotent stem cells***, F. Magdinier, MMG
14:30-14:50	***GBiM: a genomics and bioinformatics core facility toward empowering large-scale genomics and transcriptomic projects***, V. Delague, MMG & D. Salgado, MMG
14:50-15:05	***TGML a labeled platform: Expertise, Research and Development in genomics and bioinformatics in life sciences,*** D. Puthier, TAGC
15:05-15:20	***Animal facilities***, S. Zaffran, MMG
15:20-15:40	***Cell imaging: tools and challenges***, C. Matthews & P-F. Lenne, IBDM
15:40-15:55	***The ReMap resource: challenges in the integration of omic data,*** B. Ballester, TAGC
15:55-16:10	Questions & answers, J-C Roux, MMG
16:10-16:25	*coffee break*

LECTURE FROM A STAB MEMBER

16:25-17:10	***The role of patient associations in medical research: the example of Ondine syndrome,*** X. Proton de la Chapelle, AtmosR
17:10-17:20	Questions & answers, Frédérique Magdinier
17:20-17:30	Conclusion of the 1st day

Friday 11th December 2020

SESSION IV : THERAPIES FOR RARE DISEASES: STRATEGIES, PROGRESS AND CHALLENGES

09:00-09:20	***Coupling ultrasound and gene therapy to treat the central nervous system in rare diseases***, J-C Roux, MMG & S. Mensah, LMA
09:20-09:40	***The behavioral and neurobiological impacts of oxytocin administration in mouse neonates with autism syndrome disorder***, F. Muscatelli, INMED
09:40-10:00	***Gene therapies in muscular dystrophy, the special case of dysferlinopathies***, M. Bartoli, MMG
10:00-10:20	***OrphanDev: a FCRIN labelled French National Network for Rare Diseases Clinical Research,*** O. Blin, OrphanDev
10:20-10:35	Questions & answers, moderator: N. Levy, MMG
10:35-10:50	*Coffee break*

SESSION V : SHORT TALKS

10:50-11:05 ***Targets for heart regeneration: lessons from development,*** S. Payan, MMG

11:05-11:20 ***Application of CRISPR/Cas9 to human induced pluripotent stem cells (hiPSCs) to approaches congenital isolated ACTH deficiency,*** T. Mac, MMG

11:20-11:35 ***System biology approach links muscle weakening to alteration of the contractile apparatus in Facio Scapulo Humeral Dystrophy,*** C. Laberthonnière, MMG

11:35-11:50 ***Deciphering variants impact on calpain-3 catalytic activity,*** A. Salvi, MMG

11:50-12:05 Questions & answers, moderator: A. Casanova, LP3

12:05-13:15 *Lunch break*

13:15-13:30 ***Development of an in vitro model of immunotherapy-induced cardiotoxicity,*** T. Tran, TAGC

13:30-13:45 ***Optimization of a viral approach for the embryonic expression of Necdin in Necdin-KO mice: implication for Prader-Willi Syndrome*****,** J. Buron, INMED

13:45-14:00 **Laser-induced printing of stem cells for the creation of ordered neuromuscular junctions,** A. Casanova, LP3

14:00-14:15 ***Generation of a genetically reversible Tshz3 mouse model to rescue ASD-like deficit***, E. Arbeille, IBDM

14:15-14:30 Questions & answers, moderator: F. Lescroart, MMG

14:30-14:40 *Coffee break*

ROUND-TABLE: STAKEHOLDERS IN THE FIELD OF RARE DISEASES

14:40-15:40 **Moderator:** Laurence Colleaux, MMG

Invited speakers :
Brigitte Chabrol, Centre Référence Maladies Rares
Roseline Favresse, Fondation Maladies Rares
Nathalie Triclin, Alliance Maladies Rares

15:40-16:00 **Conclusion**
Thierry Brue Director-RST