

Emmanuelle JOUANGUY, PhD

A. DEGREE CONFERRAL

- 09/1995 M.Sc. (Biochemistry & Immunology), Jussieu Faculty of Sciences, Univ. Paris Pierre et Marie Curie, Pasteur Institute, France
12/1998 Ph.D. (Genetic Immunology), Necker Medical School, INSERM U429, Univ. Paris Pierre et Marie Curie, France
12/2012 HDR (Genetic Immunology), Paris René Descartes University, France

B. RESEARCH SUMMARY

My life as a scientist is devoted to working on the hypothesis that severe infectious diseases in children and young adults may result from single-gene inborn errors of immunity (IEI). During my graduate studies, I identified mutations in *IFNGR1* associated with severe disseminated infections caused by weakly virulent mycobacteria, establishing the important role type II IFN plays in immunity against mycobacteria. During my post-doc, I developed a model of viral infections in drosophila. Subsequently, as Team Leader in the Laboratory of Human Genetics of Infectious Diseases, I delved into the genetic basis of severe viral infections. I contributed to the identification of single-gene IEI in the TLR3-IFN- α/β pathway that were associated with herpes simplex encephalitis (HSE), the most common sporadic viral encephalitis in Western countries. I spearheaded investigations into selective natural killer (NK) cell deficiency (NKD), which revealed novel insights into the role of NK cells in host defense against viruses, particularly herpes viruses. My team identified genetic mutations in *MCM4* and *GINS1*, highlighting the importance of DNA integrity in NK cell development and maturation. Expanding our research to include cutaneous infections, we identified CIB1 deficiency as the third genetic etiology of isolated epidermodysplasia verruciformis (EV), CIB1 being a new partner of EVER1 and EVER2 and the first HPV restriction factor. We also reported RhoH and STK4 deficiencies as the first T cell defects associated with syndromic EV. We established a cohort of patients with recurrent respiratory papillomatosis (RRP), mainly due to alpha-HPV6 and 11, a rare and severe disease with frequent surgeries as the only available treatment. We reported gain-of-function mutation in *NLRP1* in one multiplex family with syndromic RRP. We also turned our attention to liver immunity, investigating severe liver-related conditions, including yellow fever vaccine-associated viscerotropic disease (YEL-AVD), uncovering the first genetic etiology of YEL-AVD, with a complete autosomal recessive (AR) defect in type I IFNs receptor chains or neutralizing auto-antibodies against type I IFNs. Lastly, we investigated patients with fulminant viral hepatitis (FVH), a rare, life-threatening, and characterized by a massive necrosis of the liver in individuals without any previous history of liver disease. We reported *IL18BP* and *IL10RB* deficiencies as the first genetic etiologies of FVH. These important findings not only enhanced our understanding of immune responses to viral infections, but also provide crucial insights into the genetic mechanisms underlying susceptibility to various infectious diseases.

C. POSITIONS AND HONORS

- 2013 - *Guest Investigator & Team Leader*, St. Giles Laboratory of Human Genetics of Infectious Diseases (head: Jean-Laurent Casanova), Rockefeller University, USA
Associate Professor & Theme Leader, Laboratory of Human Genetics of Infectious Diseases, Imagine institute, University of Paris Cité and INSERM, France
2009 - 2011 *Research Associate & Team Leader*, St. Giles Laboratory of Human Genetics of Infectious Diseases, Rockefeller University, USA
2008 *Visiting Assistant Professor*, Center for the Study of Hepatitis C (head: Charles M. Rice), Rockefeller University, USA
2001 - 2012 *Assistant Professor*, Laboratory of Human Genetics of Infectious Diseases, Paris René Descartes University and INSERM, France
1999 - 2001 *Post-Doctoral Fellow*, Laboratory of Immune Response and Development of *Drosophila melanogaster* (head: Jules A. Hoffmann), CNRS and Strasbourg Louis Pasteur University, France

Honors

- 2019 Prix Charles-Louis de Saulses de Freycinet, Académie Française des Sciences
2018 Elected Member, Henry Kunkel Society
2014 Prix Lucien Tartoix, Fondation de la Recherche Médicale
2007 Translational research contract INSERM-AP-HP

D. CONTRIBUTION TO SCIENCE

Complete List of Published Work in My Bibliography:

<https://www.ncbi.nlm.nih.gov/sites/myncbi/emmanuelle.jouanguy.1/bibliography/48476996/public>

Web of Sciences Citations: 18,833, H-index: 66