
BIOGRAPHICAL SKETCH

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NAME Hallmayer, Joachim Franz M.D.	POSITION TITLE Associate Professor of Psychiatry		
eRA COMMONS USER NAME JOACHIMH			
EDUCATION/TRAINING <i>(Begin with baccalaureate or other initial professional education, such as nursing, and include postdoctoral training.)</i>			
INSTITUTION AND LOCATION	DEGREE <i>(if applicable)</i>	YEAR(s)	FIELD OF STUDY
University of Cologne, Germany	M.D.	1986	Medicine
University of Cologne, Germany	Dr. med.	1989	Medicine

A. Personal Statement

The goal of this collaborative project is to recruit 40 patients with 22q11 deletion syndrome and 20 healthy controls, evaluate and document carefully their psychiatric symptoms, harvest their skin cells, transform the fibroblasts into induced pluripotent stem cells, and characterize the phenotype and transcriptome of neurons derived from these stem cells. For several years Drs. Dolmetsch and Geschwind and I have collaborated successfully on a NIMH funded project that explores the neuronal phenotype in children with autism in conjunction with known genetic abnormalities. The current proposal is a logical extension and builds on this work. I have the appropriate scientific background and experience to ensure that the project will be completed successfully. I have been involved in genetic studies on psychiatric disorders for over 20 years and have conducted several genome wide linkage and association studies. My experience encompasses both the laboratory and the analytic side. Currently, I am the Chair of the Committee of Senior Investigators of the largest collaborative research project on autism, the Autism Genome Project, with over 100 researchers from 50 centers in North America, Europe, and Asia. I am also the PI of the largest twin study on autism to date, a collaborative effort that includes five universities.

B. Positions and Honors.

Positions and Employment

1983-1986 Postgraduate Fellow, Max-Planck-Institute for Neurological Research, Cologne, Germany
1986-1987 Research Officer, Max-Planck-Institute in Cologne, Germany
1987-1989 Scientific Employee, Psychiatric Hospital of the Johannes Gutenberg University of Mainz, Germany
1989-1991 Research Fellow (German Research Association-DFG) Department of Genetics, Stanford University, Stanford, CA
1991-1994 Research Fellow, Department of Genetics, Stanford University, Stanford, CA
1994-1995 Research Associate, Nancy Pritzker Laboratories, Department of Psychiatry, Stanford University, Stanford, CA
1995-2001 Associate Professor of Psychiatry, University of Western Australia; Co-Director, Centre for Clinical Research in Neuropsychiatry, Crawley, WA, Australia
2001-2007 Associate Professor (Research), Department of Psychiatry and Behavioral Sciences, Stanford University, Stanford, CA
2007- Associate Professor with Tenure, Department of Psychiatry and Behavioral Sciences, Stanford University, Stanford, CA

Honors

1989 Fellow of the German Research Association (DFG)
1991 NARSAD Young Investigator Award
1994 Editorial Board, Psychiatric Genetics

Principal Investigator (Last, First, Middle): Hallmayer, Joachim F

2001- Section Editor, Current Psychiatry Reports - Nonschizophrenic Psychotic Disorders
2008- Autism Genetic Resource Exchange (AGRE) Steering Committee

C. Selected peer-reviewed publications (out of a total of over 150).

Most relevant to the current application

1. The Autism Genome Project Consortium (2010) Functional impact of global rare copy number variation in autism spectrum disorders. *Nature* 466:368-72. PMC3021798
2. Hallmayer J, Cleveland S, Torres A, Phillips J, Cohen B, Torigoe T, Miller J, Fedele A, Collins J, Smith K, Lotspeich L, Croen LA, Ozonoff S, Lajonchere C, Grether JK, Risch N (2011). Genetic heritability and shared environmental factors among twin pairs with autism. *Archives of General Psychiatry* Nov;68(11):1095-102.
3. Yazawa M, Hsueh B, Jia K, Pasca AM, Bernstein JA, Hallmayer J, Dolmetsch RE Using induced pluripotent stem cells to investigate cardiac phenotypes in patients with Timothy Syndrome. *Nature* 2011 Mar 10;471(7337):230-4.
4. Paşca SP, Portmann T, Voineagu I, Yazawa M, Shcheglovitov A, Paşca AM, Cord B, Palmer TD, Chikahisa S, Nishino S, Bernstein JA, Hallmayer J, Geschwind DH, Dolmetsch RE. (2011) Using iPSC-derived neurons to uncover cellular phenotypes associated with Timothy syndrome. *Nature Medicine* Nov 27;17(12):1657-62.
5. Froehlich W, Cleveland S, Torres A, Phillips J, Cohen B, Torigoe T, Miller J, Fedele A, Collins J, Smith K, Lotspeich L, Croen LA, Ozonoff S, Lajonchere C, Grether JK, Hallmayer J. Head Circumferences in Twins with and Without Autism Spectrum Disorders. *J Autism Dev Disord*. 2013 Jan 16. [Epub ahead of print]

Additional recent publications of importance to the field (in chronological order)

1. Wong D, Maybery M, Bishop DV, Maley A, Hallmayer J. Profiles of executive function in parents and siblings of individuals with autism spectrum disorders. *Genes Brain Behav*. 2006 Nov;5(8):561-76.
2. O'Hara R, Schröder CM, Gray H, Schatzberg AF, Kraemer HC, Noda A, Lin X, Hallmayer J (2007). Serotonin Transporter Polymorphism, Cortisol and Cognition in Older Adults. *Molecular Psychiatry* 12:544-555. PMC2084475
3. Singh J, Hallmayer J, Illes J (2007) Interacting and Paradoxical Forces in Neuroscience and Society. *Nature Neuroscience Review*. 8(2):153-60. PMC1885680
4. The Autism Genome Project Consortium (2007) Mapping autism risk loci using genetic linkage and chromosomal rearrangements. *Nature Genetics*. 39:319-28
5. Gotlib IH, Joormann J, Minor KL, Hallmayer J (2008) HPA axis reactivity: a mechanism underlying the associations among 5-HTTLPR, stress, and depression. *Biological Psychiatry* 63:847-51.
6. Singh J, Illes J, Lazzeroni L, Hallmayer J. Trends in US autism research funding. *J Autism Dev Disord*. 2009 May;39(5):788-95.
7. Hallmayer, J., Faraco, J., Lin, L., et al. (2009). Narcolepsy is strongly associated with the TCR alpha locus. *Nature Genetics* 41(6):708-711. PMC2803042
8. Pagnamenta AT, Khan H, Walker S, Gerrelli D, Wing K, Bonaglia MC, Giorda R, Berney T, Mani E, Molteni M, Pinto D, Le Couteur A, Hallmayer J, Sutcliffe JS, Szatmari P, Paterson AD, Scherer SW, Vieland VJ, Monaco AP. Rare familial 16q21 microdeletions under a linkage peak implicate cadherin 8 (CDH8) in susceptibility to autism and learning disability. *J Med Genet*. 2011 Jan;48(1):48-54. PMC3003876
9. Thompson W, Hallmayer J, O'Hara R Design Considerations for Characterizing Psychiatric Trajectories across the Lifespan (2011). *American Journal of Psychiatry* 168(9):894-903.
10. Anney R, and the Autism Genome Project Consortium. Individual common variants exert weak effects on the risk for autism spectrum disorders. *Hum Mol Genet*. 2012 Nov 1;21(21):4781-92.

Principal Investigator (Last, First, Middle): Hallmayer, Joachim F

D. Research Support.

ACTIVE:

R33 MH087898 (Hallmayer)

09/01/09-05/31/13

NIMH

Exploring the Neuronal Phenotype of Autism Spectrum Disorders Using Induced Pluripotent Stem Cells

The goal of this project is to develop the methods to convert skin cells from patients with autism into neurons and to characterize these neurons using high content screens. These experiments will allow researchers to study the neurons of individuals diagnosed with autism and will lead to a better understanding of the development and differentiation of neurons.

OVERLAP: None

R01 MH083972-03 (Hardan)

02/01/09 - 12/30/13

NIMH

A Neuroimaging Study of Twin Pairs with Autism

This investigation examines brain anatomy and metabolism in monozygotic and dizygotic twins with autism along with cognitive and behavioral correlates using multimodal imaging techniques including structural MRI, diffusion tensor imaging, and proton spectroscopy.

OVERLAP: None

P50 NS023724 (Mignot)

08/15/06-7/31/16

NINDS

Center for Narcolepsy and Related Disorders

Project F: Genomic Analysis in Narcolepsy-Cataplexy

To conduct a genome scan in 50 well characterized multiplex narcolepsy families and to conduct cSNP genome-wide association study in 250 well characterized trios.

OVERLAP: None

DP2 OD004445 (Kesler)

9/30/08-8/31/13

NIH

Assessment and Treatment of Cognitive Deficits in Breast Cancer

This study will determine the neurobiologic and genetic correlates of cognitive deficits in women with breast cancer as well as examine programs for rehabilitating these deficits.

OVERLAP: None

Sorenson Molecular Genealogy Fdn (Hallmayer) 9/1/06-05/31/13

Private Foundation

Targeting novel y-chromosome binary polymorphisms

OVERLAP: None

R01HL09492-01A1 (Krystal)

10/1/09 – 6/30/13

Duke University Subcontract/NIH Prime

Biomarkers of the Response to CBT for Insomnia in Major Depression

The study goal is to develop non-REM EEG and genetic predictors of the CBT response in MDD, and also improve the understanding of homeostatic and arousal-related mechanisms that play a role in sleep disturbance in insomnia and are targeted by the components of CBT.

Principal Investigator (Last, First, Middle): Hallmayer, Joachim F

OVERLAP: None

Simon's Foundation (O'Hara) 10/1/10 – 9/31/13
Private Foundation

Characterizing the Type, Severity and Impact of Sleep Disorders in ASD

To use a combination of standard and novel objective assessments of sleep including polysomnography to examine and improve current approaches for characterization of sleep disorders in children with autism spectrum disorders.

OVERLAP: None

106956 (Zwaigenbaum) 03/01/12 – 02/28/15
Alberta Heritage Foundation For Medical Research

Genomic Influence on Developmental Course and Outcomes in Infants at Risk of ASD: A BRSC Study

OVERLAP: None

P50 HG00338908 (Cho) 07/01/04-02/28/15
NIH

Center for Integrating Ethics in Genetics Research

To serve as a center of excellence in neurogenetics research, to develop a national model for bench to bedside research ethics consultation, and to provide training opportunity in biomedical ethics.

OVERLAP: None

PENDING:

OVERLAP: None

Tissue Collection for Disease Modeling (Hallmayer) 06/01/13 – 05/31/15
California Institute for Regenerative Medicine (CIRM)
Induced pluripotent stem cells from children with autism spectrum disorders

In this study our goal is to contribute blood and skin samples for hiPSC research from 200 children with an ASD and 100 control subjects to the CIRM repository. These cells will be made available to the wider research community for hiPSC research. All subjects will be assessed using a protocol that provides a thorough clinical picture as well as measurement of the intellectual abilities of the participants. This resource will enable research on hiPSC-derived neurons on a scale and depth that is unmatched anywhere else in the world.