GMGF - Génétique médicale et génomique fonctionnelle

Rapport Hcéres

HCERES

High Council for the Evaluation of Research and Higher Education

Department of Research Evaluation

Report on research unit:

Functional Genomics and Medical Genetics

GMGF

under the supervision of the following institutions and research bodies:

Aix-Marseille Université

Institut National de la Santé Et de la Recherche Médicale - INSERM

Evaluation Campaign 2016-2017 (Group C)

HCERES

High Council for the Evaluation of Research and Higher Education

Department of Research Evaluation

In the name of HCERES,[1]	*In the name of the experts committee,*[2]
Michel Cosnard, president	Meena Upadhyaya, chairwoman of the committee

Under the decree N°.2014-1365 dated 14 november 2014,

[1] The president of HCERES "countersigns the evaluation reports set up by the experts committees and signed by their chairman." (Article 8, paragraph 5)

[2] The evaluation reports "are signed by the chairman of the expert committee". (Article 11, paragraph 2)

Evaluation report

This report is the sole result of evaluation by the expert committee, the composition of which is specified below.

The assessments contained herein are the expression of an independent and collegial reviewing by the committee.

Unit name:	Functional Genomics and Medical Genetics
Unit acronym:	GMGF
Label requested:	UMR
Current number:	UMR_S-910
Name of Director (2016-2017):	Mr Nicolas LEVY
Name of Project Leader (2018-2022):	Mr Nicolas LEVY

Expert committee members

Chair:

Ms Meena UPADHYAYA, Institute of Cancer & Genetics, Cardiff University, UK

Expert:

Ms Delphine DUPREZ, Pierre et Marie Curie University, Paris

Mr Ian FRAYLING, Institute of Cancer & Genetics, Cardiff University, UK

Mr Francesc PALAU, Sant Joan de Déu Children's Hospital, Barcelona, Spain

Ms Agnès RÖTIG, INSERM U781 Hôpital Necker-Enfants Malades, Paris

Ms Christel THAUVIN-ROBINET, Centre de Génétique Hôpital d'Enfants, Dijon (representative of the CSS INSERM)

Mr Nicolas THIERRY-MIEG, Faculté de Médecine, La Tronche

Mr Jean-Luc THOMAS, ENS, Lyon (representative of supporting personnel)

Ms Elisabeth TOURNIER-LASSERVE, Paris 7 University (representative of the CNU)

Scientific delegate representing the HCERES:

Mr Bohdan WASYLYK

Representatives of supervising institutions and bodies:

Mr Pierre CHIAPPETTA, AMU

Ms Aurélie PHILIPPE, INSERM

Head of Doctoral School:

Mr Jean-Louis MEGE, Doctoral School ED n°62, "Science de la Vie et de la Santé"

1 • Introduction

History and geographical location of the unit

AMU-INSERM UMR_S-910 was created in 2008 and renewed in 2012. This unit is geographically based at the Hospital University campus - la Timone and has close links with the department of Medical Genetics. It is emerging as a reference center in Europe for clinical studies, molecular diagnosis and research on rare diseases.

Management team

The unit is directed by Mr Nicolas LEVY and the management of the unit is provided by Mr Rodolphe MOREAU (ADAENES personnel) with the help of 3 administrative assistants.

HCERES nomenclature

Main: SVE2 Biologie Cellulaire, Imagerie, Biologie Moléculaire, Biochimie, Génomique, Biologie Systémique, Développement, Biologie Structurale.

Secondary: SVE5 Physiologie, Physiopathologie, Cardiologie, Pharmacologie, Endocrinologie, Cancer, Technologies Médicales.

Scientific domains

The main themes are rare genetic and developmental diseases of the central and peripheral nervous systems, skeletal muscle and the heart, as well as pathologies associated with defects of nuclear envelope proteins that are particularly involved in premature and accelerated aging and cancer. There are strong transversal research themes, that include development, epigenetics, systems biology, bioinformatics and hiPSC.ial.

Unit workforce

Unit workforce	Number on 30/06/2016	Number on 01/01/2018
N1: Permanent professors and similar positions	24	33
N2: Permanent researchers from Institutions and similar positions	18	23
N3: Other permanent staff (technicians and administrative personnel)	24	36
N4: Other researchers (Postdoctoral students, visitors, etc.)	11	
N5: Emeritus	2	
N6: Other contractual staff (technicians and administrative personnel)	11	
N7: PhD students	20	
TOTAL N1 to N7	**110**	
Qualified research supervisors (HDR) or similar positions	25	

Unit record	From 01/01/2011 to 30/06/2016
PhD theses defended	24
Postdoctoral scientists having spent at least 12 months in the unit	11
Number of Research Supervisor Qualifications (HDR) obtained during the period	8

2 • Assessment of the unit

Global assessment of the unit

This unit comprises about 150 staff members, 8 research teams and 4 departments. It has strong links with hospital and research centers for rare diseases. Patient recruitment, cohort characterisation and availability of samples are facilitated by the hospital based clinical department of Medical Genetics at the Children Hospital "La Timone" (head: Mr Nicolas LEVY). The Medical Genetics department also offers several core facilities for diagnosis and investigation of patients as well as for research: 2 comparative genomic hybridization array platforms, NGS platforms, a platform for DNA molecular combing.

Several members of the unit lead rare disease reference centers for neuromuscular diseases and neurogenetics, developmental defects and neuroendocrine disorders. Another member leads a competence center for arteriovenous malformations. The majority of associated clinicians are connected with the research laboratories through these reference centers.

The unit also collaborates with several other hospitals and research laboratories at the national and international level. In the 5 years from 2011 to 2016, the unit has been extremely productive, with a total of 886 publications.

It must be noted that in the same period the unit has performed four major clinical trials. The effort involved in these trials is considerable and must be taken into account when considering the total output of the unit.

The unit focuses on various genetic conditions including Rett syndrome, intellectual deficits, early onset epileptic encephalopathies, constitutional hypopituitarism, FacioScapular Humeral muscular Dystrophy (FSHD), other neuromuscular diseases, cardiovascular diseases, Hutchinson-Gilford disease and selected cancers.

It has expertise in genomics, transcriptomics, proteomics, kinomics, animal models and bioinformatics. The unit also has expertise in modeling genetic diseases with induced pluripotent stem cells. During the last five years, it has also made advances through clinical trials, the discovery of new disease genes, translational research, patents, licenses, and therapeutic and biotherapeutic strategies. The unit has excellent core facilities, including 5 technical platforms, and has a coherent scientific trajectory with interest to translate basic findings into clinical trials and medical practice.

The unit has numerous partnerships and links through international networks and has obtained 12 patents and several licenses.

This unit has created 20 new locus specific databases. A private-public (Quest diagnostics / INSERM) partnership has been developed to exploit two of these databases. A spin-off company is under creation to valorize two softwares devoted to genomic-variants interpretation. The Bioinformatic team is one of the national bioinformatics platforms accredited by the French National Bioinformatic Institute and is also an ELIXIR partner.

Although this unit is publishing regularly, it should make efforts to increase the impact of its research findings. This could be achieved in two ways: 1) publishing in higher impact journals; and 2) increasing the level of first author/senior/corresponding authorship. In addition, greater promotion of the unit's work at European and international conferences should take place.

International competition is a threat, as is the low level of European and other international funding for some of the teams.

Greater involvement in bodies responsible for rare disease guidance at European/international levels should take place (e.g. the forthcoming European Rare disease Networks: ERN).

Researchers in this unit have to take care of more administrative tasks than other similar European institutions. There is also difficulty in recruiting non-tenured personnel for long periods, which threatens to result in an overall reduction in competency within the unit.

Disease gene databases should work with, and be integrated into existing and established international databases and endeavours to interpret variants, both clinically and scientifically. For the databases that the unit leads on internationally, there are clear opportunities to establish the unit's leadership at an international level.