

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](https://orcid.org/0000-0001-8164-9220)

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	PhD	1994	Genetics of Mitochondrial Disorders
University Paris 6, Paris	Master	1990	Plant Biology

A. Personal Statement

My laboratory is affiliated to the Centre National Recherche Scientifique (CNRS)/Université de Paris/Institut Pasteur and is part of the Neuroscience Department of Institut Pasteur. It gathers psychiatrists, neuroscientists and geneticists aiming at better understanding the causes of autism spectrum disorders.

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. These projects are focused on deep-phenotyping of individuals with autism or neurodevelopmental disorders (NDD) and controls including brain imaging and a battery of cognitive tests. Our group is currently developing new 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 the [INCEPTION](#) project, 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); Laureate of the 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

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)

E. Most significant publications

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** doi: 10.1038/s41588-022-01072-5. *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

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, Régnauld 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, Cloëz-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, Lemièrre 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.

2022

177. 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, Hurler ME, Geschwind DH, Børghlum AD, Robinson EB, Grove J, Martin HC, **Bourgeron T***, Baron-Cohen S*. Genetic correlates of phenotypic heterogeneity in autism. *Nature Genetics* doi: 10.1038/s41588-022-01072-5. *senior authors
176. Garcés P, Baumeister S, Mason L, Chatham CH, Holiga S, Dukart J, Jones E, Banaschewski T, Baron-Cohen S, Bölte S, Buitelaar JK, Durston S, Oranje B, Persico AM, Beckmann CF, **Bourgeron T**, Dell'Acqua F, Ecker C, Moessnang C, Charman T, Tillmann J, Murphy DGM, Johnson M, Loth E, Brandeis D, Hipp JF; EU-AIMS LEAP group authorship. Resting state EEG power spectrum and functional connectivity in autism: a cross-sectional analysis. *Mol Autism*. 2022 May 18;13(1):22.
175. Traut N, Heuer K, Lemaître G, Beggato A, Germanaud D, Elmaleh M, Bethegnies A, Bonnasse-Gahot L, Cai W, Chambon S, Cliquet F, Ghriess A, Guigui N, de Pierrefeu A, Wang M, Zantedeschi V, Boucaud A, van den Bossche J, Kegl B, Delorme R, Bourgeron T, Toro R, Varoquaux G. Insights from an autism imaging biomarker challenge: Promises and threats to biomarker discovery. *Neuroimage*. 2022 Apr 10;255:119171. doi: 10.1016/j.neuroimage.2022.119171.
174. Pretzsch CM, Schäfer T, Lombardo MV, Warrier V, Mann C, Bletsch A, Chatham CH, Floris DL, Tillmann J, Yousaf A, Jones E, Charman T, Ambrosino S, Bourgeron T, Dumas G, Loth E, Oakley B, Buitelaar JK, Cliquet F, Leblond CS, Baron-Cohen S, Beckmann CF, Banaschewski T, Durston S, Freitag CM; EU-AIMS LEAP Group, Murphy DGM, Ecker C. Neurobiological Correlates of Change in Adaptive Behavior in Autism. *Am J Psychiatry*. 2022 Mar 25;appiajp.21070711. doi: 10.1176/appi.ajp.21070711. Online ahead of print.
173. Cooke J, Molloy CJ, Cáceres ASJ, Dinneen T, **Bourgeron T**, Murphy D, Gallagher L, Loth E. The Synaptic Gene Study: Design and Methodology to Identify Neurocognitive Markers in Phelan-McDermid Syndrome and NRXN1 Deletions. *Front Neurosci*. 2022 Feb 18;16:806990. doi: 10.3389/fnins.2022.806990. eCollection 2022.
172. Pretzsch CM, Schäfer T, Lombardo MV, Warrier V, Mann C, Bletsch A, Chatham CH, Floris DL, Tillmann J, Yousaf A, Jones E, Charman T, Ambrosino S, **Bourgeron T**, Dumas G, Loth E, Oakley B, Buitelaar JK, Cliquet F, Leblond CS, Baron-Cohen S, Beckmann CF, Banaschewski T, Durston S, Freitag CM; EU-AIMS LEAP Group, Murphy DGM, Ecker C. Neurobiological Correlates of Change in Adaptive Behavior in Autism. *Am J Psychiatry*. 2022 Mar 25;appiajp.21070711. doi: 10.1176/appi.ajp.21070711.
171. Cooke J, Molloy CJ, Cáceres ASJ, Dinneen T, Bourgeron T, Murphy D, Gallagher L, Loth E. The Synaptic Gene Study: Design and Methodology to Identify Neurocognitive Markers in Phelan-McDermid Syndrome and NRXN1 Deletions. *Front Neurosci*. 2022 Feb 18; 16:806990.
170. Phelan K, Boccuto L, Powell CM, Boeckers TM, van Ravenswaaij-Arts C, Rogers C, Sala C, Verpelli C, Thurm A, Bennett, Jr. WE, Winrow CJ, Garrison S, Toro R, **Bourgeron T**. Phelan-McDermid syndrome: a classification system after 30 years of experience. *Orphanet Journal of Rare Diseases*. Orphanet J Rare Dis. 2022 Jan 29;17(1):27.

2021

169. Lutz AK, Pérez Arévalo A, Ioannidis V, Stirmlinger N, Demestre M, Delorme R, **Bourgeron T**, Boeckers TM. SHANK2 Mutations Result in Dysregulation of the ERK1/2 Pathway in Human Induced Pluripotent Stem Cells-Derived Neurons and Shank2(-/-) Mice. *Front Mol Neurosci*. 2021 26;14:773571
168. Loth E, Ahmad J, Chatham C, López B, Carter B, Crawley D, Oakley B, Hayward H, Cooke J, San José Cáceres A, Bzdok D, Jones E, Charman T, 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.
167. Loureiro LO, Howe JL, Reuter MS, Iaboni A, Calli K, Roshandel D, Pritisanac I, Moses A, Forman-Kay JD, Trost B, Zarrei M, Rennie O, Lau LYS, Marshall CR, Srivastava S, Godlewski B, Buttermore ED, Sahin M, Hartley D, Frazier T, Vorstman J, Georgiades S, Lewis SME, Szatmari P, Bradley CAL, Tabet AC, Willems M, Lumbroso S, Piton A, Lespinasse J, Delorme R, **Bourgeron T**, Anagnostou E, Scherer SW. A recurrent SHANK3 frameshift variant in Autism Spectrum Disorder. *NPJ Genom Med*. 2021 Nov 4;6(1):91.
166. de Chaumont F, Lemièrre N, Coqueran S, **Bourgeron T***, Ey E*. LMT USV Toolbox, a Novel Methodological Approach to Place Mouse Ultrasonic Vocalizations in Their Behavioral Contexts-A Study in Female and Male C57BL/6J Mice and in Shank3 Mutant Females. *Front Behav Neurosci*. 2021 Oct 13;15:735920. * Senior authors
165. Ecker C, Pretzsch CM, Bletsch A, Mann C, Schaefer T, Ambrosino S, Tillmann J, Yousaf A, Chiocchetti A, Lombardo MV, Warrier V, Bast N, Moessnang C, Baumeister S, Dell'Acqua F, Floris DL, Zabihi M, Marquand A, Cliquet F, Leblond C, Moreau C, Puts N, Banaschewski T, Jones E, Mason L, Bölte S, Meyer-Lindenberg A, Persico AM, Durston S, Baron-Cohen S, Spooren W, Loth E, Freitag CM, Charman T, Dumas G, **Bourgeron T**, Beckmann CF, Buitelaar JK, Murphy DGM. Interindividual Differences in Cortical Thickness and Their Genomic Underpinnings in Autism Spectrum Disorder. *Am J Psychiatry*. 2021 doi: 10.1176/appi.ajp.2021.20050630.
164. Lefebvre A, Cohen A, Maruani A, Amsellem F, Beggato A, Amestoy A, Moal ML, Umbricht D, Chatham C, Murtagh L, Bouvard M, Leboyer M, **Bourgeron T**, Delorme R. Discriminant value of repetitive behaviors in families with autism spectrum disorder and obsessional compulsive disorder probands. *Autism Res*. 2021 Jul 19. doi: 10.1002/aur.2570.
163. Kikusui T, Sonobe M, Yoshida Y, Nagasawa M, Ey E, de Chaumont F, **Bourgeron T**, Nomoto K, Mogi K. Testosterone Increases the Emission of Ultrasonic Vocalizations With Different Acoustic Characteristics in Mice. *Front Psychol*. 2021 Jun 25;12:680176.
162. Bertelsen N, Landi I, Bethlehem RAI, Seidlitz J, Busuoli EM, Mandelli V, Satta E, Trakoshis S, Auyeung B, Kundu P, Loth E, Dumas G, Baumeister S, Beckmann CF, Bölte S, **Bourgeron T**, Charman T, Durston S, Ecker C, Holt RJ, Johnson MH, Jones E, Mason L, Meyer-Lindenberg A, Moessnang C, Oldehinkel M, Persico AM, Tillmann J, Williams SCR, Spooren W, Murphy DGM, Buitelaar JK; EU-AIMS LEAP group, Baron-Cohen S, Lai MC, Lombardo MV. Imbalanced social-communicative and restricted repetitive behavior subtypes of autism spectrum disorder exhibit different neural circuitry. *Commun Biol*. 2021 May 14;4(1):574.
161. Leblond CS, Le TL, Malesys S, Cliquet F, Tabet AC, Delorme R, Rolland T, **Bourgeron T**. Operative list of genes associated with autism and neurodevelopmental disorders based on database review. *Mol Cell Neurosci*. 2021 Jun;113:103623.
160. 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* 2021 Mar;31(3):484-496.

159. 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* 2021 Jan 7. doi: 10.1038/s41380-020-00985-z
158. Pagan C, Benabou M, Leblond CSL, Cliquet F, Mathieu A, Lemièrre N, Goubran-botros H, Delorme R, Leboyer M, Callebort J, **Bourgeron T***. Launay JM* Decreased phenol sulfotransferase activities associated with hyperserotonemia in Autism Spectrum Disorders. *Trans Psychiatry* 2021 11:23. *senior authors.
157. 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, Motttron 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.

2020

156. Dumas G, Goubran-Botros H, Matondo M, Pagan C, Boulègue C, Chaze T, Chamot-Rooke J, Maronde E, **Bourgeron T**. Mass-spectrometry analysis of the human pineal proteome during night and day and in autism. *J Pineal Res*. 2020 Dec 23:e12713.
155. 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.
154. **Bourgeron T**. The gnomAD project and the importance of having more than only one reference human genome! *C R Biol*. 2020 Oct 9;343(2):123-125.
153. Moreau CA, Urchs SGW, Kuldeep K, Orban P, Schramm C, Dumas G, Labbe A, Huguet G, Douard E, Quirion PO, Lin A, Kushan L, Grot S, Luck D, Mendrek A, Potvin S, Stip E, Bourgeron T, Evans AC, Bearden CE, Bellec P, Jacquemont S. Mutations associated with neuropsychiatric conditions delineate functional brain connectivity dimensions contributing to autism and schizophrenia. *Nat Commun*. 2020 Oct 19;11(1):5272.
152. 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
151. Gialluisi A, Andlauer TFM, Mirza-Schreiber N, Moll K, Becker J, Hoffmann P, Ludwig KU, Czamara D, Pourcain BS, Honbolygó F, Tóth D, Csépe V, Huguet G, Chaix Y, Iannuzzi S, Demonet JF, Morris AP, Hulslander J, Willcutt EG, DeFries JC, Olson RK, Smith SD, Pennington BF, Vaessen A, Maurer U, Lyytinen H, Peyrard-Janvid M, Leppänen PHT, Brandeis D, Bonte M, Stein JF, Talcott JB, Fauchereau F, Wilcke A, Kirsten H, Müller B, Francks C, **Bourgeron T**, Monaco AP, Ramus F, Landerl K, Kere J, Scerri TS, Paracchini S, Fisher SE, Schumacher J, Nöthen MM, Müller-Myhsok B, Schulte-Körne G Genome-wide association study reveals new insights into the heritability and genetic correlates of developmental dyslexia. *Mol Psychiatry*. 2020 Oct 14. doi: 10.1038/s41380-020-00898
150. Baillin F, Lefebvre A, Pedoux A, Beauxis Y, Engemann DA, Maruani A, Amsellem F, **Bourgeon T**, Delorme R, Dumas G. Interactive Psychometrics for Autism with the Human Dynamic Clamp: Interpersonal Synchrony from Sensory-motor to Socio-cognitive Domains. *Frontiers in Psychiatry*, section Social Cognition. 11:510366.
149. Vitrac A, Pons S, Balkota M, Lemièrre N, Raïs C, Bourgeois JP, Maskos U, **Bourgeron T**, Cloëz-Tayarani I. A chimeric mouse model to study human iPSC-derived neurons: the case of a truncating SHANK3 mutation. *Sci Rep*. 2020 Aug 7;10(1):13315.
148. 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.
147. 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.
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
144. Biton A, Traut N, Poline JB, Aribisala BS, Bastin ME, Bülow R, Cox SR, Deary IJ, Fukunaga M, Grabe HJ, Hagenaars S, Hashimoto R, Kikuchi M, Muñoz Maniega S, Nauck M, Royle NA, Teumer A, Valdés Hernández M, Völker U, Wardlaw JM, Wittfeld K, Yamamori H; Alzheimer's Disease Neuroimaging Initiative, **Bourgeron T***, Toro R*. Polygenic Architecture of Human Neuroanatomical Diversity. *Cereb Cortex*. (2020) 30(4):2307-2320.. * senior authors.

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. * senior authors.
141. de Lombares C, Heude E, Alfama G, Fontaine A, Hassouna R, Vernochet C, de Chaumont F, Olivo-Marin C, Ey E, Parnaud 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. * 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.
 137. Maruani A, Dumas G, Beggiato A, Traut N, Peyre H, Cohen-Freoua A, Amsellem F, Elmaleh M, Germanaud D, Launay JM, **Bourgeron T**, Toro R, Delorme R. Morning Plasma Melatonin Differences in Autism: Beyond the Impact of Pineal Gland Volume. *Front Psychiatry*. 2019 Feb 6;10:11. doi: 10.3389/fpsyt.2019.00011. eCollection 2019.
 136. Gialluisi A, Andlauer TFM, Mirza-Schreiber N, Moll K, Becker J, Hoffmann P, Ludwig KU, Czamara D, St Pourcain B, Brandler W, Honbolygó F, Tóth D, Csépe V, Huguet G, Morris AP, Hulslander J, Willcutt EG, DeFries JC, Olson RK, Smith SD, Pennington BF, Vaessen A, Maurer U, Lyytinen H, Peyrard-Janvid M, Leppänen PHT, Brandeis D, Bonte M, Stein JF, Talcott JB, Fauchereau F, Wilcke A, Francks C, **Bourgeron T**, Monaco AP, Ramus F, Landerl K, Kere J, Scerri TS, Paracchini S, Fisher SE, Schumacher J, Nöthen MM, Müller-Myhsok B, Schulte-Körne G. Genome-wide association scan identifies new variants associated with a cognitive predictor of dyslexia. *Transl Psychiatry*. 2019 Feb 11;9(1):77. doi: 10.1038/s41398-019-0402-0.
 135. Leblond CS, Cliquet F, Carton C, Huguet G, Mathieu A, Kergrohen T, Buratti J, Lemièrre N, Cuisset L, Bienvenu T, Boland A, Deleuze JF, Stora T, Biskupstoe R, Halling J, Andorsdóttir G, Billstedt E, Gillberg C, **Bourgeron T**. Both rare and common genetic variants contribute to autism in the Faroe Islands. *NPJ Genom Med*. 2019 21;4:1.
 134. Gouder L, Vitrac A, Goubiran-Botros H, Danckaert A, Tinevez JY, André-Leroux G, Atanasova E, Lemièrre N, Leblond CS, Poulet A, Benchoua A, Delorme R, **Bourgeron T*** and Cloëz-Tayarani I* Altered spinogenesis in iPSC-derived cortical neurons from patients with autism carrying de novo SHANK3 mutations. * senior authors. *Scientific Reports* 2019 14;9(1):94.
 133. Bouvet L, Amsellem F, Maruani A, Tonus-Vic Dupont A, Mathieu A, **Bourgeron T**, Delorme R, Mottron L. Synesthesia & autistic features in a large 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.
- 2018**
132. Lefebvre A, Delorme R, Delanoë C, Amsellem F, Beggiato A, Germanaud D, **Bourgeron T**, Toro R, Dumas G. Alpha Waves as a Neuromarker of Autism Spectrum Disorder: The Challenge of Reproducibility and Heterogeneity. *Front Neurosci*. 2018 Oct 1;12:662.
 131. Ey E., Torquet N, de Chaumont F, Lévi-Strauss J, Ferhat AT, Le Sourd AM, Boeckers TM and **Bourgeron T** Shank2 Mutant Mice Display Hyperactivity Insensitive to Methylphenidate and Reduced Flexibility in Social Motivation, but Normal Social Recognition *Front. Mol. Neurosci.*, 2018 Oct 4;11:365.
 130. Lahbib S, Leblond CS, Hamza M, Regnault B, Lemée L, Mathieu A, Jaouadi H, Mkaouer R, Youssef-Turki IB, Belhadj A, Kraoua I, **Bourgeron T**, Abdelhak S. Homozygous 2p11.2 deletion supports the implication of ELMOD3 in hearing loss and reveals the potential association of CAPG with ASD/ID etiology. *J Appl Genet*. 2018 Oct 4. doi: 10.1007/s13353-018-0472-3.
 129. Septier M, Peyre H, Amsellem F, Beggiato A, Maruani A, Poumeyreau M, Amestoy A, Scheid I, Gaman A, Bolognani F, Honey G, Bouquet C, Ly-Le Moal M, Bouvard M, Leboyer M, **Bourgeron T**, Delorme R. Increased risk of ADHD in families with ASD. *Eur Child Adolesc Psychiatry*. 2018 Sep 28. doi: 10.1007/s00787-018-1206-0.
 128. Aubart M, Gazal S, Arnaud P, Benarroch L, Gross MS, Buratti J, Boland A, Meyer V, Zouali H, Hanna N, Milleron O, Stheneur C, **Bourgeron T**, Desguerre I, Jacob MP, Gouya L, Génin E, Deleuze JF, Jondeau G, Boileau C. Association of modifiers and other genetic factors explain Marfan syndrome clinical variability. *Eur J Hum Genet*. 2018 Aug 7.
 127. Bonnet A, Levy-Leduc C, Gassiat E, Toro R, **Bourgeron T**. Improving heritability estimation by a variable selection approach in sparse high dimensional linear mixed models. *Journal of the Royal Statistical Society* 2018, 67 (4), pp.813-839.
 126. 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. * senior authors
 125. Warrier V, Roberto Toro, Bhismadev Chakrabarti, The iPSYCH-BROAD Autism Group, Jakob Grove, Anders Børghlum, 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.* senior authors
 124. Traut N, Beggiato A, **Bourgeron T**, Delorme R, Rondi-Reig L, Paradis AL, Toro R. Cerebellar volume in autism: Meta-analysis and analysis of the ABIDE cohort *Biological Psychiatry* 2018 1;83(7):579-588..
 123. Warrier V, Grasby K, Uzefovsky F, Toro R, Smith P, Chakrabarti B, Khadake J, Litterman N, Hottenga J-J, Lubke G, Boomsma DI, Martin NG, Hatemi PK, Medland SE , Hinds DA , **Bourgeron T***, Baron-Cohen S*. Genome-wide meta-analysis of cognitive empathy: heritability, and correlates with sex, neuropsychiatric conditions and brain anatomy. *Molecular Psychiatry* 2017 2018 Jun;23(6):1402-1409. * senior authors
- 2017**
122. 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.
 121. 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 EJJ, 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.
120. Loth E, Charman T, Mason L, Tillmann J, Jones EJH, Wooldridge C, Ahmad J, Auyeung B, Brogna C, Ambrosino S, Banaschewski T, Baron-Cohen S, Baumeister S, Beckmann C, Brammer M, Brandeis D, Bölte S, **Bourgeron T**, Bours C, de Bruijn Y, Chakrabarti B, Crawley D, Cornelissen I, Acqua FD, Dumas G, Durston S, Ecker C, Faulkner J, Frouin V, Garces P, Goyard D, Hayward H, Ham LM, Hipp J, Holt RJ, Johnson MH, Isaksson J, Kundu P, Lai MC, D'ardhuy XL, Lombardo MV, Lythgoe DJ, Mandl R, Meyer-Lindenberg A, Moessnang C, Mueller N, O'Dwyer L, Oldehinkel M, Oranje B, Pandina G, Persico AM, Ruigrok ANV, Ruggeri B, 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): design and methodologies to identify and validate stratification biomarkers for autism spectrum disorders. *Mol Autism*. 2017 Jun 23;8:24. doi: 10.1186/s13229-017-0146-8.
 119. Tabet AC, Rolland T, Ducloy M, Lévy J, Buratti J, Mathieu A, Haye D, Perrin L, Dupont C, Passemard S, Capri Y, Verloes A, Drunat S, Keren B, Mignot C, Marey I, Jacqueline A, Whalen S, Pipiras E, Benzacken B, Chantot-Bastarud S, Afenjar A, Héron D, Le Caignec C, Beneteau C, Pichon O, Isidor B, David A, El Khattabi L, Kemeny S, Gouas L, Vago P, Mosca-Boidron AL, Faivre L, Missirian C, Philip N, Sanlaville D, Edery P, Satre V, Coutton C, Devillard F, Dieterich K, Vuillaume ML, Rooryck C, Lacombe D, Pinson L, Gatinois V, Puechberty J, Chiesa J, Lespinasse J, Dubourg C, Quelin C, Fradin M, Journal H, Toutain A, Martin D, Benmansour A, Leblond CS, Toro R, Amsellem F, Delorme R, **Bourgeron T**. A framework to identify contributing genes in patients with Phelan-McDermid syndrome. *NPJ Genom Med*. 2017 2:32.
 118. Ferhat AT, Halbedl S, Schmeisser MJ, Kas MJ, **Bourgeron T**, Ey E. Behavioural Phenotypes and Neural Circuit Dysfunctions in Mouse Models of Autism Spectrum Disorder. *Adv Anat Embryol Cell Biol*. 2017;224:85-101. doi: 10.1007/978-3-319-52498-6_5.
 117. Kleijer KTE, Huguet G, Tastet J, **Bourgeron T**, Burbach JPH. Anatomy and Cell Biology of Autism Spectrum Disorder: Lessons from Human Genetics. *Adv Anat Embryol Cell Biol*. 2017; 224:1-25. doi: 10.1007/978-3-319-52498-6_1.
 116. Autism Spectrum Disorders Working Group of The Psychiatric Genomics Consortium. Meta-analysis of GWAS of over 16,000 individuals with autism spectrum disorder highlights a novel locus at 10q24.32 and a significant overlap with schizophrenia. *Mol Autism*. 2017 May 22; 8:21. doi: 10.1186/s13229-017-0137-9. eCollection 2017.
 115. Pagan C, Goubran-Botros H, Delorme R, Benabou M, Lemièrre N, Murray K, Amsellem F, Callebert J, Chaste P, Jamain S, Fauchereau F, Huguet G, Maronde E, Leboyer M, **Bourgeron T**. Disruption of melatonin synthesis is associated with impaired 14-3-3 and miR-451 levels in patients with autism spectrum disorders. *Scientific Reports* 2017 18;7(1):2096.
 114. Gillberg C, Fernell E, Kočovská E, Minnis H, **Bourgeron T**, Thompson L, Allely CS. The role of cholesterol metabolism and various steroid abnormalities in autism spectrum disorders: A hypothesis paper. *Autism Res*. 2017 Apr 12. doi: 10.1002/aur.1777. [Epub ahead of print]
 113. Kishimoto K, Nomura J, Ellegood J, Fukumoto K, Lerch JP, Moreno-De-Luca D, **Bourgeron T**, Tamada K, Takumi T. Behavioral and neuroanatomical analyses in a genetic mouse model of 2q13 duplication. *Genes Cells*. 2017 Mar 29. doi: 10.1111/gtc.12487. [Epub ahead of print]
 112. Pfaender S., Sauer AK, Hagmeyer S, Mangus K, Linta L, Liebau S, Bockmann J, Huguet G, **Bourgeron T**, Boeckers TM, Grubbrucker AM. Zinc deficiency and low enterocyte zinc transporter expression in human patients with autism related mutations in SHANK3. *Scientific Reports* 2017 27;7:45190.
 111. 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, Régault 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, Cloëz-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.
- 2016**
110. Beggiato A, Peyre H, Maruani A, Scheid I, Rastam M, Amsellem F, Gillberg CI, Leboyer M, **Bourgeron T**, Gillberg C, Delorme R. Gender differences in autism spectrum disorders: Divergence among specific core symptoms. *Autism Res*. 2016 10(4):680-689.
 109. Torquet N, de Chaumont F, Faure P, **Bourgeron T**, Ey E. mouseTube - a database to collaboratively unravel mouse ultrasonic communication. *F1000Res*. 2016 Sep 16;5:2332.
 108. **Bourgeron T**. Current knowledge on the genetics of autism and propositions for future research. *C R Biol*. 2016 Jul-Aug;339(7-8):300-7.
 107. Torrico B, Chiochetti AG, Bacchelli E, Trabetti E, Hervás A, Franke B, Buitelaar JK, Rommelse N, Yousaf A, Duketis E, Freitag CM, Caballero-Andaluz R, Martinez-Mir A, Scholl FG, Ribasés M; ITAN, Battaglia A, Malerba G, Delorme R, Benabou M, Maestrini E, **Bourgeron T**, Cormand B, Toma C. Lack of replication of previous autism spectrum disorder GWAS hits in European populations. *Autism Res*. 2016 Jul 15. doi: 10.1002/aur.1662.
 106. Darville H, Poulet A, Rodet-Amsellem F, Chatrousse L, Pernelle L, Boissart C, Héron D, Nava C, Perrier A, Jarrige M, Cogé F, Millan MJ, **Bourgeron T**, Peschanski M, Delorme R, Benchoua A Human Pluripotent Stem Cell-derived Cortical Neurons for High Throughput Medication Screening in Autism: A Proof of Concept Study in SHANK3 Haploinsufficiency Syndrome. *EBioMedicine* 2016 9:293-305.
 105. Ferhat AT, Torquet N, Le Sourd AM, de Chaumont F, Olivo-Marin JC, Faure P, **Bourgeron T**, Ey E. Recording mouse ultrasonic vocalizations in the laboratory to evaluate social communication. *JoVE Jun 5;(112)*. doi: 10.3791/53871.
 104. Loth E, Spooren W, Ham LM, Isaac MB, Auriche-Benichou C, Banaschewski T, Baron-Cohen S, Broich K, Bölte S, **Bourgeron T**, Charman T, Collier D, de Andres-Trelles F, Durston S, Ecker C, Elferink A, Haberkamp M, Hemmings R, Johnson MH, Jones EJ, Khwaja OS, Lenton S, Mason L, Mantua V, Meyer-Lindenberg A, Lombardo MV, O'Dwyer L, Okamoto K, Pandina GJ, Pani L, Persico AM, Simonoff E, Tauscher-Wisniewski S, Llinares-Garcia J, Vamvakas S, Williams S, Buitelaar JK, Murphy DG. *Nature Reviews Drug Discovery*. 2015 Dec 31;15(1):70-3.
 103. Mosca-Boidron AL, Gueneau L, Huguet G, Goldenberg A, Henry C, Gigot N, Palesi-Pocachard E, Falace A, Duplomb L, Thevenon J, Duffourd Y, St-Onge J, Chambon P, Rivière JB, Thauvin-Robinet C, Callier P, Marle N, Payet M, Ragon C, Botros HG, Buratti J, Calderari S, Dumas G, Delorme R, Lagarde N, Pinoit JM, Rosier A, Masurel-Paulet A, Cardoso C, Mugneret F, Saugier-Verber P, Campion D, Faivre L and **Bourgeron T**. A de novo microdeletion of SEMA5A in a boy with autism spectrum disorder and intellectual disability. *Eur J Hum Genet*. 2016 24(6):838-43.
- 2015**

102. Maruani A, Huguet G, Beggiato A, ElMaleh M, Toro R, Leblond C, Mathieu A, Amsellem F, Lemièrre N, Verloes A, Leboyer M, Gillberg C, **Bourgeron T**, Delorme R. 11q24.2-25 micro-rearrangements in Autism Spectrum Disorders: Relation to Brain Structures. *Am J Med Genet A*. 2015 Sep 3. doi: 10.1002/ajmg.a.37345. [Epub ahead of print]
 101. Gouder L, Tinevez JY, Goubran-Botros H, Benchoua A, **Bourgeron T**, Clôez-Tayarani I. Three-dimensional quantification of dendritic spines from pyramidal neurons derived from human inducible pluripotent stem cells. *JoVE* 2015 Oct 10;(104). doi: 10.3791/53197.
 100. **Bourgeron T**. From the genetic architecture to synaptic plasticity in autism spectrum disorders. *Nature Reviews Neuroscience* 16, 551–563.
 99. **Bourgeron T**. The genetics and neurobiology of ESSENCE: The third Birgit Olsson lecture. *Nord J Psychiatry*. 2015 May 14:1-9.
 98. Ferhat AT, Le Sourd AM, de Chaumont F, Olivo-Marin JC, **Bourgeron T**, Ey E. Social communication in mice - are there optimal cage conditions? *PLoS One*. 2015 Mar 25;10(3):e0121802.
 97. Lefebvre A, Beggiato A, **Bourgeron T**, Toro R. Neuroanatomical Diversity of Corpus Callosum and Brain Volume in Autism: Meta-analysis, Analysis of the Autism Brain Imaging Data Exchange Project, and Simulation. *Biological Psychiatry*. (2015) doi: 10.1016/j.biopsych.2015.02.010.
 96. Pagan C, Delorme R, Callebort J, Goubran Botros H, Amsellem F, Drouot X, Boudebessé C, Le Dudal K, Ngo-Nguyen N, Laouamri H, Gillberg C, Leboyer M, **Bourgeron T**, Launay JM. The serotonin-N-acetylserotonin-melatonin pathway as a biomarker for autism spectrum disorders. *Translational Psychiatry* (2015) 4:e479.
 95. Toro R., Poline JB, Huguet G., Loth E, Frouin V, Banaschewski T., Barker GJ, Bokde A, Buchel C., Carvalho FM, Conrod P, Fauth-Bühler M., Flor H, Gallinat J, Garavan H, Gowland P., Heinz A, Ittermann B, Lawrence C, Lemaitre H, Mann K, Nees F, Paus T, Pausova Z, Rietschel M, Robbins T, Smolka MN, Stroble A, Schumann G, and **Bourgeron T** Genomic architecture of human neuroanatomical diversity *Molecular Psychiatry* (2015) 20(8):1011-6
- 2014**
94. Kočovská E, Andorsdóttir G, Weihe P, Halling J, Fernell E, Stóra T, Biskupstø R, Gillberg IC, Shea R, Billstedt E, **Bourgeron T**, Minnis H, Gillberg C. Vitamin D in the General Population of Young Adults with Autism in the Faroe Islands. *J Autism Dev Disord*. 2014 Jun 14.
 93. Leblond CS, Nava C, Polge A, Gauthier J, Huguet G, Lumbroso S, Giuliano F, Stordeur C, Depienne C, Mouzat K, Pinto D, Howe J, Lemièrre 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, Jacqueline 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.
 92. Karayannis T, E. Au, J. Patel, I. Kruglikov, S. Mark, R. Delorme, D. Héron, J. Glessner, S. Restituito, A. Gordon, L. Rodriguez-Murillo, N.C. Roy, J. Gogos, B. Rudy, M.E. Rice, M. Karayiorgou, H. Hakonarson, B. Keren, G. Huguet, **Bourgeron T**, C. Hoeffler, R.W. Tsien, E. Peles, G. Fishell. Cntnap4 Differentially Contributes to GABAergic and Dopaminergic Synaptic Transmission *Nature* 2014 May 25. doi: 10.1038
 91. Pinel P, Lalanne C, **Bourgeron T**, Fauchereau F, Poupon C, Artiges E, Le Bihan D, Dehaene-Lambertz G, Dehaene S. Genetic and Environmental Influences on the Visual Word Form and Fusiform Face Areas. *Cereb Cortex*. 2014 May 13.
 90. Pinto D, Delaby E, Merico D, Barbosa M, Merikangas A, Klei L, Thiruvahindrapuram B, Xu X, Ziman R, Wang Z, Vorstman JA, Thompson A, Regan R, Pilorge M, Pellecchia G, Pagnamenta AT, Oliveira B, Marshall CR, Magalhaes TR, Lowe JK, Howe JL, Griswold AJ, Gilbert J, Duketis E, Dombroski BA, De Jonge MV, Cuccaro M, Crawford EL, Correia CT, Conroy J, Conceição IC, Chiochetti AG, Casey JP, Cai G, Cabrol C, Bolshakova N, Bacchelli E, Anney R, Gallinger S, Cotterchio M, Casey G, Zwaigenbaum L, Wittemeyer K, Wing K, Wallace S, van Engeland H, Tryfon A, Thomson S, Soorya L, Rogé B, Roberts W, Poustka F, Moug S, Minshew N, McInnes LA, McGrew SG, Lord C, Leboyer M, Le Couteur AS, Kolevzon A, Jiménez González P, Jacob S, Holt R, Guter S, Green J, Green A, Gillberg C, Fernandez BA, Duque F, Delorme R, Dawson G, Chaste P, Café C, Brennan S, **Bourgeron T**, Bolton PF, Bölte S, Bernier R, Baird G, Bailey AJ, Anagnostou E, Almeida J, Wijsman EM, Veland VJ, Vicente AM, Schellenberg GD, Pericak-Vance M, Paterson AD, Parr JR, Oliveira G, Nurnberger JI, Monaco AP, Maestrini E, Klauck SM, Hakonarson H, Haines JL, Geschwind DH, Freitag CM, Folstein SE, Ennis S, Coon H, Battaglia A, Szatmari P, Sutcliffe JS, Hallmayer J, Gill M, Cook EH, Buxbaum JD, Devlin B, Gallagher L, Betancur C, Scherer SW. Convergence of genes and cellular pathways dysregulated in autism spectrum disorders. *Am J Hum Genet*. 2014 May 1;94(5):677-94.
 89. Huguet G., Nava C., Lemièrre N, Ey E, Brice A., Leboyer M., Szepetowski P, Gillberg C., Depienne C., Delorme R., **Bourgeron T**. Heterogeneous pattern of selective pressure for PRRT2 in human populations, but no association with autism spectrum disorders. *PLoS One* 9 (3); e886000
 88. Fernell E, Wilson P, Hadjikhani N, **Bourgeron T**, Neville B, Taylor D, Minnis H, Gillberg C. Screening, Intervention and Outcome in Autism and Other Developmental Disorders: The Role of Randomized Controlled Trials. *J Autism Dev Disord*. 2014 Feb 20.
 87. Waltes R, Duketis E, Knapp M, Anney RJ, Huguet G, Schlitt S, Jarczok TA, Sachse M, Kämpfer LM, Kleinböck T, Poustka F, Bölte S, Schmötzer G, Voran A, Huy E, Meyer J, **Bourgeron T**, Klauck SM, Freitag CM, Chiochetti AG. Common variants in genes of the postsynaptic FMRP signalling pathway are risk factors for autism spectrum disorders. *Hum Genet*. 2014 Jan 19.
 86. Kleijer KT, Schmeisser MJ, Krueger DD, Boeckers TM, Scheiffele P, **Bourgeron T**, Brose N, Burbach JP. Neurobiology of autism gene products: towards pathogenesis and drug targets. *Psychopharmacology (Berl)*. 2014 Mar;231(6):1037-62.

2013

85. Scheid I, Maruani A, Huguët G, Leblond CS, Nygren G, Anckarsäter H, Beggiato A, Rastam M, Amsellem F, Gillberg IC, Elmaleh M, Leboyer M, Gillberg C, Betancur C, Coleman M, Hama H, Cook EH, **Bourgeron T**, Delorme R. Heterozygous FA2H mutations in autism spectrum disorders. *BMC Med Genet*. 2013 Dec 3;14(1):124.
84. Guilmatre A, Huguët G, Delorme R, **Bourgeron T**. The emerging role of SHANK genes in neuropsychiatric disorders. *Dev Neurobiol*. 2013 74(2): 113-22.
83. Ey E, Torquet N, Le Sourd AM, Leblond CS, Boeckers TM, Faure P and **Bourgeron T**. (2013) The Autism ProSAP1/Shank2 Mouse Model Displays Quantitative and Structural Abnormalities in Ultrasonic Vocalisations. *Behav Brain Res*. doi:pii: S0166-4328
82. Boissart C, Poulet A, Georges P, Darville H, Julita E, Delorme R, **Bourgeron T**, Peschanski M, Benchoua A. (2013) Differentiation from human pluripotent stem cells of cortical neurons of the superficial layers amenable to psychiatric disease modeling and high-throughput drug screening. *Transl Psychiatry*. 3:e294.
81. Huguët G, Ey E and **Bourgeron T**. (2013) The genetic landscapes of autism spectrum disorders. *Annu Rev Genomics Hum Genet*. 14:191-213.
80. Delorme R, Ey E, Toro R, Leboyer M, Gillberg C, and **Bourgeron T**. (2013) Progress towards treatments for synaptic defects in autism. *Nature Medicine* 19(6):685-94
79. Mercati O, Danckaert A, André-Leroux G, Bellinzoni M, Gouder L, Watanabe K, Shimoda Y, Grailhe R, de Chaumont F, **Bourgeron T** and Cloëz-Tayarani I (2013) Contactin 4, -5 and -6 differentially regulate neuritogenesis while they display identical PTPRG binding sites. *Biology Open* 2:324-34
78. Wang L, Li J, Ruan Y, Lu T, Liu C, Jia M, Yue W, Liu J, **Bourgeron T**, Zhang D. (2013) Sequencing ASMT Identifies Rare Mutations in Chinese Han Patients with Autism. *PLoS One*. 8(1):e53727.

2012

77. El-Kordi A, Winkler D, Hammerschmidt K, Kästner A, Krueger D, Ronnenberg A, Ritter C, Jatho J, Radyushkin K, **Bourgeron T**, Fischer J, Brose N, Ehrenreich H. Development of an autism severity score for mice using Nlgn4 null mutants as a construct-valid model of heritable monogenic autism. *Behav Brain Res*. 2012 Nov 23.
76. Nava C, Lamari F, Héron D, Mignot C, Rastetter A, Keren B, Cohen D, Faudet A, Bouteiller D, Gilleron M, Jacqueline A, Whalen S, Afenjar A, Périsse D, Laurent C, Dupuits C, Gautier C, Gérard M, Huguët G, Caillet S, Leheup B, Leboyer M, Gillberg C, Delorme R, **Bourgeron T**, Brice A, Depienne C. Analysis of the chromosome X exome in patients with autism spectrum disorders identified novel candidate genes, including TMLHE. *Transl Psychiatry*. 2012 Oct 23;2:e179.
75. Ey E, Yang M, Katz AM, Woldeyohannes L, Silverman JL, Leblond CS, Faure P, Torquet N, Le Sourd AM, **Bourgeron T**, Crawley JN. Absence of deficits in social behaviors and ultrasonic vocalizations in later generations of mice lacking neuroligin4. *Genes Brain Behav*. 2012 Sep 18.
74. Anney R, Klei L, Pinto D, Almeida J, Bacchelli E, Baird G, Bolshakova N, Bølte S, Bolton PF, **Bourgeron T** et al. Individual common variants exert weak effects on the risk for autism spectrum disorders. *Hum Mol Genet*. 2012 Aug 8. [Epub ahead of print]
73. Le Floch E, Guillemot V, Frouin V, Pinel P, Lalanne C, Trinchera L, Tenenhaus A, Moreno A, Zilbovicius M, **Bourgeron T**, Dehaene S, Thirion B, Poline JB, Duchesnay E. Significant correlation between a set of genetic polymorphisms and a functional brain network revealed by feature selection and sparse Partial Least Squares. *Neuroimage* 2012 Jul 8;63(1):11-24.
72. Chaste P, Betancur C, Gérard-Blanluet M, Bargiacchi A, Kuzbari S, Drunat S, Leboyer M, **Bourgeron T**, Delorme R. High-Functioning Autism Spectrum Disorder and Fragile X Syndrome: Report of Two Affected Sisters *Mol Autism*. 2012 3(1): 5
71. Goubran Botros H, Legrand P, Pagan C, Bondet V, Weber P, Ben Abdallah M, Lemièrre N, Huguët G, Bellalou J, Maronde E, Beguin P, Haouz A, Shepard W and **Bourgeron T**. Crystal structure and functional mapping of ASMT, the last enzyme of the melatonin synthesis pathway. *J. Pineal Res*. 54: 46–57.
70. Etain B, Bellivier F, Dumaine A, Deshommes J, Pagan C, Goubran-Botros H, Moreno S, Moustafa K, Mathieu F, Henry C, Kahn JP, Launay JM, **Bourgeron T**, Leboyer M, Jamain S. Functional alterations of ASMT, the last enzyme of the melatonin pathway, in patients with bipolar disorders. *Hum. Mol. Genet*. (2012) 21:4030-7.
69. Sato D, Lionel AC, Leblond CS, Prasad A, Pinto D, Walker S, O'Connor I, Russell C, Drmic IE, Hamdan FF, Michaud JL, Endris V, Roeth R, Delorme R, Huguët G, Leboyer M, Rastam M, Gillberg C, Lathrop M, Stavropoulos DJ, Anagnostou E, Weksberg R, Fombonne E, Zwaigenbaum L, Fernandez BA, Roberts W, Rappold GA, Marshall CR, **Bourgeron T**, Szatmari P and Scherer SW. SHANK1 Deletions in Males with Autism Spectrum Disorder. *Am. J. Hum. Genet*. (2012) 90:879-87
68. 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, Grubbrucker AM, Shoichet SA, Schmitz D, Kreutz MR, **Bourgeron T**, Gundelfinger ED and Boeckers TM. Hyperactivity and autistic-like behaviours in mice lacking ProSAP1/Shank2. *Nature* (2012) 486 : 256-60
67. Leblond CS, Heinrich J, Delorme R, Proepper C, Betancur C, Huguët G, Konyukh M, Chaste P, Ey E, Rastam M, Anckarsäter H, Nygren G, Gillberg IC, Melke J, Toro R, Regnault B, Fauchereau F, Mercati O, Lemièrre N, Skuse D, Poot M, Holt R, Monaco AP, Järvelä I, Kantojärvi K, Vanhala R, Curran S, Collier DA, Bolton P, Chiocchetti A, Klauck SM, Poustka F, Freitag CM, Waltes R, Kopp M, Duketis E, Bacchelli E, Minopoli F, Ruta L, Battaglia A, Mazzone L, Maestrini E, Sequeira AF, Oliveira B, Vicente A, Oliveira G, Pinto D, Scherer SW, Zelenika D, Delepine M, Lathrop M, Bonneau D, Guinchat V, Devillard F, Assouline B, Mouren MC, Leboyer M, Gillberg C, Boeckers TM, **Bourgeron T**. Genetic and functional analyses of SHANK2 mutations provide evidence for a multiple hit model of autism spectrum disorders. *PLoS Genetics* 2012 8:e1002521.
66. Pinel P, Fauchereau F, Moreno A, Barbot A, Lathrop M, Zelenika D, Le Bihan D, Poline JB, **Bourgeron T**, Dehaene S. Genetic Variants of FOXP2 and KIAA0319/TTRAP/THEM2 Locus Are Associated with Altered Brain Activation in Distinct Language-Related Regions. *J Neurosci*. (2012) 32:817-825.
65. Chabaut J, Serreau P, Ey E, Bellier L, Aubin T, **Bourgeron T**, Granon S. Adult male mice emit context-specific ultrasonic vocalizations that are modulated by prior isolation or group rearing environment. *PLoS One*. 2012;7(1):e29401.
64. Casey JP, Magalhaes T, Conroy JM, Regan R, Shah N, Anney R, Shields DC, Abrahams BS, Almeida J, Bacchelli E, Bailey AJ, Baird G, Battaglia A, Berney T, Bolshakova N, Bolton PF, **Bourgeron T**, et al. A novel approach of homozygous haplotype sharing identifies candidate genes in autism spectrum disorder. *Hum Genet*. (2012) 131(4):565-79.

2011

63. Poot M, van der Smagt JJ, Brilstra EH, **Bourgeron T**. Disentangling the Myriad Genomics of Complex Disorders, Specifically Focusing on Autism, Epilepsy, and Schizophrenia. *Cytogenet Genome Res.* (2011) 135(3-4):228-40.
62. Chaste P, Clement N, Botros HG, Guillaume JL, Konyukh M, Pagan C, Scheid I, Nygren G, Anckarsäter H, Rastam M, Ståhlberg O, Gillberg IC, Melke J, Delorme R, Leblond C, Toro R, Huguet G, Fauchereau F, Boudarene L, Serrano E, Lemièrre N, Launay JM, Heath S, Lathrop M, Boland A, Zelenica D, Leboyer M, Jockers R, Gillberg C and **Bourgeron T** Genetic variations of the melatonin pathway in patients with Attention Deficit and Hyperactivity Disorders *J. Pineal Research* 51(4):394-9.
61. Durand CM, Perroy J, Loll F, Fagni L, **Bourgeron T**, Montcouquiol M, Sans N. SHANK3 mutations identified in autism lead to modification of dendritic spine morphology via an actin-dependent mechanism. (2011) *Molecular Psychiatry* 17(1):71-84.
60. Maronde E, Saade A, Ackermann K, Goubran-Botros H, Rami A, **Bourgeron T**, Dehghani F, Stehle JH. Dynamics in enzymatic protein complexes offer a novel principle for the regulation of melatonin synthesis in the human pineal gland. (2011) *J. Pineal Research* 51(1):145-55.
59. Amaral D, Rogers SJ, Baron-Cohen S, **Bourgeron T**, Caffo E, Fombonne E, Fuentes J, Howlin P, Rutter M, Klin A, Volkmar F, Lord C, Minshew N, Nardocci F, Rizzolatti G, Russo S, Scifo R, van der Gaag RJ. Against le packing: a consensus statement. *J Am Acad Child Adolesc Psychiatry.* 2011 Feb;50(2):191-2.
58. Pagan C, Goubran-Botros H, Poirier K, Dumaine A, Jamain S, Moreno S, de Brouwer A, Van Esch H, Delorme R, Launay JM, Tzschach A, Kalscheuer VM, Lacombe D, Briault S, Laumonnier F, Raynaud M, van Bon BW, Willemsen MH, Leboyer M, Chelly J and **Bourgeron T**. Mutation screening of ASMT, the last enzyme of the melatonin pathway, in a large sample of patients with Intellectual Disability. (2011) *BMC Medical Genetics* 2(1):17.
57. Konyukh M, Delorme R, Chaste P, Leblond C, Lemièrre N, Nygren G, Anckarsäter H, Rastam M, Ståhlberg O, Gillberg IC, Mouren-Simeoni MC, Gillberg C, Leboyer M and **Bourgeron T** Mutations of the candidate gene *SEZ6L2* on chromosome 16p11.2 in patients with autism spectrum disorders and in human populations. (2011) *PLoS ONE* 6, e17289
56. Ey E., Leblond C, **Bourgeron T**. Behavioral profiles of mouse models for autism spectrum disorders. *Autism Research.* (2011) 4(1):5-16.
55. Ben-Abdallah M, Bondet V, Fauchereau F, Béguin P, Goubran-Botros H, Pagan C, **Bourgeron T** and Bellalou J. Production of soluble, active acetyl serotonin methyl transferase in *Leishmania tarentolae*. *Protein Expression and Purification* (2011) 75:114-118.

2010

54. Anney R, Klei L, Pinto D, Pagnamenta A, 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.** A genomewide scan for common risk variants nominates phospholipase D and polysialyltransferase proteins for a role in autism. *Hum. Mol. Genet.* (2010) 19:4072-82.
53. Chaste P, Clement N, Mercati O, Guillaume JL, Delorme R, Botros HG, Pagan C, Périvier S, Scheid I, Nygren G, Anckarsäter H, Rastam M, Ståhlberg O, Gillberg IC, Serrano E, Lemièrre N, Launay JM, Mouren-Simeoni MC, Leboyer M, Gillberg C, Jockers R and **Bourgeron T** Identification of pathway-biased and deleterious melatonin receptor mutants in autism spectrum disorders and in the general population *PLoS One* (2010) 5:e11495.
52. Toro R, Konyukh M, Delorme R, Leblond C, Chaste P, Fauchereau F, Coleman M, Leboyer M, Gillberg C and **Bourgeron T**. Key role for gene dosage and synaptic homeostasis in autism spectrum disorders. *Trends in Genetics* (2010) 26:363-372.
51. 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** (+255 authors) Betancur C. Functional impact of global rare copy number variation in autism *Nature* (2010) 466 : 368-72.

2009

50. Delorme R., Scheid I, Anckarsäter H, Chaste P, Schuroff F, Nygren G, Herbrecht E, Jamain S, Mouren MC, Råstam M, Leboyer M, Gillberg C and **Bourgeron T** Mutation Screening of *NOS1AP* Gene in a Large Sample of Psychiatric Patients and Controls *BMC Medical Genetics* (2010) 11:108.
49. **Bourgeron T** A synaptic trek to autism. *Curr. Opin. Neurobiol.* (2009) 19:1-4.
48. Delorme R, Betancur C, Chaste P, Kernéis S, Stopin A, Mouren MC, Collet C, **Bourgeron T**, Leboyer M, and Launay JM Reduced 3-O-methyl-dopa levels in OCD patients and their unaffected parents is associated with the low activity M158 COMT allele. *Am. J. Med Genet.* 2009 153B(2):542-548.
47. Gong X, Delorme R, Fauchereau F, Durand CM, Chaste P, Betancur C, Goubran-Botros H, Nygren G, Anckarsäter H, Rastam M, Gillberg IC, Kopp S, Mouren-Simeoni MC, Gillberg C, Leboyer M, and **Bourgeron T** Normal coding sequence and transcript level of the X-linked gene *RPL10* in autism spectrum disorders. *BMC Medical Genetics* 2009, 10:7

2008

46. Jamain S., Radyushkin K, Hammerschmidt K, Granon S, Boretius S, Varoqueaux F, Ramanantsoa N, Gallego J, Ronnenberg A, Winter D, Frahm J, Fischer J, **Bourgeron T**, Ehrenreich H, and Brose N. Reduced social interaction and ultrasonic communication in a mouse model of monogenic heritable autism. *Proc Natl Acad Sci U S A.* 2008 105:1710-1715.
45. Gong X, Delorme R, Fauchereau F, Durand CM, Chaste P, Betancur C, Goubran-Botros H, Nygren G, Anckarsäter H, Rastam M, Gillberg IC, Mouren MC, Gillberg C, Leboyer M, and **Bourgeron T**. Ascertainment of X chromosome inactivation in autism spectrum disorders reveals families with X-linked mutations. *Am. J. Med. Genetics*, 2008 147B(6):830-5.
44. Melke J, Goubran-Botros H, Chaste P, Betancur C, Nygren G, Anckarsäter H, Rastam M, Ståhlberg O, Gillberg IC, Delorme R, Chabane N, Mouren-Simeoni MC, Fauchereau F, Durand CM, Chevalier F, Drouot X, Collet C, Launay JM, Leboyer M, Gillberg C, and **Bourgeron T** Abnormal Melatonin Synthesis in Autism Spectrum Disorders *Molecular Psychiatry* 2008 13:90-98.
43. Durand CM and **Bourgeron T**. Genetic, neurobiological and clinical findings related to *SHANK3* mutations and 22q13 chromosomal rearrangements in autism spectrum disorders. *European Psychiatry Review* (2008) Vol 1 Issue 1.

Before 2008

42. The Autism Genome Project Consortium; Szatmari P, Paterson AD, Zwaigenbaum L, Roberts W, Brian J, Liu XQ, Vincent JB, Skaug JL, Thompson AP, Senman L, Feuk L, Qian C, Bryson SE, Jones MB, Marshall CR, Scherer SW, Veland VJ, Bartlett C, Mangin LV, Goedken R, Segre A, Pericak-Vance MA, Cuccaro ML, Gilbert JR, Wright HH, Abramson RK, Betancur C, **Bourgeron T**, (+ 110 authors) Meyer KJ. Mapping autism risk loci using genetic linkage and chromosomal rearrangements. *Nature Genetics*. 2007 39:319-28.
41. Durand C, Betancur C, Boeckers TM, Bockmann J, Chaste P, Fauchereau F, Nygren G, Rastam M, Gillberg IC, Ankarsäter 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.
40. Belmonte MK and **Bourgeron T**. Fragile X Syndrome and Autism at the Intersection of Genetic and Neuronal Networks. *Nature Neuroscience* (2006) 9:1221-1225.
39. Persico A. and **Bourgeron T**. Searching for ways out of the autism maze: Genetic, epigenetic, and environmental clues. *Trends in Neuroscience* (2006) 29, 349-358
38. Etain B., Mathieu F., Rietschel M., Wolfgang M, Albus M, Mckeon P, Roche S., Kealey C., Blackwood D, Muir WJ, Bellivier F., Henry C, Dina C., Gallina S, Gurling H., Malafosse A, Preisig M., Ferrero F, Cichon S, Schumacher J, Ohlraun S., Borrmann hassenbach M, Propping P, Abou jamra R., Schulze TG, Marusic A., Dernovsek M, Giros B., **Bourgeron T**, Lemaingue A, Bacq D., Betard C, Charon C, Nöthen M.M., Lathrop M, Leboyer M. Genome-wide scan for genes involved in bipolar affective disorder in 70 European families ascertained through a bipolar type in early onset proband : supportive evidence for linkage at 3p14. *Molecular Psychiatry* 2006 11(7):685-694.
37. Groupe 1: Delorme R, Durand CM, Betancur C, Wagner M, Ruhrmann S, Grabe HJ, Nygren G, Gillberg C, Leboyer M, and **Bourgeron T**; Groupe 2: Courtet P, Jollant F; Groupe 3: Buresi C, Aubry JM, Baud P, Bondolfi G, Bertschy G, Malafosse A. No *hTPH2* R441H mutation in a large cohort of psychiatric patients and controls. *Biological Psychiatry* (2006) 60:202-203.
36. Durand CM, Kappeler C, Betancur C, Delorme R, Quach H, Goubran-Botros H, Melke J, Nygren G, Chabane N, Bellivier F, Szoke A, Schurhoff F, Rastam M, Ankarsäter H, Gillberg C, Leboyer M and **Bourgeron T** Expression and genetic variability of *PCDH11Y*, a gene specific to *Homo sapiens* and candidate for susceptibility to psychiatric disorders. *Am. J. Med. Genet.* (2006) 141, 67-70.
35. Delorme R., C Betancur, M Wagner, MO Krebs, P Gorwood, P Pearl, G Nygren, CM Durand, F Buhtz, P Pickering, J Melke, S Ruhrmann, H Ankarsäter, N Chabane, Kipman A, Reck C, Millet B, Roy I, Mouren-Simeoni MC, Maier W, Råstam M, Gillberg C, Leboyer Mr, and **Bourgeron T**. Support for the association between the serotonin transporter gene and susceptibility to obsessive compulsive disorder. *Molecular Psychiatry* (2005) 10, 1059-1061.
34. Bah J, Quach H, Ebstein RP, Segman RH, Melke J, Jamain S, Rietschel M, Modai I, Kanas K, Karni O, Lerer B, Gourion D, Krebs MO, Etain B, Schürhoff F, Szöke A, Leboyer M and **Bourgeron T**. Maternal transmission disequilibrium of the glutamate receptor *GRIK2* in schizophrenic patients. *Neuroreport* (2004) 15, 1987-1991.
33. Delorme R., Krebs MO., Chabane N., Roy I., Millet B., Mouren-Simeoni MC., Maier W., **Bourgeron T** and Leboyer M. Frequency and Transmission of Glutamate Receptors *GRIK2* and *GRIK3* Polymorphisms in Patients with Obsessive Compulsive Disorder; *Neuroreport* (2004) 15, 699-702.
32. Jamain S, Quach H, Betancur C, Råstam M, Colineaux C, Gillberg IC, Soderstrom H, Giros B, Leboyer M, Gillberg C, **Bourgeron T** and the Paris study. Mutations of the X-linked neuroligins *NLGN3* and *NLGN4* are associated with autism *Nature Genetics* (2003) 34, 27-29.
31. Jamain S, Betancur C, Quach H, Philippe A, Fellous M, Giros B, Gillberg C, Leboyer M, **Bourgeron T** and the Paris Study. Linkage and association between glutamate receptor 6 and autism. *Molecular Psychiatry* (2002) 7, 302-310.
30. Jamain S, Quach H, Quintana-Murci L, Betancur C, Philippe A, Gillberg C, Sponheim E, Skjeldal OH, Fellous M, Leboyer M, and **Bourgeron T**. Y chromosome haplogroups in autistic subjects. *Molecular Psychiatry* (2002) 7, 217-219.
29. Jamain S, Girondot M, Leroy P, Clergue M, Quach H, Fellous M and **Bourgeron T**. Transduction of the Human Gene *FAM8A1* by Endogenous Retrovirus During Primate Evolution. *Genomics* (2001) 78, 38-45.
28. Quintana-Murci L, Rötig A, Munnich A, Rustin P and **Bourgeron T**. Mitochondrial DNA inheritance in patients with deleted mtDNA. *J Med Genet* (2001), 38, e28
27. Jamain S, Quach M, Fellous M, and **Bourgeron T**. Identification of the human KINESIN-13A gene homologous to Drosophila kinesin-73 and candidate for schizophrenia. *Genomics* (2001) 74, 36-44.
26. Krausz C, Quintana-Murci L, Barbaux S, Siffroi JP, Rouba H, Delafontaine D, Souleyreau-Therville N, Arvis G, Antoine JM, Erdei E, Taar JP, Tar A, Jeandidier E, Plessis G, **Bourgeron T**, Dadoune JP, Fellous M, McElreavey K. A high frequency of Y chromosome deletions in males with nonidiopathic infertility. *J Clin Endocrinol Metab* (1999) 84, 3606-12.
25. Krausz C, Siffroi J, Souleyreau-Therville N, **Bourgeron T**, McElreavey K, Fellous M. Genetic control of spermatogenesis: Y chromosome and male infertility. *Ann Biol Clin.* (1999) 57, 309-173.
24. Algonik A.I., Zharkikh A., and Boettger-Tong H (Groupe 1)
Bourgeron T., McElreavey K., and Bishop C.E. (Groupe 2) Evolution of the DAZ gene family suggests that Y-linked DAZ plays little, or a limited, role in spermatogenesis but underlines a recent African origin for human populations. *Hum. Mol. Genet.* (1998) 7, 1371-1377
23. Rustin P, **Bourgeron T**, Parfait B, Chretien D, Munnich A, Rötig A. Inborn errors of the Krebs Cycle. *Biochem Biophys Acta* (1997) 1361, 185-197
22. Seboun E, Barbaux S, **Bourgeron T**, Nishi S, Algonik A, Egashira M, Nikkawa N, Bishop C, Fellous M, McElreavey K, Kasahara M. Gene sequence, localisation and evolutionary conservation of DAZLA, a candidate male sterility gene. *Genomics* (1997) 41, 227-235
21. Munnich A, Rotig A, Chretien D, Cormier V, **Bourgeron T**, Bonnefont JP, Saudubray JM, Rustin P. Clinical presentation of mitochondrial disorders in childhood. *J Inher Metab Dis* (1996) 19, 521-527
20. Rustin P, Parfait B, Chretien D, **Bourgeron T**, Djouadi F, Bastin J, Rötig A, Munnich A. Fluxes of nicotinamide adenine dinucleotides through mitochondrial membranes in human cultured cells. *J Biol Chem* (1996) 271, 14785-14790
19. **Bourgeron T**, Rustin P, Chretien D, Birch-Machin M, Bourgeois M, Viegas-Péquignot E, Munnich A, Rötig A. A mutation in the flavoprotein subunit gene of the succinate dehydrogenase: the first nuclear gene mutation in mitochondrial respiratory chain deficiency. *Nature Genetics* (1995) 11, 144-148
18. Rötig A, **Bourgeron T**, Rustin P, Munnich A. Phenotypic expression of mitochondrial genotypes in cultured skin fibroblasts and in Epstein-Barr virus-transformed lymphocytes in Pearson syndrome. *Muscle & Nerve* (1995) 3, S159-64

17. Rötig A, **Bourgeron T**, Chretien D, Rustin P, Munnich A. Spectrum of mitochondrial DNA rearrangements in the Pearson-marrows syndrome. *Hum Mol Genet* (1995) 4, 1327-1330
16. Chretien D, Pourrier M, **Bourgeron T**, Séné M, Rötig A, Munnich A, Rustin P. An improved spectrophotometric assay of pyruvate dehydrogenase in lactate dehydrogenase-contaminated mitochondrial preparations from human skeletal muscle. *Clin Chim Acta* (1995) 240, 129-136
15. Rötig A, Lehnert A, Rustin P, Chretien D, **Bourgeron T**, Niaudet P, Munnich A. Kidney involvement in mitochondrial disorders. *Advances in Nephrology from the Necker Hospital* (1995) 24, 367-78
14. **Bourgeron T**, Chretien D, Poggi-Bach J, Doonan S, Rabier D, Letouze P, Munnich A, Rötig A, Landrieu P, Rustin P. Mutation of the fumarase gene in two siblings with progressive encephalomyopathy and fumarase deficiency. *J Clin Invest* (1994) 93, 2514-2518
13. Poulton J, Sewry C, Potter CG, **Bourgeron T**, Chretien D, Wijburg FA, Morten KJ, Brown G. Variation in mitochondrial DNA levels in muscle from normal controls. Is depletion of mtDNA in patients with mitochondrial myopathy a distinct clinical syndrome? *J Inher Metab Dis* (1994) 18, 4-20
12. Chretien D, Rustin P, **Bourgeron T**, Rötig A, Saudubray JM, Munnich A. Reference charts for respiratory chain activities in human tissues. *Clin Chim Acta* (1994) 228, 53-70
11. Rustin P, Chretien D, Gérard B, **Bourgeron T**, Rötig A, Saudubray JM, Munnich A. Biochemical and molecular investigations in respiratory chain deficiencies. *Clin Chim Acta* (1994) 228, 35-51
10. Rustin P, Le Bidois J, Chretien D, **Bourgeron T**, Piechaud JF, Rötig A, Munnich A, Sidi D. Endomyocardial biopsies for early detection of mitochondrial disorders in hypertrophic cardiomyopathies. *J Pediatr* (1994) 124, 224-228
9. **Bourgeron T**, Chretien D, Rötig A, Munnich A, Rustin P. Fate and expression of the deleted mitochondrial DNA differ between heteroplasmic skin fibroblast and Epstein-Barr virus-transformed lymphocyte cultures. *J Biol Chem* (1993) 268, 19369-19376
8. **Bourgeron T**, Chretien D, Amati P, Rötig A, Munnich A, Rustin P. Expression of respiratory chain deficiencies in human cultured cells. *Neuromusc Disord* (1993) 3, 605-608
7. Rustin P, Chretien D, **Bourgeron T**, Le Bidois J, Sidi D, Rötig A, Munnich A. The investigation of respiratory chain activity in human heart. *Biochem Med Metab Biol* (1993) 50, 120-126
6. Rustin P, Le Bidois J, Chretien D, **Bourgeron T**, Piechaud JF, Rötig A, Sidi D, Munnich A. The investigation of respiratory chain disorders in heart using endomyocardial biopsies. *J Inher Metab Dis* (1993) 16, 541-544
5. **Bourgeron T**, Chretien D, Rötig A, Munnich A, Rustin P. Isolation and characterization of mitochondria from human B lymphoblastoid cell lines. *Biochem Biophys Res Commun* (1992) 186, 16-23
4. **Bourgeron T**, Chretien D, Rötig A, Munnich A, Rustin P. Prenatal diagnosis of cytochrome c oxidase deficiency in cultured amniocytes is hazardous. *Prenatal Diagnosis* (1992) 12, 548-549
3. Gérard B, **Bourgeron T**, Chretien D, Rötig A, Munnich A, Rustin P. Uridine preserves the expression of respiratory enzyme deficiencies in cultured fibroblasts. *Eur J Pediatr* (1992) 152, 270
2. Rustin P, Chretien D, **Bourgeron T**, Wucher A, Saudubray JM, Rötig A, Munnich A. Assessment of the mitochondrial respiratory chain. *Lancet* (1991) 338, 60
1. Chretien D, **Bourgeron T**, Rötig A, Munnich A, Rustin P. The measurement of the rotenone-sensitive NADH cytochrome c reductase in mitochondria isolated from minute amount of human skeletal muscle. *Biochem Biophys Res Commun* (1990) 173, 26-33

BOOK CHAPTERS

11. Ferhat AT, Halbedl S, Schmeisser MJ, Kas MJ, **Bourgeron T**, Ey E. Behavioural Phenotypes and Neural Circuit Dysfunctions in Mouse Models of Autism Spectrum Disorder. In *Translational Anatomy and Cell Biology of Autism Spectrum Disorder 2017 Volume 224 of the series Advances in Anatomy, Embryology and Cell Biology* pp 85-101
10. Huguet G. and **Bourgeron T**. The genetic causes of autism spectrum disorders. In *Synaptic Dysfunction in Autism Spectrum Disorder and Intellectual Disability A Time for Metabolism and Hormones 2016* pp 101-129
9. **Bourgeron T**. Genetics and Epigenetics of autism spectrum disorders. In *Epigenetics, Brain and Behavior*. 2012. Ed. Editors: Paolo Sassone Corsi, Yves Christen, pp 105-132
8. Pagan C, Delorme R, Launay JM and **Bourgeron T**. The alterations of the serotonin-melatonin pathway in autism spectrum disorders: from biological evidence to clinical consequence. In *The Autisms: Molecules to Model Systems*.
7. **Bourgeron T**. Cell adhesion Molecules in Synaptopathies. In *The Sticky Synapse. Cell Adhesion Molecules and Their Role in Synapse Formation and Maintenance*. Hortsch, Michael (Ed.) (2009)
6. **Bourgeron T** Genetics of Autism Spectrum Disorders. *Encyclopedia of Molecular Mechanisms of Disease*. Lang, Florian (Ed.) 2009, LXXXVI, 2270 p. 646.
5. **Bourgeron T** Genes, Synapses and autism spectrum disorders. In *Synaptic plasticity and the mechanism of Alzheimer's disease*. Ed. DJ Selkoe, A. Triller, Y. Christen. (2008) 169-180.
4. **Bourgeron T** The Possible Interplay of Synaptic and Clock Genes in Autism Spectrum Disorders. *Cold Spring Harb Symp Quant Biol*. 2007;72:645-54.
3. **Bourgeron T**, Jamain S and Granon S. Animal models of autism : Focus on genetics models and behavioral paradigms. (2005) In *Transgenic and Knockout Models of Neuropsychiatric Disorders* Fisch, Gene S. Ed. Humana Press. 450p
2. **Bourgeron T** and Giros B. Genetic Markers in Psychiatry. In *Psychiatry Genetics Methods and Protocols* Ed. Leboyer M. and Bellivier F. (2003) *Methods Mol Med*. 77, 63-98.
1. **Bourgeron T**. Mitochondrial function and male infertility. In *The genetic basis of male infertility*. K. McElreavey Ed. *Results Probl. Cell. Differ.* (2000) 28, 187-210