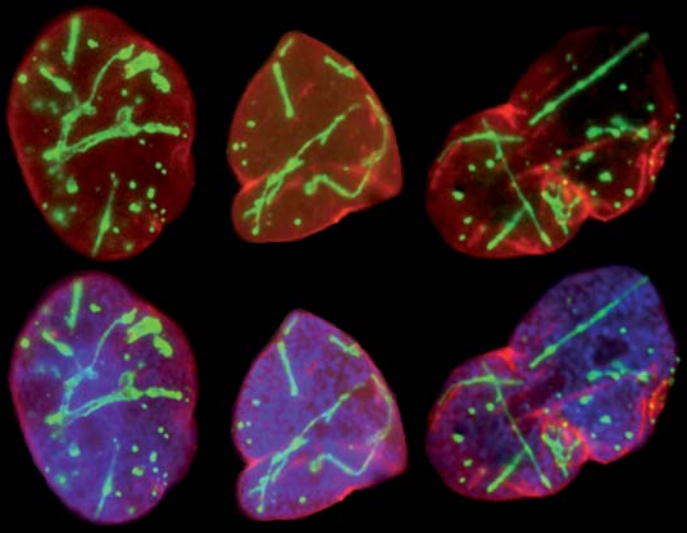
THE 1ST FRENCH-ITALIAN MEETING ON LAMINOPATHIES

& OTHER NUCLEAR ENVELOPE-RELATED DISEASES



JANUARY 15 & 16, 2015

MARSEILLE LA TIMONE

Assistance Publique Hôpitaux de Marseille

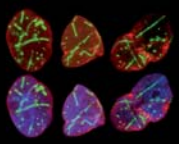
Inserm

Aix Marseille université

AFMTÉLÉTHON

A. I. Pro. Sa. R.

progeria family circle



THE 1ST FRENCH-ITALIAN MEETING ON LAMINOPATHIES

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PROFESSIONALS' MEETING JANUARY 15

THURSDAY, JANUARY 15 2015

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*

12.30-12.45 Impairment of LaminA/C-Polycomb crosstalk as a possible epigenetic cause of Emery Dreifuss Muscular Dystrophy (EDMD). *C. Lanzuolo*

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. Herraa*
17.00-17.15 Irisin assessment in lipodystrophic laminopathies. *M.C. Vantyghem*
17.15-17.30 Lamins involvement in spermatogenesis. *M. Mitchell, C. Guillemain*

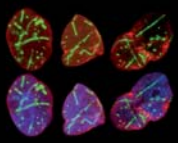
17.30-18.00 COFFEE BREAK AND VISIT OF THE EXHIBITION

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



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FAMILIES & PATIENTS ASSOCIATIONS' MEETING JANUARY 16

FRIDAY, JANUARY 16 2015

08.30-09.00 WELCOME COFFEE

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

09.15-09.45 Laminopathies : clinical presentations and management - a multifaceted approach. *R. Hennekam*

09.45-12.00 **SESSION 1 : ADVANCES IN THERAPEUTIC APPROACHES FOR LAMINOPATHIES**

09.45-10.15 Congenital muscular dystrophies linked to the LMNA gene and their early management. *S. Quijano-Roy, A. D'Amico*

10.15-10.45 Implantable devices and cardiac management in striated muscle laminopathies and utility of the molecular genetics diagnosis in the clinical management. *K. Wahbi, G. Boriani*

10.45-11.00 Advances in muscle imaging for Emery-Dreifuss Muscular Dystrophy. *N. Carboni*

11.00-11.30 COFFEE BREAK AND VISIT OF THE EXHIBITION

11.30-12.00 Results of the leptin-based clinical trials in LMNA-linked lipodystrophies. *C. Vigouroux*

12.00-13.30 LUNCH BREAK AND VISIT OF THE EXHIBITION

13.30-14.00 Update on the morpholino trial preparation for HGPS & HGPS-like patients. *A. De Sandre-Giovannoli*

14.00-15.00 **Round table : discussions with families and associations**
chairs: A. Gambineri, T. Mongini

15.00-17.30 **SESSION 2: FOCUS ON REGISTRIES AND DATABASES**

15.00-15.30 Utility of patients' registries to gather clinical, epidemiological and molecular informations. *C. Béroud*

15.30-16.15 COFFEE BREAK AND VISIT OF THE EXHIBITION

16.15-16.30 Clinical aspects of cardiolaminopathies and prospects for a cardiolaminopathy registry. *L. Politano, S. Benedetti.*

16.30-17.00 A common French-Italian laminopathies registry - update and future prospects. *G. Bonne*

17.00-17.20 ECLip- the European consortium on lipodystrophies: an update. *D. Araujo-Vilar*

17.20-17.30 **Conclusions and closure** *A. De Sandre-Giovannoli, N. Lévy*

NOTES

ASSISTANCE PUBLIQUE HÔPITAUX DE PARIS

INSTITUTE OF MYOLOGY
CURE THROUGH INNOVATION

Alessandra Proietti O.N.L.U.S.

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Istituto Ortopedico Rizzoli di Bologna
Istituto di Ricovero e Cura a Carattere Scientifico

DIATHEVA

NETWORK ITALIANO LAMINOPATIE
ITALIAN NETWORK FOR LAMINOPATHIES

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

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