Program (version 6)

11th Japanese-French Workshop "New insights in personalized medicine for neuromuscular diseases: From Basic to Applied Myology"

Proposed dates: **June 14th – 16th, 2018** - National Center of Neurology and Psychiatry, Tokyo, Japan.

Thursday, June 14th

13:00 Welcome address: Shin'ichi Takeda and Gisèle Bonne

13:30 **1. Molecular Pathology and Mechanism of Neuromuscular Disorders**

13:30 Dr. Ichizo Nishino (NCNP, Tokyo): Inflammatory myopathies in Japan.

14:00 Pr. Olivier Benveniste (CRM, Inserm U974, Paris): Inflammatory myopathies in France, from classification to pathophysiogenesis.

14:30 Dr. Masanori Takahashi (Osaka Univ.): Novel insights into the pathomechanism of periodic paralyses and myotonic disorders

15:00 Coffee break

15:30 Dr. Denis Furling (CRM, Inserm U974, Paris): Targeting CUG expanded-transcripts in myotonic dystrophy.

16:00 Dr. Masayuki Nakamori (Osaka Univ.): Disease mechanism in congenital myotonic dystrophy: linking pathogenesis to therapeutic targets

16:30 Dr. Kinji Ohno (Nagoya University): Splicing regulations in neuromuscular diseases.

17:00 Dr. Guillaume Bassez (Reference Center of Neuromuscular Diseases, Paris): New aspects of congenital myasthenic syndromes revealed by whole exome sequencing.

17:30 Dr. Antoine Muchir (CRM, Inserm U974, Paris): Molecular events leading to cardiomyopathy caused by *LMNA* mutations.

18:00 Dr. Matteo Garibaldi (Histopathology laboratory, Institut de Myologie, Paris) PhD student.: Morphological spectrum of recessive RYR1 myopathies: clinical, biochemical and genetic correlations.

Friday, June 15th

9:00 Dr. France Piétri-Rouxel (CRM, Inserm U974, Paris): Genotype-Phenotype link in Becker patients with a deletion of exons 45-55.

9:30 Dr. Edoardo Malfallti (Histopathology laboratory, Institut de Myologie, Paris): Role of muscle morphological analysis in Pompe disease (GSDII) and Debranching enzyme deficiency (GSDIII) in the perspective of novel therapeutic approaches

10:00 Coffee break

10:30 Dr. Tatsushi Toda (University of Tokyo): Fukutin is Ribitol 5-phosphate Transferase: Recent Advance in Fukuyama Muscular Dystrophy, ISPD, and LGMD2I

11:00 Dr. Tamao Endo (Tokyo Metropolitan Geriatric Hospital & Institute of Gerontology): Glycosylation defects in muscular dystrophies

11:30 Dr. Valérie Allamand (CRM, Inserm U974, Paris): Shear Wave Elastography to assess skeletal muscle stiffness in a COLVI-deficient mouse model.

12:00 Dr. Makiko Osawa (Tokyo Women's Medical University): Fukuyama congenital muscular dystrophy.

12:30 Dr. Motoi Kanagawa (Kobe University): Pathological analysis of model mice for Fukuyama congenital muscular dystrophy and exploration of therapeutic strategy.

13:00 Lunch break

14:00 2. Gene Therapy and Genome Editing based Therapy

14:00 Dr. Maria-Grazia BIFERI (CRM, Inserm U974, Paris): A novel AAV-mediated exon skipping gene therapy for SOD1-linked ALS.

14:30 Dr. Yoshitsugu Aoki (NCNP, Tokyo): Targeting RNA to treat neuromuscular diseases.

15:00 Dr. Takashi Okada (Nippon Medical School): AAV vector transduction strategy with immune-modulation to treat DMD.

15:30 Coffee break

16:00 Dr. Akitsu Hotta (CiRA, Kyoto University): Genome editing exon skipping approach for Duchenne muscular dystrophy

16:30 Dr. Capucine Trollet (CRM, Institut de Myologie, Paris): Therapy for Oculopharyngeal muscular dystrophy.

17:00 Dr. Akiyoshi Uezumi (Tokyo Metropolitan Geriatric Hospital & Institute of Gerontology): Controlling mesenchymal progenitors for the inhibition of fatty and fibrous degeneration in skeletal muscle.

17:30 Poster viewing.

Saturday, June 16th

9:00 3. Stem Cells and Cell-based Therapy of Muscular Dystrophy

9:00 Dr. Soichiro Fukada (Osaka University): Muscle Stem Cells and muscle hypertrophy.

9:30 Dr. Shinichiro Hayashi (NCNP, Tokyo): Regulation of muscle stem cell function during murine fetal development

10:00 Coffee break

10:30 Dr. Cécile Martinat (I-Stem, Inserm U861, Evry): Use of human pluripotent stem cells for neuromuscular diseases.

11:00 Dr. Hidetoshi Sakurai (CiRA, Kyoto University) : Development of novel therapeutic options for muscular dystrophy utilizing iPS cell technology.

11:30 4. Clinical Trials and Arrangement of Clinical Trials

11:30 Dr. Laurent Servais (Clinical Trials team, Institut de Myologie, Paris): 2 possible topics >>> **a)** *'Actimyo'*: its development for monitoring motor function for disease natural history and clinical trial **or b)** Neonatal Screening of spinal muscular atrophy (confirmed). If we choose topic b it could be move to another session may be.

12:00 Dr. Yoshihide Sunada (Kawasaki Medical School): Development of anti-myostatin therapeutics.

12:30 Dr. Jean-Yves Hogrel (Neuromuscular Physiology Laboratory, Institut de Myologie, Paris): Outcome measures in clinical trials for neuromuscular disorders.

13:00 Dr. Benjamin Marty (NMR laboratory Institut de Myologie, Paris): NMR outcome measures for NMDs.

5. Young Researcher Poster Session (to be scheduled and distribute throughout the program)

- Dr. Teresa Gidaro (Reference Center of Neuromuscular Diseases, Institute of Myology, Paris):
  Topic: Nusinersen in spinal muscular atrophy type 1 patients older than 7 months: a cohort study.
- Dr. Sestina Falcone (CRM, Institut de Myologie, Paris)
  Topic: CaVb1: The missing link from voltage sensing to muscle mass homeostasis
- Dr. Lorenzo Giordani (CRM, Institut de Myologie, Paris)
  Topic: Single-cell based analysis of functional populations in aged and dystrophic muscle
- Mrs. Blanca Morales Rodriguez, PhD student (CRM, Institut de Myologie, Paris)
  Topic: Pathogenic role of Sarcolipin in cardiomyopathy caused by mutation in lamin A/C gene
- Mrs. Mona Bensalah, PhD student (CRM, Institut de Myologie, Paris)
  Topic: Peculiar behavior of interstitial non myogenic cells from human fibrotic muscle
- Dr. Chiseko Ikenaga, and others (Tokyo University)
  Topic: E-cadherin is ectopically expressed in the muscle fiber of inclusion body myositis

Several young Japanese researchers will participate.