

Stéphanie BAULAC, PhD | CV

Group leader (Mosaic Lab) at Paris Brain Institute/Institut du Cerveau (ICM)

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ORCID N°: 0000-0001-6430-4693

Nationality: French

EDUCATION

2008: Habilitation to supervise research, Sorbonne University

2001: PhD in Neurogenetics, University Paris Cité

RESEARCH EXPERIENCE

2023- Research director (INSERM DR1) & Group leader at ICM (Mosaic Team)

2013-2022 Research director (INSERM DR2) & Group leader at ICM (Mosaic Team)

2009-2013: Senior Researcher (INSERM CR1), ICM, Paris

2009-2010: Visiting Researcher, Kyoto University, Japan

2005-2009: Junior Researcher (INSERM CR2), Pitié-Salpêtrière hospital, Paris

2002-2005: Postdoctoral fellow, Harvard Medical School, Boston, USA

1998-2001: PhD training, UMR289, Pitié-Salpêtrière hospital, Paris

RESEARCH INTERESTS

Dr Stéphanie Baulac's research focuses on somatic mosaicism and cortical malformations associated with focal epilepsies. Using a translational approach from genetic studies capitalizing on a large biobank of resected epileptic brain tissues, to *in vitro* functional testing using patient-derived cortical organoids, and *in utero*-based mouse models, the ultimate goal is to provide insights into targeted therapeutic opportunities. Aims are to: (1) identify new causative epilepsy genes; (2) unveil molecular and cellular mechanisms underlying neurodevelopmental epilepsies using cortical organoids and mouse models; (3) determine the cell-type specific determinants underlying mosaic neurodevelopmental disorders using human tissues; (4) investigate targeted rescue strategies for therapeutic purposes.

MAJOR RESEARCH FUNDINGS

2024 Hope Foundation for Hypothalamic Hamartomas

2024 Fund Victoria on Autism & Epilepsy

2023 ERC Proof of Concept

2022 FRM Equipe 2022

2021 ERA-Net NEURON (coordinator)

2021 National Research Agency (ANR coordinator)

2020 ANR - IA - RHU

2020 AXA Research Funds

2016 European Research Council (ERC), consolidator

2015 Fondation pour la Recherche Médicale, FRM équipe label

2010 National Research Agency (ANR-JCJC)

AWARDS AND HONORS

2025	Fondation Roger de Spoelberch Prize
2024	Lamonica Prize in Neurology from the French Academy of Science
2019	Michael Prize Award (International Prize in Epileptology)
2019	Camille Woringer Prize (FRM)
2019	Grand Prix Robert Debré pour la recherche médicale
2015	National medical translational stipend with Paris Hospital, AP-HP (3-year)
2009	Invitational Fellowships for Research in Japan (Long-term JSPS)
2014	Prize from the French Foundation for Research in Epilepsy (Prix Valérie Chamaillard)
2013	INSERM Prize for scientific excellence
2012	Young investigator award at the 10th European Congress on Epileptology
2003	Prize from the Simone et Cino Del Duca Foundation
2002	Postdoctoral fellowship from the Singer-Polignac Foundation
2000	Prize for best contribution in basic sciences at 4th European Congress Epileptology

INSTITUTIONAL RESPONSIBILITIES AND SCIENTIFIC COMMITTEES

2025-2029	Member of the Precision Therapy for Epilepsy Task Force of the Neurobiology Commission of the ILAE
2025-	Member of the scientific board of Plateforme Génomique APHP
2024	Member of the scientific board of ASTB (Association Sclérose Tubéreuse de Bourneville)
2022-	Member of the College des experts de l'ITMO Neurosciences (AVIESAN)
2022-25	Committee member of the SFN Award for Education in Neuroscience
2022-	Member of the SAB of the Epilepsy Research Centre Prague
2020-	Member of scientific board of French Foundation for Research in Epilepsy (FFRE)
2019-	Member of the Scientific and Medical Steering Committee ("COFIL") of ICM
2015-2025	Scientific director of the sequencing core facility of ICM (iGenSeq)
2012-	Member of scientific board of French League against Epilepsy

EDITORIAL BOARD COMMITTEES

2021-	Member of the Editorial Advisory Board of <i>Oxford Open Neuroscience</i>
2019-2022	Member of the Editorial Board of <i>Annals of Neurology</i>
2019-	Member of the Editorial Board of <i>Annals of Child Neurology</i>
2019-	Member of the Editorial Board of <i>EBioMedicine</i>

REVIEWING ACTIVITIES

Grant reviews: ERC H2020 EU, MSCA H2020 EU, Era-Net NEURON H2020 EU, FWO-FNRS (Belgium); Telethon (Italy); ANR (France); FWF (Austria); "La Caixa" Foundation Research (Spain); European Science Foundation; SFI (Ireland), UK Epilepsy Research, ...

Journal referee (selection): Science, Nature, Nat. Genet, Nat. Com., Hum. Mol. Genet., J. Clin Invest, Neurology, Brain, Annals of Neurol., Am J. of Hum Genet, Scientific Report, Epilepsia, J. Med Genet., J. Neuroscience, The JCI, Neurobiology of Disease, etc.

TEACHING EXPERIENCE

Lecturer in several master's and doctoral programs of the University Paris Cité and 7 (10 hrs/yr); 4EU+ Course on the Pathogenesis of Epilepsy. Member of >20 PhD thesis and Research Habilitation committees in France and Abroad.

CAREER BREAKS

3 maternity leaves (2006, 2008, 2011)

INVITED PRESENTATIONS IN MEETINGS & SEMINARS

Over the last decade, I have delivered > than **50 invited lectures at international conferences** including FENS, SFN, EHSG, International Epilepsy Congress, American Epilepsy Society (AES), Keystone Conference (2025), Gordon Research Conference (2024, 2022)), gave > 20 seminars and chaired scientific sessions at the AES congress and SFN and was part of the organizing committee (TOR de France 2018, 2020, 2023, 2025; JFE 2020; FENS 2022, WONOEP 2022).

SELECTED PUBLICATIONS (10 of 134 publications, >14,000 citations, h-index 59 Google Scholar)

1. Maletic M, Bizzotto S, Ribierre T, Guerdoud K, Raoux C, Doladilhe M, Dalle C, Picard F, Baulac S. Mosaic human cortical organoids model mTOR-related focal cortical dysplasia through DEPDC5 loss-of-function. *Brain*. 2026. Mar 6:awag086. doi: 10.1093/brain/awag086
2. Baldassari S, Klingler E, Gomez Teijeiro L, Doladilhe M, Raoux C, Roig Puiggros S, Bizzotto S, Sami L, Ribierre T, Aronica E, Adle-Biassette H, Chipaux M, Jabaudon J and Baulac S. Single-cell genotyping and transcriptomic profiling in focal cortical dysplasia. *Nat Neurosci*. 2025. doi.org/10.1038/s41593-025-01936-z
3. Ribierre T, Bacq A, Baldassari S, Maletic M, Roussel D, Donneger F, Doladilhe M, Le Roux I, Chassoux F, Devaux B, Adle-Biassette H, Ferrand-Sorbets S, Dorfmueller G, Chipaux M, Poncer JC and Baulac S. Targeting pathogenic cells reduces seizures in neurodevelopmental mTOR-related epilepsy. *Nat Neurosci*. 2024 Jun;27(6):1125-1136. *Highlighted in Nat Rev Drug Discov. 2024*.
4. Blümcke I, Budday S, Poduri A, Lal D, Kobow K and Baulac S (2021). Neocortical development and epilepsy: insights from focal cortical dysplasia and brain tumours (review). *Lancet Neurology* Nov;20(11):943-955.
5. Baldassari S, Ribierre T, Marsan E, Adle-Biassette H, Ferrand-Sorbets S, Bulteau C, Dorison N, Fohlen M, Polivka M, Weckhuysen S, Dorfmueller G, Chipaux M, and Baulac S (2019). Dissecting the genetic basis of focal cortical dysplasia: a large cohort study. *Acta Neuropathologica*, Dec;138(6):885-900
6. Ribierre T, Deleuze C, Bacq A, Baldassari S, Marsan E, Chipaux M, Muraca G, Roussel D, Navarro V, Leguern E, Miles R and Baulac S (2018). Second-hit mosaic mutation in mTORC1 repressor DEPDC5 causes focal cortical dysplasia-associated epilepsy. *Journal of Clinical Investigation*, Jun 1; 128(6): 2452-2458. *Highlighted by the editor in the "JCI this month"*.
7. Baulac S, Ishida S, Marsan E, Miquel C, Biraben A, Nguyen DK, Nordli D, Cossette P, Nguyen S, Lambrecq V, Vlaicu M, Daniau M, Bielle F, Andermann E, Andermann F, Leguern E, Chassoux F, Picard F (2015). Familial focal epilepsy with focal cortical dysplasia due to DEPDC5 mutations. *Annals of Neurology*. Apr;77(4):675-83. *Highlighted as Best Advances of 2015: Picks from the Neurology Today Editorial Advisory Board*
8. Ishida S, Picard F, Rudolf G, Noé E, Achaz G, Thomas P, Genton P, Mundwiler E, Wolff M, Marescaux C, Miles R, Baulac M, Hirsch E, Leguern E and Baulac S (2013). Mutations of DEPDC5 cause autosomal dominant focal epilepsies. *Nature Genetics*, Apr 26;45(5):552-5. *Highlighted in Nature Review Neurology*
9. Baulac S, Huberfeld G, Gourfinkel-An I, Mitropoulou G, Beranger A, Prud'homme JF, Baulac M, Brice A, Bruzzone R and Leguern E (2001). First genetic evidence of GABA_A receptor dysfunction in epilepsy: a mutation in the gamma 2 subunit gene. *Nature Genetics*; 28:46-8.
10. Escayg A*, Baulac S*, Moulard B*, MacDonald BT, Meisler MH, Huberfeld G, An-Gourfinkel I, Brice A, Leguern E, Chaigne D, Buresi C and Malafosse A (2000). Mutations of SCN1A, encoding a neuronal sodium channel, in two families with GEFS+2. *Nature Genetics*; 24:343-5. (*equal contribution)