MolCelRD

Rapport Hcéres

Research evaluation

REPORT ON THE RESEARCH UNIT:

Molecular and cellular bases of rare diseases (MolCelRD)

UNDER THE SUPERVISION OF THE FOLLOWING INSTITUTIONS AND RESEARCH BODIES:

Institut national de la santé et de la recherche médicale - Inserm

Université Pierre et Marie Curie

Aix Marseille Université

EVALUATION CAMPAIGN 2017-2018

GROUP D

In the name of Hcéres[1]:

Michel Cosnard, President

In the name of the expert committee[2]:

Marguerite Neerman-Arbez, Chairwoman of the committee

Under the decree No.2014-1365 dated 14 November 2014,

[1] The president of Hcéres "countersigns the evaluation reports set up by the expert 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).

This report is the sole result of the unit's 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 PRESENTATION

Unit name:	Molecular and cellular bases of rare diseases
Unit acronym:	MolCelRD
Requested label:	UMR
Application type:	Renewal
Current number:	UMR_S933
Head of the unit (2017-2018):	Mr Serge AMSELEM
Project leader (2019-2023):	Mr Serge AMSELEM
Number of teams:	1

COMMITTEE MEMBERS

Chair:	Ms Marguerite NEERMAN-ARBEZ, Université de Genève, Switzerland (representative of CNU)
Experts:	Mr Benoît ARVEILER, Université de Bordeaux
	Mr Christophe BEROUD, Aix-Marseille Université (representative of Inserm CSS)
	Ms Sylvie TUFFERY-GIRAUD, Université de Montpellier (supporting personnel)
HCERES scientific officer:	
	Mr Jean-Paul LALLÈS
Representatives of supervising institutions and bodies:	
	Ms Catherine N'GUYEN, Inserm
	Mr Bernard ZALC, UPMC

INTRODUCTION

HISTORY AND GEOGRAPHICAL LOCATION OF THE UNIT

This is a renewal application for the single-team research unit UMR_S933 previously renewed in 2014. UMR_S933 is a joint structure between Inserm and the University of Paris 6 unit located at the Armand-Trousseau hospital, where the research unit (150 m^2) shares premises with the clinical laboratory.

MANAGEMENT TEAM

The UMR_S933 is directed by Mr Serge AMSELEM (PUPH), with no deputy head.

HCERES NOMENCLATURE

SVE2; SVE5.

SCIENTIFIC DOMAIN

The UMR_S933 research unit is studying three groups of rare diseases: rare pulmonary disorders (primary ciliary dyskinesia (PCD) and interstitial lung diseases), auto-inflammatory disorders and several developmental disorders including growth disorders of endocrine origin, disorders of sex development, premature ovarian insufficiency and infertility. The goals are: to identify the molecular defects responsible for these diseases in patients; to understand the pathophysiology of those rare diseases by designing various disease/gene models; to characterize the molecular networks impacted by the mutations and ultimately to search for new therapeutic options for patients.

UNIT WORKFORCE

Unit workforce	Number 30/06/2017	Number 01/01/2019
Permanent staff		
Full professors and similar positions	6	6
Assistant professors and similar positions	4	5
Full time research directors (Directeurs de recherche) and similar positions	0	0
Full time research associates (Chargés de recherche) and similar positions	1	1
Other scientists ("Conservateurs, cadres scientifiques des EPIC, fondations, industries, etc.")	3	3
High school teachers	0	0
Supporting personnel (ITAs, BIATSSs and others, notably of EPICs)	4	3
TOTAL permanent staff	**18**	**18**
Non-permanent staff		

Non-permanent professors and associate professors, including emeritus	1	
Non-permanent full time scientists, including emeritus, post-docs	6	
Non-permanent supporting personnel	1	
PhD Students	5	
TOTAL non-permanent staff	**13**	
TOTAL unit	**31**	

GLOBAL ASSESSMENT OF THE UNIT

The unit has continuously produced high-level translational research resulting in an impressive publication record both quantitatively and qualitatively. Teaching and training activities at the pre-graduate and post-graduate level are numerous and diverse. During the last contract, the unit has further elucidated the molecular basis of three groups of rare diseases i.e. pulmonary diseases including PCD, auto-inflammatory disorders and developmental disorders. Thanks to the team's long-term expertise in these diseases, their close relationship with the diagnostic laboratory, the access to large cohorts of clinically well characterized patients and involvement of the unit in the national diagnostic/health networks, the unit has significantly contributed to a better knowledge of the genetic basis of these clinically and genetically heterogeneous groups of disorders. The unit occupies a leader position at the international level for the investigation of PCD in particular and has growing international visibility in the fields of autoimmune diseases and developmental disorders. This research has already led to improved diagnosis, genetic counselling and care of the patients affected by these diseases. The next contract period should continue to produce clinically important results and may give rise to new therapeutic approaches.

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