BIOGRAPHICAL SKETCH

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

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

EDUCATION/TRAINING

INSTITUTION AND LOCATION	DEGREE	Completion Date	FIELD OF STUDY
Université de Paris, Paris	Professor	2005	Human Genetics
Université de Paris, Paris	HDR	2003	Genetics of Psychiatric Disorders
University Paris 6, Paris	Ph.D	1994	Genetics of Mitochondrial Disorders
University Paris 6, Paris	Master	1990	Plant Biology

A. Personal Statement

My laboratory is affiliated to the CNRS/Université de Paris/Institut Pasteur and is part of the Neuroscience Department of Institut Pasteur. It gathers psychiatrists, neuroscientists and geneticists to understand the causes of autism. We previously identified the first mutations in 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 am the principal investigator of the genetic and database work packages of AIMS2-TRIALS and CANDY, the largest European projects on autism research. The projects are focused on deep-phenotyping of individuals with autism or neurodevelopmental disorders (NDD) and controls including brain imaging (EEG and MRI) and a battery of cognitive tests. Our group is currently developing new methods for analyzing whole genome and brain imaging data as well as new paradigms for characterizing mouse social and vocal behaviors (see on our website). I am also the PI of *INCEPTION*, a project at Institut Pasteur 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); ROGER DE SPOELBERCH Prize 2021; 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

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. 10 publications

de Chaumont F, Ey E, Torquet N, Lagache T, Dallongeville S, Imbert A, Le Sourd AM, Faure P*, **Bourgeron T*** and Olivo-Marin JC* Live

Mouse Tracker : real-time behavior analysis of group of mice. **Nature Biomedical Engineering** 2019 3(11):930-942. *equally contributed to the work as senior authors

Huguet G, Schramm C, Douard E, Jiang L, Labbe A, Tihy F, Mathonnet G, Nizard S, Lemyre E, Mathieu A, Poline JB, Loth E, Toro R, Schumann G, IMAGEN Consortium; Conrod P, Pausova Z, Greenwood C, Paus T, **Bourgeron T***, Sébastien J* Measuring and predicting the effect of copy number variants on general intelligence in community-based samples. **JAMA Psychiatry** 2018 75(5):447-457. *equally contributed to the work as senior authors

Mercati O, Huguet G, Danckaert A, André-Leroux G, Maruani A, Bellinzoni M, Rolland T, Gouder L, Mathieu A, Buratti J, Amsellem F, Benabou M, Van-Gils J, Beggiato, Konyukh M, Bourgeois J-P, Gazzellone M, Yuen RKC, Walker S, Delépine M, Boland A, Régnault B, Francois M, Van Den Abbeele T, Mosca-Boidron AL, Faivre L, Shimoda Y, Watanabe K, Bonneau D, Rastam M, Leboyer M, Scherer S, Gillberg C, Delorme R, Cloët-Tayarani I and **Bourgeron T.** CNTN6 mutations are risk factors for abnormal auditory sensory perception in autism spectrum disorders. **Molecular Psychiatry** 2017 22(4):625-633.

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.

Full Publication list (>150 publications; Web of Science: h-index 54; 15 293 citations, average citations per item 68)

2021

Dumas G, Malesys, **Bourgeron T**. Systematic detection of brain protein-coding genes under positive selection during primate evolution and their roles in cognition. *Genome Research In press*

Huguet G, Schramm C, Douard E, Petra T, Main A, Monin P, England J, Jizi K, Renne T, Poirier M, Nowak S, Martin CO, Younis N, Knoth IS, Martineau JL, Saci Z, Auger M, Tihy F, Mathonnet G, Maftei C, Léveillé F, Porteous D, Davies G, Redmond P, Harris SE, Hill WD, Lemyre E, Schumann G, **Bourgeron T**, Pausova T, Paus T, Karama S, Lippe S, Deary IJ, Almasy L, Labbe A, Glahn D, Greenwood C, Jacquemont S. Genome wide analysis of gene dosage in 24,092 individuals shows that 10,000 genes modulate cognitive ability. *Molecular Psychiatry* In press.

Pagan C, Benabou M, Leblond CSL, Cliquet F, Mathieu A, Lemière N, Goubran-botros H, Delorme R, Leboyer M, Callebert J, **Bourgeron T***. Launay JM* Decreased phenol sulfotransferase activities associated with hyperserotonemia in Autism Spectrum Disorders. *Trans Psychiatry* 11:23.*equally contributed to the work as senior authors.

Douard E, Zeribi A, Schramm C, Tamer P, Loum MA, Nowak S, Saci Z, Lord MP, Rodríguez-Herreros B, Jean-Louis M, Moreau C, Loth E, Schumann G, Pausova Z, Elsabbagh M, Almasy L, Glahn DC, **Bourgeron T**, Labbe A, Paus T, Mottron L, Greenwood CMT, Huguet G, Jacquemont S. Effect Sizes

of Deletions and Duplications on Autism Risk Across the Genome. *Am J Psychiatry.* 2021 178(1):87-98.

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156. Zabihi M, Floris DL, Kia SM, Wolfers T, Tillmann J, Arenas AL, Moessnang C, Banaschewski T, Holt R, Baron-Cohen S, Loth E, Charman T, **Bourgeron T**, Murphy D, Ecker C, Buitelaar JK, Beckmann CF, Marquand A; EU-AIMS LEAP Group. Fractionating autism based on neuroanatomical normative modeling. *Transl Psychiatry.* 2020 Nov 6;10(1):384.

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153. Ayrolles A, Brun F, Chen P, Djalovski A, Beauxis Y, Delorme R, **Bourgeron T**, Dikker S, Dumas G. HyPyP: a Hyperscanning Python Pipeline for inter-brain connectivity analysis. *Soc Cogn Affect Neurosci.* 2020 Oct doi: 10.1093/scan/nsaa141. PMID: 33031496

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149. Lin BD, Colas F, Nijman IJ, Medic J, Brands W, Parr JR, van Eijk KR, Klauck SM, Chiocchetti AG, Freitag CM, Maestrini E, Bacchelli E, Coon H, Vicente A, Oliveira G, Pagnamenta AT, Gallagher L, Ennis S, Anney R, **Bourgeron T**, Luykx JJ, Vorstman J. The role of rare compound heterozygous events in autism spectrum disorder. *Transl Psychiatry.* 2020 Jun 22;10(1):204.

148. Myers SM, Challman TD, Bernier R, **Bourgeron T**, Chung WK, Constantino JN, Eichler EE, Jacquemont S, Miller DT, Mitchell KJ, Zoghbi HY, Martin CL, Ledbetter DH. Insufficient Evidence for "Autism-Specific" Genes. *Am J Hum Genet.* 2020 May 7;106(5):587-595.

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2019

143. Florian et al. Familial Adult Myoclonic Epilepsy linked to chromosome 5p15 is caused by unstable 2 TTTTA/TTTCA expansions in intron 1 of MARCH6. *Nature Communication* 2019 29;10(1):4919.

142. Warrier V, Toro R, Won H, Leblond CS, Cliquet F, Delorme R, de Witte W, Bralten J, Chakrabarti B, EU-AIMS LEAP group, the iPSYCH-Broad autism group, Børglum AD, Grove J, Poelmans G, the 23andMe Research Team, Hinds DA, **Bourgeron T*** and Baron-Cohen S*. Social and non-social autism symptom/trait domains are genetically dissociable. *Communications Biology* 2019 3;2:328.*equally contributed to the work as senior authors.

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139. Pinel P, Forgeot d'Arc B, Dehaene S, **Bourgeron T**, Thirion B, Le Bihan D, Poupon C. The functional database of the ARCHI project: Potential and perspectives. *Neuroimage*. 2019 ;197:527-543.

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C, Turecki G, Gravel S, Mechawar N, Rossignol E, Michaud JL, Lessard J, Ernst C, Campeau PM. Mutations in ACTL6B Cause Neurodevelopmental Deficits and Epilepsy and Lead to Loss of Dendrites in Human Neurons. *Am J Hum Genet.* 2019 ;104(5):815-834.

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