BIOGRAPHICAL SKETCH

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NAME: Tischfield, Jay A.

eRA COMMONS USER NAME (credential, e.g., agency login): tischfield

POSITION TITLE: MacMillan Distinguished Professor of Genetics, Pediatrics and Psychiatry/ Executive Director, Human Genetics Institute of New Jersey

EDUCATION/TRAINING *(Begin with baccalaureate or other initial professional education, such as nursing, include postdoctoral training and residency training if applicable. Add/delete rows as necessary.)*

INSTITUTION AND LOCATION	DEGREE *(if applicable)*	Completion Date MM/YYYY	FIELD OF STUDY
Brooklyn College, Brooklyn, NY	B.S.	06/1967	Biology
Yale University, New Haven, CT	M.Ph.	06/1969	Biology
Yale University, New Haven, CT	Ph.D.	06/1973	Biology

A. Personal Statement

My current research interests include the genetic architecture(s) of alcohol and drug abuse and Tourette disorder. Gene discovery has led us to functional disease models utilizing both neuronally-differentiated induced pluripotent stem cells (iPSCs), which we now also produce as a service to two NIH institutes, as well as to engineered mouse models. I have co-authored 320 research papers and am PI on awards that collect, process, bank and analyze human samples from throughout the world, with the lab work originally done at RUCDR Infinite Biologics®, of which I was CEO. Implementing new technologies, state-of-the-art science, and high-throughput automation within RUCDR provides opportunities to facilitate the research of hundreds of basic and clinical scientists and to provide training opportunities for predoctoral and postdoctoral students. It has also led to rewarding “big-science” collaborations. In June 2020, RUCDR assets were privatized with the formation of Infinite BiologiX®, a company in which I have no ownership interest.

Ongoing and recently completed projects that I would like to highlight include:

Ongoing Research Support

1. U24 MH068457-19 Tischfield/Brzustowicz (MPI) 09/26/98 - 02/28/25
NIH/NIMH – Limited Competition: Continuation of the Center for Genomic Studies on Mental Disorders
Cooperative Agreement to establish a repository of DNA, cell lines, RNA/cDNA and other biomaterials from individuals with various mental disorders and to maintain complete genotype and phenotype databases of subjects and to distribute these to NIMH-approved researchers. We also develop new molecular analysis protocols for biosamples, web-based bioinformatics approaches to data queries, and statistical/computational methods for genetic data from the NIMH Genetics Initiative.

2. U10AA008401 Porjesz/Tischfield (Co-PI) 09/01/89 - 08/31/23
NIH/NIAAA - Collaborative Study on the Genetics of Alcoholism
With seven participating institutions we aim to identify genes for vulnerability to alcoholism and alcohol abuse. Rutgers maintains the NIAAA/COGA Sharing Repository and conducts gene and transcriptome expression studies and functional studies on blood and cell lines.
Role: Co-Investigator (Site PI)

3. R01 MH115958-03 Heiman/Tischfield (MPI) 06/15/18-03/31/24
NIH/NIMH- 1/7 Collaborative Genomic Studies of Tourette Disorder

The major goals of this project are to collect an additional 1000+ subjects affected with Tourette disorder, and their family members, to identify specific genes underpinning the risk for TD and make collected samples available to the scientific community.
Role: MPI

4. HHSN271201800023I Tischfield 09/30/99 - 03/13/23
NIH/NIDA – NIDA Center for Genetic Studies
This contract maintains and expands the NIDA Center for Genetic Studies to create phenotypic and genotypic databases, cell lines, DNA, RNA/cDNA from samples and materials received by projects funded under other grants supporting research on the genetics of addiction vulnerability. The data and biomaterials are widely distributed to qualified investigators and consultation on their use and analysis is provided.
Role: PI

5. 2U24NS095914-06 Tischfield 09/30/15 - 07/31/25
NIH/NINDS - The NINDS Human Cell and Data Repository
The NINDS Repository was created to collect and distribute a variety of human cells such as skin fibroblasts and blood cells and convert these to iPSCs for research. Given the complexity and cost of generating iPSC lines of high quality, and the emerging importance of disease modeling and regenerative medicine, there is a tremendous need in the neuroscience field for a reliable centralized source of these materials.
Role: PI

6. FY23 Tourette Syndrome Repository Tischfield/Heiman (MPI) 07/01/16 - 06/30/23
State of NJ/NJCTS–Center of Excellence for Tourette Syndrome & Associated Disorders Genetics Repository
This project is to ascertain, clinically phenotype, and reposit biologics from families with a proband affected with Tourette disorder (TD). These cell lines and DNA become part of a public repository of Tourette samples.
Role: PI

7. HHSN275201800001C Tischfield (PI) 12/01/22 – 11/30/27
NESARC-III DNA Storage
The goal of this project is to safely and efficiently provide for all storage activities for NESARC III DNA Repository
Role: PI

B. Positions, Scientific Appointments, and Honors

Positions and Employment

2005 - 2022 Executive Director, Human Genetics Institute of New Jersey, Piscataway, NJ
2001 - Professor, Center for Alcohol Studies, Rutgers Univ., Piscataway, NJ
2000 - Professor, Toxicology, EOHSI, Rutgers Univ., Piscataway, NJ
1999 - Professor, Pediatrics and Psychiatry, Robert Wood Johnson Med School, Piscataway, NJ
1998 - 2010 Founding Chair, Dept. of Genetics, Rutgers University, Piscataway, NJ
1998 - MacMillan Distinguished Professor, Dept. of Genetics, Rutgers Univ., Piscataway, NJ
1987 - 1998 Professor & Division Chief, Medical & Molecular Genetics, Indiana Univ. School of Medicine, Indianapolis, IN
1978 - 1986 Assoc. Prof/Prof, Anatomy, Cell & Molec. Biol., & Pediatrics, Med Col of GA, Augusta, GA
1972 - 1978 Assistant Prof, Biology & Pediatrics, Case Western Reserve University, Cleveland, OH

Other Experience (selected, since 1990)

2019 - Board of Directors NJ Alliance for Clinical and Translational Science
2019 - Member, Internal Advisory Board, Rutgers Center for Advanced Biotechnology and Medicine
2016 - 2019 Member, Genetics Diagnostics to Cancer Treatment Program Oversight Committee
2016 - Member, Functional Genomics Shared Resource Advisory Committee
2015 - Member, Internal Advisory Board Rutgers Brain Health Institute
2013 - 2018 Head, Medical Advisory Board, New Jersey Center for Tourette Syndrome
2012 - 2020 Board of Directors, The BioProcessing Solutions Alliance
2012 - CINJ/RWJMS Precision Medicine Initiative Medicine Internal Advisory Board
2006 - Scientific Advisory Board, Center for Treatment of Addictions, Rockefeller University, NY
2006 - 2012 Steering Committee, Stem Cell Institute of NJ
2005 - 2010 Advisor, Singapore Tissue Network

2004 - 2009 Member, New Jersey Tourette Syndrome Center of Excellence Task Force
2002 - 2018 Chair, Scientific Advisory Board, Motif Bio, plc
2001 - 2019 Editorial Board, Mutation Research
2001 - 2005 Scientific Advisory Board, Genome Institute of Singapore
1999 - Internal Advisory Board, Cancer Center of New Jersey
1998 - 2020 Founder, CEO and Scientific Director, RUCDR Infinite Biologics®, Piscataway, NJ
1994 - 1996 Vice President, Indiana University Medical Genetics, Inc., Indianapolis, IN
1992 - 2007 Editorial Advisory Board, *J. of Molecular Medicine*

Professional Certification

1993 - Founding Fellow, American College of Medical Genetics
1993 - 2019 Diplomate, American Board of Medical Genetics, Clinical Molecular Geneticist (expired 2019)
1987 - Diplomate, American Board of Medical Genetics Ph.D. Medical Geneticist (current)

Honors

BioNJ 2022 Annual Dinner Meeting & Innovation Celebration. The event honored New Jersey's COVID-19 Heroes – BioNJ Members who made a significant impact on New Jersey and the world in the battle against COVID-19. Dr. Andrew Brooks (posthumously): Led the Discovery of the First COVID-19 Saliva-Based Test. Rutgers, The State University of New Jersey: FIRST Saliva COVID-19 Test (2022)

Tourette Syndrome Lifetime Achievement Award from the NJ Center for Tourette Syndrome during the NJCTS Annual Awards Celebration & Tourette Syndrome Awareness Day Luncheon (2022)

Research & Development Council of NJ 41st Edison Patent Awards Ceremony. Healthcare Institute of NJ presented Rutgers-Infinity BiologiX with a research award recognizing the development of the SARS-CoV2 saliva test (2020)

Health Care Institute of New Jersey (HINJ) Recognition Award for Research and Innovation (2019)

NJBIZ Healthcare Heroes Awards Program. TIC (Tourette International Collaborative) Genetics was a finalist in the Innovation Hero- Organization category (2019)

NJBIZ's 100 Most Powerful People in New Jersey Business, #85 (2011)

Rutgers University Board of Trustees Award for Excellence in Research (2011)

Election as American Association for the Advancement of Science Fellow (2007)

Duncan and Nancy MacMillan Endowed Chair in Genetics (1999 -)

Elliot Osserman Distinguished Service Award in Support of Cancer Research, Israel Cancer Res Fund (1994)

Distinguished Alumnus Award and Medal, Brooklyn College of the City University of New York (1990)

C. Contribution to Science

My contributions to science have been several, as described in ~335 peer-reviewed publications as a consequence of 40 years of continuous NIH funding.

I. I became interested in mutagenesis and genomic instability, which led to ~70 publications over a 35-year period describing the selectable adenine phosphoribosyltransferase (APRT) gene and its mutagenesis. Our group was the first to describe in detail, mitotic recombination in mammalian cells (human and mouse) and we created human cell culture and mouse models to study the process.

1. Shao, C., Deng, L., Henegariu, O., Liang, L., Raikwar, N., Sahota, A., Stambrook, P.J., and Tischfield, J.A. Mitotic recombination produces the majority of recessive fibroblast variants in heterozygous mice. Proc. Natl. Acad. Sci., USA 96:9230-9235 (1999). PMCID: PMC17762
2. Shao, C., Stambrook, P.J., and Tischfield, J.A. Mitotic recombination is suppressed by chromosomal divergence in hybrids of distantly related mouse strains. Nature Genetics 28:169-172 (2001). SEE NEWS AND VIEWS by Vrieling, Nature Genetics 28:101 - 103 (2001)
3. Serrano L, Liang L, Chang Y, Deng L, Maulion C, Nguyen SC, Tischfield JA. Homologous Recombination conserves DNA sequence integrity throughout the cell cycle in embryonic stem cells. Stem Cells Dev. 2011 Feb;20(2):363-74. Epub 2010 Oct 29. PMCID: PMC3128761.
4. Vazquez BN, Thackray JK, Simonet NG, Chahar S, Kane-Goldsmith N, Newkirk SJ, Lee S, Xing J, Verzi MP, An W, Vaquero A, Tischfield JA, Serrano L. SIRT7 mediates L1 elements transcriptional repression and their association with the nuclear lamina. Nucleic Acids Res. 2019 Sep 5;47(15):7870-7885. PMCID: PMC6735864.

II. My lab produced and described, from ES cells targeted in-house, the first mouse models for two inherited human diseases that exhibit kidney stones as the most prominent phenotype. We are now engaged in drug discovery for stone prevention.

1. Engle, S.J., Stockelman, M.G., Chen, J., Boivin, G., Yum, M-N., Davies, P.M., Ying, M.Y., Sahota, A.S., Simmonds, H.A., Stambrook, P.J., and Tischfield, J.A. Adenine phosphoribosyltransferase-deficient mice develop 2,8-dihydroxadenine nephrolithiasis. Proc. Nat. Acad. Sci., USA, 93:5307-5312 (1996). PMCID: PMC39241
2. Evan, A.P., Bledsoe, S., Connors, B.A., Deng, L., Shao, C., Liang, L., Fineberg, N., Grynpass, M.D., Stambrook, P.J., Sahota, A. and Tischfield, J.A. Sequential analysis of kidney stone formation in the Aprt knockout mouse. Kidney Int. 60:910-923 (2001). COVER PAGE PHOTO
3. Sahota A, Parihar JS, Capaccione KM, Yang M, Noll K, Gordon D, Reimer D, Yang I, Buckley BT, Polunas M, Reuhl KR, Lewis MR, Ward MD, Goldfarb DS, Tischfield JA. Novel cystine ester mimics for the treatment of cystinuria-induced urolithiasis in a knockout mouse model. Urology. 2014 Nov;84(5):1249.e9-15. doi: 10.1016/j.urology.2014.07.043. Epub 2014 Oct 24. PMCID: PMC4498569
4. Sahota A, Tischfield JA, Goldfarb DS, Ward MD, Hu L. Cystinuria: genetic aspects, mouse models, and a new approach to therapy. Urolithiasis. 2019 Feb;47(1):57-66. Epub 2018 Dec 4. PMCID: PMC6592844

III. In the course of research on Batten disease, my graduate student (Ju Chen, now American Heart Association Professor at UCSD) stumbled on the first and second novel low molecular weight phospholipases. We then proceeded to describe this interesting family of genes and proteins in a series of 12 publications that had profound effect on inflammation research, causing two of the largest pharmaceutical companies to reevaluate their programs.

1. Chen, J., Engle, S.J., Seilhamer, J.J., and Tischfield, J.A. Cloning and recombinant expression of a novel human low molecular weight Ca^{2+}-dependent phospholipase A_2*. J. Biol. Chem., 269:2365-2368 (1994).
2. Tischfield, J.A. Minireview: A reassessment of the low molecular weight phospholipase A2 gene family in mammals. J. Biol. Chem. 272:17247-17250 (1997).
3. Murakami, M., Shimbara, S., Kambe, T., Kuwata, H., Winstead, M.V., Tischfield, J.A., and Kudo, I. The functions of five distinct mammalian phospholipase A_2s in regulating arachidonic acid release. J. Biol. Chem. 273(23):14411-14423 (1998).

IV. I continue with my interest in the genetics of alcoholism and drug abuse. Much of the team science on alcohol abuse has been over the past 31 years as one of seven NIAAA-funded centers of the **C**ollaborative **O**n the **G**enetics of **A**lcoholism (COGA), coauthoring ~75 publications. This work has produced much of the known body of work on the genetics of alcoholism.

1. Bond, C., LaForge, S., Tian, M., Melia, D., Zhang, S., Borg, L., Gong, J., Schluger, J., Strong, J.A., Leal, S.M., Tischfield, J.A., Kreek, M.J., and Yu, L. Single nucleotide polymorphism in the human mu opioid receptor gene alters β-endorphin binding and activity: Possible implications for opioid addiction. Proc. Nat. Acad. Sci., USA 95: 9608 – 9613 (1998). PMCID: PMC21386
2. McClintick JN, Tischfield JA, Deng L, Kapoor M, Xuei X, Edenberg HJ. Ethanol Activates Immune Response In Lymphoblastoid Cells. Alcohol. 2019 Jan 9;79:81-91. [Epub ahead of print] PMCID: PMC6616005
3. Oni EN, Halikere A, Li G, Toro-Ramos AJ, Swerdel MR, Verpeut JL, Moore JC, Bello NT, Bierut LJ, Goate A, Tischfield JA, Pang ZP, Hart RP. Increased nicotine response in iPSC-derived human neurons carrying the CHRNA5 N398 allele. Sci Rep. 6:34341. (2016) PMCID: PMC5048107.
4. Halikere A, Popova D, Scarnati MS, Hamod A, Swerdel MR, Moore JC, Tischfield JA, Hart RP, Pang ZP. Addiction associated N40D mu-opioid receptor variant modulates synaptic function in human neurons. Mol Psychiatry. 2020 Jul;25(7):1406-1419. doi: 10.1038/s41380-019-0507-0. Epub 2019 Sept.3. PMCID: PMC7051890.

V. In recent years I became interested in neurogenetics, neuroscience and behavior. Specifically, this interest has manifested in studies of Tourette Disorder (TD). Realizing that there were few human samples from TD individuals or families, I and two colleagues started the **T**ourette **I**nternational **C**ollaborative Genetics Project (TIC Genetics), which now encompasses over 15 centers in 7 countries. Over the past 4 years we have collected and banked ~1800 subjects whose genetics we have studied through CNV analysis, whole exome sequencing and other methods. We have described several rare gene variants causing TD and contributed to an understanding of the genetic architecture of the disorder.

1. Ercan-Sencicek AG, Stillman AA, Ghosh AK, Bilguvar K, O'Roak BJ, Mason CE, Abbott T, Gupta A, King RA, Pauls DL, Tischfield JA, Heiman GA, Singer HS, Gilbert DL, Hoekstra PJ, Morgan TM, Loring E, Yasuno K, Fernandez T, Sanders S, Louvi A, Cho JH, Mane S, Colangelo CM, Biederer T, Lifton RP, Gunel M, State MW. L-histidine decarboxylase and Tourette's syndrome. N Engl J Med. 2010 May 20;362(20):1901-8. Epub 2010 May 5. PMCID: PMC2894694.

2. Sun N, Nasello C, Deng L, Wang N, Zhang Y, Xu Z, Song Z, Kwan K, King RA, Pang ZP, Xing J, Heiman GA, Tischfield JA. The PNKD gene is associated with Tourette Disorder or Tic disorder in a multiplex family. Mol Psychiatry. 2017 Sep 12.doi:10.1038/mp.2017.179.[Epub ahead of print] PMCID: PMC5847395.
3. Willsey J, Fernandez TV, Yu D, King RA, Dietrich A, Xing J, Sanders SJ, Mandell JD, Huang AY, Richer P, Smith L, Dong S, Samocha KE, Tourette International Collaborative Genetics (TIC Genetics),Tourette Syndrome Association International Consortium for Genetics (TSAICG), Neale, BM, Coppola G, Mathews CA, Tischfield JA, Scharf JM, State MW, Heiman GA. De Novo Coding Variants Are Strongly Associated with Tourette Disorder. Neuron. 2017 May 3;94(3):486-499.e9. PMCID: PMC5769876
4. Wang S, Mandell JD, Kumar Y, Sun N, Morris MT, Arbelaez J, Nasello C, Dong S, Duhn C, Zhao X, Yang Z, Padmanabhuni SS, Yu D, King RA, Dietrich A, Khalifa N, Dahl N, Huang AY, Neale BM, Coppola G, Mathews CA, Scharf JM; Tourette International Collaborative Genetics Study (TIC Genetics); Tourette Syndrome Genetics Southern and Eastern Europe Initiative (TSGENESEE); Tourette Association of America International Consortium for Genetics (TAAICG), Fernandez TV, Buxbaum JD, De Rubeis S, Grice DE, Xing J, Heiman GA, Tischfield JA, Paschou P, Willsey AJ, State MW. De Novo Sequence and Copy Number Variants Are Strongly Associated with Tourette Disorder and Implicate Cell Polarity in Pathogenesis. Cell Rep. 2018 Sept. 25;24(13):3441-3454.e12. doi: 10.1016/j.celrep.2018.08.082. PMCID: PMC6475626.

Complete List of Published Work in MyBibliography:
http://www.ncbi.nlm.nih.gov/sites/myncbi/jay.tischfield.1/bibliography/41163396/public/?sort=date&direction=descending