BIOGRAPHICAL SKETCH

NAME: **THOMAS BOURGERON**

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 part of the Neuroscience Department of the Institut Pasteur and 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, the largest European project on autism research. The project is focused on deep-phenotyping of individuals with autism 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 co-PI of *INCEPTION*, a project at the 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); Elected INSAR Fellow (International Society for Autism Research) and chairman of the scientific committee for IMFAR 2013; IPSEN Prize for Neuronal Plasticity, 2015; Chair of excellence of the Foundation Bettencourt-Schueller 2012 and 2017; Vallery Radot award from the Pasteur Institute 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

2020-2024: H2020 CANDY (300 k€ /5 years)
2020-2023: ERA-NET Neuron ALTRUISM (200k€ / 4 years)
2018-2022: IMI2 AIMS2-TRIALS (900 k€ /5 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. *equally contributed to the work as senior authors. **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.

F. Full Publication list (>140 publications; Google Scholar H index: 88; Citations: 38,765)

Papers submitted

Pagan et al. Decreased phenol sulfotransferase activities associated with hyperserotonemia in Autism Spectrum Disorders.

Dumas G, Malesys S, **Bourgeron T**. Systematic detection of divergent brain protein coding-genes in human evolution and their roles in cognition. *bioRxiv* doi: https://doi.org/10.1101/658658

Warrier V, the 23andMe Research Team, **Bourgeron T**, Baron-Cohen S. Genome-wide association study of social relationship satisfaction: significant loci and correlations with psychiatric conditions. *bioRxiv 196071*; doi: https://doi.org/10.1101/196071 *equally contributed to the work as senior authors

2020

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.

147. Ey E, **Bourgeron T**, Boeckers TM, Kim E, Han K. Editorial: Shankopathies: Shank Protein Deficiency-Induced Synaptic Diseases.Front Mol Neurosci. 2020 Feb 7;13:11.

146. Lutz AK, Pfaender S, Incearap B, Ioannidis V, Ottonelli I, Föhr KJ, Cammerer J, Zoller M, Higelin J, Giona F, Stetter M, Stoecker N, Alami NO, Schön M, Orth M, Liebau S, Barbi G, Grabrucker AM, Delorme R, Fauler M, Mayer B, Jesse S, Roselli F, Ludolph AC, **Bourgeron T**, Verpelli C, Demestre M, Boeckers TM. Autism-associated SHANK3 mutations impair maturation of neuromuscular junctions and striated muscles. Sci Transl Med. 2020 Jun 10;12(547):eaaz3267.

145. Ey E, **Bourgeron T**, Boeckers TM, Kim E, Han K. Editorial: Shankopathies: Shank Protein Deficiency-Induced Synaptic Diseases. *Front Mol Neurosci.* 2020 Feb 7;13:11. doi: 10.3389/fnmol.2020.00011. eCollection 2020

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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.

141. de Lombares C, Heude E, Alfama G, Fontaine A, Hassouna R, Vernochet C, de Chaumont F, Olivo-Marin C, Ey E, Parnaudeau S, Tronche F, **Bourgeron T**, Luquet S, Levi G, Narboux-Nême N. Dlx5 and Dlx6 expression in GABAergic neurons controls behavior, metabolism, healthy aging and lifespan." *Aging*. 2019;11(17):6638-6656.

140. de Chaumont F, Ey E, Torquet N, Lagache T, Dallongeville S, Imbert A, Legou T, Le Sourd AM, Faure P*, **Bourgeron T***, 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. *equally contributed to the work as senior authors.

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.

138. Bell S, Rousseau J, Peng H, Aouabed Z, Priam P, Theroux JF, Jefri M, Tanti A, Wu H, Kolobova I, Silviera H, Manzano-Vargas K, Ehresmann S, Hamdan FF, Hettige N, Zhang X, Antonyan L, Nassif C, Ghaloul-Gonzalez L, Sebastian J, Vockley J, Begtrup AG, Wentzensen IM, Crunk A, Nicholls RD, Herman KC, Deignan JL, Al-Hertani W, Efthymiou S, Salpietro V, Miyake N, Makita Y, Matsumoto N, Østern R, Houge G, Hafström M, Fassi E, Houlden H, Klein Wassink-Ruiter JS, Nelson D, Goldstein A, Dabir T, van Gils J, **Bourgeron T**, Delorme R, Cooper GM, Martinez JE, Finnila CR, Carmant L, Lortie A, Oegema R, van Gassen K, Mehta SG, Huhle D, Abou Jamra R, Martin S, Brunner HG, Lindhout D, Au M, Graham JM Jr, Coubes 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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family: Evidence for spatial imagery as a common factor. *Behav Brain Res.* 2019 Apr 19;362:266-272. doi: 10.1016/j.bbr.2019.01.014.

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125. Warrier V, Roberto Toro, Bhismadev Chakrabarti, The iPSYCH-BROAD Autism Group, Jakob Grove, Anders Børglum, The 23andMe Research Team, David Hinds, **Bourgeron T***, and Simon Baron-Cohen*. Genome-wide analyses of self-reported empathy: correlations with autism, schizophrenia, and anorexia nervosa" *Translational Psychiatry* 2018 8(1):35.*equally contributed to the work as senior authors

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2017

123. Benabou M, Rolland T, Leblond CS, Millot GA, Huguet G, Delorme R, Leboyer M, Pagan C, Callebert J, Maronde E, **Bourgeron T.** Heritability of the melatonin synthesis variability in autism spectrum disorders. *Scientific Reports.* 2017 7(1):17746.

122. Charman T, Loth E, Tillmann J, Crawley D, Wooldridge C, Goyard D, Ahmad J, Auyeung B, Ambrosino S, Banaschewski T, Baron-Cohen S, Baumeister S, Beckmann C, Bölte S, **Bourgeron T**, Bours C, Brammer M, Brandeis D, Brogna C, de Bruijn Y, Chakrabarti B, Cornelissen I, Acqua FD, Dumas G, Durston S, Ecker C, Faulkner J, Frouin V, Garcés P, Ham L, Hayward H, Hipp J, Holt RJ, Isaksson J, Johnson MH, Jones EJH, Kundu P, Lai MC, D'ardhuy XL, Lombardo MV, Lythgoe DJ, Mandl R, Mason L, Meyer-Lindenberg A, Moessnang C, Mueller N, O'Dwyer L, Oldehinkel M, Oranje B, Pandina G, Persico AM, Ruggeri B, Ruigrok ANV, Sabet J, Sacco R, Cáceres ASJ, Simonoff E, Toro R, Tost H, Waldman J, Williams SCR, Zwiers MP, Spooren W, Murphy DGM, Buitelaar JK. The EU-AIMS Longitudinal European Autism Project (LEAP): clinical characterisation. *Mol Autism.* 2017 Jun 23;8:27. doi: 10.1186/s13229-017-0145-9.

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