THE 1ST FRENCH-ITALIAN MEETING ON LAMINOPATHIES & OTHER NUCLEAR ENVELOPE-RELATED DISEASES

JANUARY 15 & 16, 2015
MARSEILLE LA TIMONE
THE 1ST FRENCH-ITALIAN MEETING ON LAMINOPATHIES & OTHER NUCLEAR ENVELOPE-RELATED DISEASES

PROFESSIONALS' MEETING JANUARY 15
08.30-09.00  WELCOME COFFEE

09.00-09.15  Welcome and opening comments.
A. De Sandre-Giovannoli, N. Lévy

Presentation of the French Network on EDMD and other nucleopathies. G. Bonne, R. Ben Yaou
Presentation of the Italian Network on laminopathies. G. Lattanzi

09.15-15.45  SESSION 1: PRECLINICAL RESEARCH ON LAMINOPATHIES

09.15-09.45  Mechanisms involved in natural and pathological aging, an overview. C. Lopez-Otín

1.1 - IPS CELLS AND OTHER IN VITRO MODELS OF LAMINOPATHIES
09.45-10.00  High-throughput screening of drugs using HGPS-iPS cells. X. Nissan
10.00-10.15  Development of a SMCs model from HGPS-iPS and proofs of principle. L. Ferreira
10.15-10.30  Human induced pluripotent stem cells to model genetic lipodystrophies: LMNA p.R482W mutation alters adipocyte and endothelial cell differentiation. N. Briand
10.30-10.45  3D culture system of muscle precursor cell to reveal mechanosensing defects in nuclear envelope related disorders. C. Coirault
10.45-11.00  Prelamin A accumulation in endothelial cells induces premature senescence and functional impairment. C. Badens

11.00-11.30  COFFEE BREAK AND VISIT OF THE EXHIBITION

1.2 - IN VIVO MODELS OF LAMINOPATHIES
11.30-11.45  Hypothalamic involvement in premature aging laminopathies. C. Cavadas
11.45-12.00  Investigation of pathomechanisms of ventricular arrhythmias in cardiac laminopathies. A. Muchir

1.3 - NOVEL THERAPEUTIC APPROACHES / PROOFS OF PRINCIPLE IN LAMINOPATHIES
12.00-12.15  New therapeutic approaches to HGPS based on progerin inhibition. C. Pellegrini
12.15-12.30  Compound X preclinical study in HGPS. K. Harhouri
12.45-13.00  Gene therapy approaches for striated muscle laminopathies. F. Azibani
13.00-14.00  LUNCH BREAK AND VISIT OF THE EXHIBITION

14.00-14.15  Metreleptin in lipodystrophic laminopathies - effects on insulin secretion.  C. Vatier
14.15-14.30  Vascular consequences of farnesylated prelamin A accumulation: role of LMNA mutations and antiHIV - protease inhibitor treatment.  C. Vigouroux

1.4 - NOVEL BIOMARKERS IN LAMINOPATHIES

14.30-15.00  Chromatin dynamics and in vitro biomarkers in laminopathies, an overview.  G. Lattanzi
15.00-15.15  LMNA p.R482W mutation related to FPLD2 alters SREBP1-LMNA interactions in human fibroblasts and adipose stem cells.  B. Buendia
15.15-15.30  Altered cytokine profiles in laminopathic patients.  P. Bernasconi
15.30-15.45  Micro-RNAs deregulation in Hutchinson-Gilford Progeria.  P. Roll

15.45-19.30  SESSION 2: CLINICAL RESEARCH

2.1 - CASE REPORTS / NOVEL MUTATIONS AND PHENOTYPES / OTHER NUCLEAR ENVELOPATHIES

15.45-16.00  Update of Emerinopathies’ clinical genetic spectrum.  F. Leturcq
16.00-16.15  FHL1 isoforms and X-linked Emery-Dreifuss muscle Dystrophy.  E. Ziat
16.15-16.30  LMNA-associated myopathies: The Italian experience in a large cohort of patients.  L. Maggi
16.30-16.45  Transportin 3 defects in LGMD1F.  S. Ortolano
16.45-17.00  Emerin oligomerisation properties, impact on lamin and actin recognition.  I. Herrera
17.00-17.15  Irisin assessment in lipodystrophic laminopathies.  M.C. Vantyghem
17.15-17.30  Lamins involvement in spermatogenesis.  M. Mitchell, C. Guillemain

17.30-18.00  COFFEE BREAK AND VISIT OF THE EXHIBITION

2.2 CLINICAL TRIAL DESIGN FOR RARE DISEASES

18.00-18.30  Which support from the French Rare Disease Foundation towards clinical trials set up in rare diseases.  L. Ravagnan
18.30-19.00  Systemic antisense morpholino approaches in genetic diseases.  R. Kole (Sarepta Therapeutics)
19.00-19.30  Round table: potential collaborations and future prospects  
            chairs: G. Lattanzi, G. Bonne
19.30-19.35  Closure of the first day  
            A. De Sandre-Giovannoli, N. Lévy
FAMILIES & PATIENTS
ASSOCIATIONS' MEETING JANUARY 16
**FRIDAY, JANUARY 16 2015**

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<td>09.15-09.45</td>
<td>Laminopathies: clinical presentations and management - a multifaceted approach.</td>
<td>R. Hennekam</td>
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<td>Congenital muscular dystrophies linked to the LMNA gene and their early management.</td>
<td>S. Quijano-Roy, A. D'Amico</td>
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<td>Implantable devices and cardiac management in striated muscle laminopathies and utility of the molecular genetics diagnosis in the clinical management.</td>
<td>K. Wahbi, G. Boriani</td>
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<td>10.45-11.00</td>
<td>Advances in muscle imaging for Emery-Dreifuss Muscular Dystrophy.</td>
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<td>Results of the leptin-based clinical trials in LMNA-linked lipodystrophies.</td>
<td>C. Vigouroux</td>
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<td>Update on the morpholino trial preparation for HGPS &amp; HGPS-like patients.</td>
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<td>14.00-15.00</td>
<td>Round table: discussions with families and associations</td>
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<td>Utility of patients’ registries to gather clinical, epidemiological and molecular informations.</td>
<td>C. Béroud</td>
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<td>Clinical aspects of cardiolaminopathies and prospects for a cardiolaminopathy registry.</td>
<td>L. Politano, S. Benedetti</td>
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<td>A common French-Italian laminopathies registry - update and future prospects.</td>
<td>G. Bonne</td>
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<td>ECLip- the European consortium on lipodystrophies: an update.</td>
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<td>Conclusions and closure</td>
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