BIOGRAPHICAL SKETCH

NAME: **THOMAS BOURGERON (born in Paris on November 9, 1965)**

POSITION TITLE: **PROFESSOR AT UNIVERSITE DE PARIS**

ORCID number: **0000-0001-8164-9220**

EDUCATION/TRAINING

INSTITUTION AND LOCATION	DEGREE	Completion Date	FIELD OF STUDY
Université de Paris, Paris	Habilitation to lead research (HDR)	2003	Genetics of Psychiatric Disorders
University Paris 6, Paris	Thesis PhD	1994	Genetics of Mitochondrial Disorders
University Paris 6, Paris	Master 2	1990	Plant Biology

A. Personal Statement

My laboratory is affiliated to the Centre National Recherche Scientifique (CNRS)/Université de Paris Cité/Institut Pasteur and is part of the Neuroscience Department of the Institut Pasteur. It gathers psychiatrists, neuroscientists and geneticists aiming at better understanding the causes of autism and neurodevelopmental disorders (NDD). We previously identified the first mutations in autism of the NLGN-NRXN-SHANK pathway, which has a key role in synapse formation within the brain. Our results highlight the genetic heterogeneity of autism, but also point at common biological mechanisms that could constitute relevant targets for new treatments. I coordinate R2D2-Mental Health, a new EU funded project which aims to identify both **R**isk and **R**esilience factors for NDD and co-develop with the stakeholder new measures of Mental Health more align with the concept of **D**evelopmental **D**iversity. I am the principal investigator of the genetic and database work packages of the European AIMS2-TRIALS and CANDY projects on autism research. These projects are focused on individuals with autism, NDD and controls with deep-phenotyping data including brain imaging and a battery of cognitive tests. Our group is developing methods to analyze whole genome and brain imaging data and is developing new paradigms for characterizing mouse social and vocal behaviors (see on our website). I am also the PI of INCEPTION, an Institut Pasteur project that applies integrative biology and social sciences to understand the emergence of diseases in populations and individuals.

B. Laboratory

Human Genetics and Cognitive Functions Unit; Institut Pasteur, 25 rue du Docteur Roux; 75724 Paris Cedex 15, France; Tel: + 33 1 40 61 32 16; email: thomasb@pasteur.fr; website: https://research.pasteur.fr/en/team/human-genetics-and-cognitive-functions/

C. Positions and Honors

Member of the French Academy of Sciences, the European Molecular Biology Organisation (EMBO), the *Academia Europaea* and the National Ethical Committee (CCNE); Asperger-Kanner medal 2024, Duquesne Prize 2023, Roger De Spoelberch Prize Prize 2020; IPSEN Prize for Neuronal Plasticity 2015; Chair of excellence of the Foundation Bettencourt-Schueller 2012 and 2017; Elected INSAR Fellow (International Society for Autism Research) and chairman of the scientific committee for IMFAR 2013; Vallery Radot award from Institut Pasteur and the French Academy of Sciences, 2008; Lacassagne award from the Collège de France, 2007; Jean Bernard award of the victories of medicine 2007; French Academy of Sciences award 2007 (Biological discoveries of 2007); Awarded ENI (European Neuroscience Institutes) Young Investigator (2005)

D. Main Grants

2023: ANR AUTISMSHANK (722 k€/ 3 years)
2022: R2D2-Mental Health; Horizon Europe Framework Programme (HORIZON) (1,081 k€ /5 years)
2021: ROGER DE SPOELBERCH Prize (598,5 k€)
2020-2024: H2020 CANDY (366 k€ /5 years)
2020-2023: ERA-NET Neuron ALTRUISM (200k€ / 4 years)
2018-2022: IMI2 AIMS2-TRIALS (900 k€ /5 years)
2017-2026: INCEPTION (12 M€ /10 years)
2017-2022: Chair of excellence Foundation Bettencourt Schueller (1,500 k€ / 5 Years)
2016-2020: H2020 COSYN (225€ / 5 years)

E. Conferences since 2017 (selected)

2024. Kanner-Medal Award, Frankfurt Germany – La semaine du Cerveau, Geneva, Switzerland – SFARI Gene meeting, USA New York. **2023** SFARI Investigator Meeting – USA New York EuroNDD 2023 – Netherlands Amsterdam 17th Troina Meeting on Genetics of Neurodevelopmental Disorders – Italy Troina International Society for Autism Research (INSAR) Annual Meeting – Sweden Stockholm Genomics of Brain Disorders – UK Hinxton NeuroFrance 2023 – France Lyon Crick-Pasteur Neuro-Immune Workshop – UK London ; **2022** 11e Assises de Génétique Humaine et Médicale – France Rennes Simons Foundation Autism Research Initiative (SFARI) Investigator meeting – USA New York Bordeaux Neurocampus Conference : Neurodevelopmental disorders – France Bordeaux ALTRUISM Consortium Meeting – Italy Milan PMS Consensus Meeting – Netherlands Groningen CSHL - Genetics & Neurobiology of Language 2022 – USA Woodbury AIMS-2-TRIALS General Assembly – Portugal Lisbon Harvard Conte Center Conference – USA Boston SYNGO2022 – USA New York Utopiales – France Nantes Max Planck Institut Colloquium – Netherlands Nijmegen ; **2021** XXVe colloque ACLF 2021 (Association des Cytogénéticiens de Langue Française – France Grenoble The Synaptic Dimension of Brain Disorders Workshop – Spain Baeza ; **2020** 10e Assises de Génétique Humaine et Médicale – France Tours COSYN Annual Meeting – Spain Barcelona 3rd Conference on the Neurobiology of Mental Health – Switzerland Geneva Ecole de Recherche Translationnelle en Neurosciences – France Langon ; **2019** COSYN Meeting – Malta Valletta 14th Troina Meeting on Genetics of Neurodevelopmental Disorders – Italy Troina International Society for Autism Research (INSAR) Annual Meeting – Canada Montreal NeuroFrance 2019 – France Marseille Phelan McDermid Syndrom Foundation Conference – France Nice AIMS-2-TRIALS General Assembly – Spain Fuerteventura Austime Europe International Congress – France Nice CSR and Autism Forum – CHINA Beijing A new beginning for neurdevelopment disorders: science-based intervention for individual patients Colloquium – Netherlands Amsterdam World Congress of Psychiatric Genetics (WCPG) – USA Los Angeles Max Planck Institute for Psycholinguistics SAB Meeting – Netherlands NIjmegen European Molecular Biology Laboratory Meeting – Italy Rome FHU NeuroGenycs Seminar – France Strasbourg ; **2018** ESSENCE Conference – Sweden Gothenburg 13th Troina Meeting on Genetics of Neurodevelopmental Disorders – Italy Troina MIT'Science Foundation – USA Boston The Inaugural Transforming Autism Care Consertium Symposium – Canada Montreal Symposium "New Horizons in Biology and Health" – France Montpellier SynPathy Mid-Term Meeting – Belgium Leuven 9e Assises de Génétique Humaine et Médicale – France Nantes SIKON Conference – Denmark Odense International Society for Autism Research (INSAR) Annual Meeting – Netherlands Rotterdam National Autism Conference – NORWAY Tønsberg AIMS-2-TRIALS Annual Meeting – Germany Munich Towards Understanding "INDIVIDUALITY" Symposium – JAPAN Kyoto Radboud Research Rounds Plus 2018 – Netherlands Nijmegen ; **2017** European College of Neuropsychopharmacology Congress (ECNP) - France Nice International Convention of Psychological Science (ICPS) – Austria Vienna International Society for Autism Research (IMFAR) Annual Meeting – USA San Francisco NeuroFrance 2017 – France Bordeaux AIMS-2-TRIALS Ice-Breaker Meeting – Belgium Brussels COSYN meeting – Germany Berlin SFARI Investigator Meeting – USA New York

F. 10 significant publications

Rolland T, Cliquet F, Anney R, Moreau C, Traut N, Mathieu A, Huguet G, Duan J, Warrier V, Portalier S, Dry L, Leblond CS, Douard E Amsellem F, Malesys S, Maruani A, Toro R, Børglum AD, Grove J, Baron-Cohen S, Packer A, Chung WK, Jacquemont S, Delorme R, Bourgeron T. Phenotypic effects of genetic variants associated with autism. **Nature Medicine** 2023 ;29(7):1671-1680.

Warrier W, Zhang X, Reed P, Havdahl A, Moore TM, Cliquet F, Leblond CS, Rolland T, Rosengren A, EU-AIMS-LEAP, iPSYCH-Autism Working Group, Spectrum 10K and APEX Consortium, Rowitch DH, Hurles ME, Geschwind DH, Børglum AD, Robinson EB, Grove J, Martin HC, Bourgeron T*, Baron-Cohen S*. Genetic correlates of phenotypic heterogeneity in autism. **Nature Genetics** 2022 54(9):1293-1304 *senior authors

de Chaumont F, Ey E, Torquet N, Lagache T, Dallongeville S, Imbert A, Le Sourd AM, Faure P*, **Bourgeron T*** and Olivo-Marin JC* Real-time analysis of the behaviour of groups of mice via a depth-sensing camera and machine learning **Nature Biomedical Engineering** 2019 3(11):930-942. * senior authors

Bourgeron T. From the genetic architecture to synaptic plasticity in autism spectrum disorders. **Nature Reviews Neuroscience** (2015) 16, 551–563.

Leblond CS, Nava C, Polge A, Gauthier J, Huguet G, Lumbroso S, Giuliano F, Stordeur C, Depienne C, Mouzat K, Pinto D, Howe J, Lemière N, Durand CM, Guibert J, Ey E, Toro R, Peyre H, Mathieu A, Amsellem F, Rastam M, Gillberg IC, Rappold GA, Holt R, Monaco AP, Maestrini E, Galan P, Heron D, Jacquette A, Afenjar A, Rastetter A, Brice A, Devillard F, Assouline B, Laffargue F, Lespinasse J, Chiesa J, Rivier F, Bonneau D, Regnault B, Zelenika D, Delepine M, Lathrop M, Sanlaville D, Schluth-Bolard C, Edery P, Perrin L, Tabet AC, Schmeisser MJ, Boeckers TM, Coleman M, Sato D, Szatmari P, Scherer SW, Rouleau GA, Betancur C, Leboyer M, Gillberg C, Delorme R, **Bourgeron T.** Meta-analysis of SHANK mutations in Autism Spectrum Disorders: A gradient of severity in cognitive impairments. **PLOS Genetics**. (2014) 10, e1004580.

Delorme R, Ey E, Toro R, Leboyer M, Gillberg C, and **Bourgeron T**. Progress towards treatments for synaptic defects in autism. **Nature Medicine** (2013) 19:685-94.

Schmeisser MJ, Ey E, Kuebler A, Bockmann J, Wegener S, Stempel AV, Kuebler A, Janssen AL, Udvardi PT, Shiban E, Spilker C, Balschun D, Skryabin BV, tom Dieck S, Smalla KH, Montag D, Leblond CS, Faure P, Torquet N, Le Sourd AM, Toro R, Grabrucker AM, Shoichet SA, Schmitz D, Kreutz MR, **Bourgeron T,** Gundelfinger ED and Boeckers TM. (2012) Hyperactivity and autistic-like behaviours in mice lacking ProSAP1/Shank2. **Nature** (2012) 486: 256-60

Pinto D, Pagnamenta A, Klei L Merico D, Anney R, Merico D, Regan R, Conroy J, Magalhaes T, Correia C, Abrahams BS, Almeida J, Bacchelli E, Bader GD, Bailey AJ, Baird G, Battaglia A, Berney T, Bolshakova N, Bölte S, Bolton PF, **Bourgeron T** et al. Functional impact of global rare copy number variation in autism. **Nature** (2010) 466: 368-72.

Durand C, Betancur C, Boeckers TM, Bockmann J, Chaste P, Fauchereau F, Nygren G, Rastam M, Gillberg IC, Anckarsäter H, Sponheim E, Goubran-Botros H, Delorme R, Chabane N, Mouren-Simeoni MC, de Mas P, Bieth E, Rogé B, Héron D, Burglen L, Gillberg C, Leboyer M, **Bourgeron T** Mutations of the synaptic scaffolding protein SHANK3 are associated with autism spectrum disorders. **Nature Genetics** (2007) 39:25-7.

Jamain S, Quach H, Betancur C, Råstam M, Colineaux C, Gillberg IC, Soderstrom H, Giros B, Leboyer M, Gillberg C, **Bourgeron T**. Mutations of the X-linked neuroligins NLGN3 and NLGN4 are associated with autism. **Nature Genetics** (2003) 34, 27-29.

G. Book

T. Bourgeron. Des gènes, des synapses, des autismes. https://www.odilejacob.fr/catalogue/sciences/genetique/des-genes-des-synapses-des-autismes_9782415003906.php

H. Full Publication list (ORCID number **0000-0001-8164-9220**).

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200. Black MH, Remnélius KL, Alehagen L, Bourgeron T, Bölte S. From Symptomatology to Functioning - Applying the ICF to Autism Measures to Facilitate Neurodiversity-Affirmative Data Harmonization. J Autism Dev Disord. 2023 Dec 11

199. Lefebvre A, Traut N, Pedoux A, Maruani A, Beggiato A, Elmaleh M, Germanaud D, Amestoy A, Ly-Le Moal M, Chatham C, Murtagh L, Bouvard M, Alisson M, Leboyer M, Bourgeron T, Toro R, Dumas G, Moreau C, Delorme R. Exploring the multidimensional nature of repetitive and restricted behaviors and interests (RRBI) in autism: neuroanatomical correlates and clinical implications. Mol Autism. 2023 Nov 27;14(1):45.

198. Dejean C, Dupont T, Verpy E, Gonçalves N, Coqueran S, Michalski N, Pucheu S, Bourgeron T, Gourévitch B. Detecting Central Auditory Processing Disorders in Awake Mice.Brain Sci. 2023 Oct 31;13(11):1539

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196. COVID-19 Host Genetics Initiative. A second update on mapping the human genetic architecture of COVID-19. Nature. 2023 Sep;621(7977):E7-E26. doi: 10.1038/s41586-023-06355-3.

195. Heraty S, Lautarescu A, Belton D, Boyle A, Cirrincione P, Doherty M, Douglas S, Plas JRD, Van Den Bosch K, Violland P, Tercon J, Ruigrok A, Murphy DGM, **Bourgeron T**, Chatham C, Loth E, Oakley B, McAlonan GM, Charman T, Puts N, Gallagher L, Jones EJH. Bridge-building between communities: Imagining the future of biomedical autism research. Cell. 2023 Aug 31;186(18):3747-3752.

194. Isakoglou C, Haak KV, Wolfers T, Floris DL, Llera A, Oldehinkel M, Forde NJ, Oakley BFM, Tillmann J, Holt RJ, Moessnang C, Loth E, **Bourgeron T**, Baron-Cohen S, Charman T, Banaschewski T, Murphy DGM, Buitelaar JK, Marquand AF, Beckmann CF. Fine-grained topographic organization within somatosensory cortex during resting-state and emotional face-matching task and its association with ASD traits. Transl Psychiatry. 2023 Jul 27;13(1):270. doi: 10.1038/s41398-023-02559-3.

193. Gaudfernau F, Lefebvre A, Engemann DA, Pedoux A, Bánki A, Baillin F, Landman B, Maruani A, Amsellem F, **Bourgeron T**, Delorme R, Dumas G. Cortico-Cerebellar neurodynamics during social interaction in Autism Spectrum Disorders. Neuroimage Clin. 2023 Jun 28;39:103465.

192. Rolland T, Cliquet F, Anney R, Moreau C, Traut N, Mathieu A, Huguet G, Duan J, Warrier V, Portalier S, Dry L, Leblond CS, Douard E Amsellem F, Malesys S, Maruani A, Toro R, Børglum AD, Grove J, Baron-Cohen S, Packer A, Chung WK, Jacquemont S, Delorme R, **Bourgeron T**. Phenotypic effects of genetic variants associated with autism. *Nature Medicine* 2023 doi: 10.1038/s41591-023-02408-2

191. Leyhausen J, Schäfer T, Gurr C, Berg LM, Seelemeyer H, Pretzsch CM, Loth E, Oakley B, Buitelaar JK, Beckmann CF, Floris DL, Charman T, **Bourgeron T**, Banaschewski T, Jones EJ, Tillmann J, Chatham C; EU-AIMS LEAP Group; Murphy D, Ecker C. Differences in Intrinsic Gray-Matter Connectivity and their genomic underpinnings in Autism Spectrum Disorder. Biol Psychiatry. 2023 Jun 20:S0006-3223(23)01363-X. doi: 10.1016/j.biopsych.2023.06.010.

190. Ferhat AT, Verpy E, Biton A, Forget B, De Chaumont F, Mueller F, Le Sourd AM, Coqueran S, Schmitt J, Rochefort C, Rondi-Reig L, Leboucher A, Boland A, Fin B, Deleuze JF, Boeckers TM, Ey E, **Bourgeron T.** Excessive self-grooming, gene dysregulation and imbalance between the striosome and matrix compartments in the striatum of Shank3 mutant mice. *Front Mol Neurosci.* 2023 Mar 16;16:1139118.

189. Schön M, Pablo L, Julián N, Mattina T, Gunnarsson C, Hadzsiev K, Verpelli C, **Bourgeron T**, Sarah J, van Ravenswaaij-Arts CMA; European Phelan-McDermid syndrome consortium; Hennekam RC. Definition and clinical variability of SHANK3-related Phelan-McDermid syndrome. *Eur J Med Genet.* 2023 Mar 30:104754.

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187. de Coo IFM, Jesse S, Le TL, Sala C; European Phelan-McDermid syndrome consortium. Consensus recommendations on Epilepsy in Phelan-McDermid syndrome. *Eur J Med Genet.* 2023 Mar 24;66(6):104746.

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AC, **Bourgeron T**. Dissecting the 22q13 region to explore the genetic and phenotypic diversity of patients with Phelan-McDermid syndrome. *Eur J Med Genet.* 2023 May;66(5):104732.

185. Baumeister S, Moessnang C, Bast N, Hohmann S, Aggensteiner P, Kaiser A, Tillmann J, Goyard D, Charman T, Ambrosino S, Baron-Cohen S, Beckmann C, Bölte S, **Bourgeron T**, Rausch A, Crawley D, Dell'Acqua F, Dumas G, Durston S, Ecker C, Floris DL, Frouin V, Hayward H, Holt R, Johnson MH, Jones EJH, Lai MC, Lombardo MV, Mason L, Oakley B, Oldehinkel M, Persico AM, San José Cáceres A, Wolfers T, Loth E, Murphy DGM, Buitelaar JK, Tost H, Meyer-Lindenberg A, Banaschewski T, Brandeis D; EU-AIMS LEAP Group. Processing of social and monetary rewards in autism spectrum disorders. *Br J Psychiatry*. 2023 Mar;222(3):100-111.

184. Hollestein V, Poelmans G, Forde NJ, Beckmann CF, Ecker C, Mann C, Schäfer T, Moessnang C, Baumeister S, Banaschewski T, **Bourgeron T**, Loth E, Dell'Acqua F, Murphy DGM, Puts NA, Tillmann J, Charman T, Jones EJH, Mason L, Ambrosino S, Holt R, Bölte S, Buitelaar JK, Naaijen J. Excitatory/inhibitory imbalance in autism: the role of glutamate and GABA gene-sets in symptoms and cortical brain structure. *Transl Psychiatry.* 2023 Jan 21;13(1):18.

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Beckmann C, **Bourgeron T**, Toro R, Buitelaar J, Murphy D, Dumas G. The meaning of significant mean group differences for biomarker discovery. *PLoS Comput Biol.* 2021 Nov 18;17(11):e1009477.

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