BIOGRAPHICAL SKETCH

Provide the following information for the Senior/key personnel and other significant contributors. Follow this format for each person. DO NOT EXCEED FIVE PAGES.

NAME: Fisher, Simon E

POSITION TITLE: Director, Language & Genetics Department, Max Planck Institute for Psycholinguistics

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
Trinity Hall, Cambridge University, UK	B.A. Hons.	07/1991	Natural Sciences
Trinity Hall, Cambridge University, UK	M.A.	07/1995	Natural Sciences
St. Catherine's College, Oxford University, UK	D.Phil.	01/1996	Genetics

A. Personal Statement

I am a director of the Max Planck Institute for Psycholinguistics and Professor of Language and Genetics at the Donders Institute for Brain, Cognition and Behaviour, in Nijmegen, the Netherlands. I obtained a Natural Sciences degree at Cambridge University, UK, followed by a doctorate in human genetics at Oxford University, UK. For my postdoctoral research at the Wellcome Trust Centre for Human Genetics (WTCHG) in Oxford, I focused on genetic analyses of human neurodevelopmental disorders, and isolated the first gene implicated in speech and language deficits. In 2002, I was awarded a Royal Society University Research Fellowship and became head of my own WTCHG laboratory, investigating how language-related genes influence the brain. In 2010 I was appointed director of the Nijmegen MPI, leading a new department devoted to tracing functional links between genetics and language. My work involves extensive supervision of post-doctoral scientists, research assistants and students, and interdisciplinary collaborations worldwide. I am author of 15 book chapters, and >260 journal articles, with original research in Nature, Science, Cell, New England Journal of Medicine, Current Biology, PNAS, Nature Communications, Nature Neuroscience, Nature Genetics, Nature Human Behaviour, and reviews in Annual Review of Genetics/Neuroscience, Nature Reviews Genetics/Neuroscience, Trends in Genetics/Cognitive Sciences and Current Opinion in Genetics & Development/Neurobiology, among others. According to Google Scholar, my publications have received >37,000 citations (h-index of 96). I have given >160 invited talks at colloquia and international conferences, organized expert meetings at the Royal Society (UK) and KNAW (NL), and established a unique Cold Spring Harbor Lab course on Genetics & Neurobiology of Language. I am co-founder/leader of major consortia: GenLang (an international network to facilitate large-scale genomic investigations of speech, language, reading, and related skills) and MusicGens (promotes worldwide research on musicality genetics, genomics, and phenomics). Awards include the Francis Crick Medal and Lecture (2008) and the Eric Kandel Young Neuroscientists Prize (2009). My research integrates data from genetics, neuroscience, developmental biology and evolutionary anthropology.

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

Dutch Research Council (Nederlandse Organisatie voor Wetenschappelijk Onderzoek; NWO)

07/01/2013 - 06/30/2023

Gravitation award 024.001.006: Language in Interaction

Role: Work Package Leader and Scientific Board Member; Peter Hagoort (Main Applicant)

NHMRC (Australia): APP2015727

2023-2028

Translational Centre for Speech Disorders

Role: Chief investigator; Angela Morgan (lead CI)

NIH (US): 1R01DC016977-01A1

2019-2024

Neurobiological Markers of Rhythm: Risk and Resilience for Language Acquisition

Role: Co-investigator; Reyna Gordon (PI)

NIH (US): 1R01MH134004-02

2023-2028

Global studies into the Genetic Architecture of the Brain's White Matter Network through Harmonized and Coordinated

Analyses in the ENIGMA-Consortium

Role: Co-investigator; Neda Jahanshad (PI)

B. Positions and Honors

Positions and Employment

2012-present	Professor of Language and Genetics; Donders Institute for Brain, Cognition & Behaviour, Faculty of Science, Radboud University Nijmegen, Netherlands
2010-present	Director; Max Planck Institute for Psycholinguistics, Nijmegen, Netherlands
2010-2012	Honorary Research Fellow; WTCHG, Oxford University, UK
2002-2010	Head of Molecular Neuroscience research group; WTCHG, Oxford University, UK; Research Area:
	Genetic dissection of neural pathways involved in speech & language acquisition.
1996-2002	Senior post-doctoral research scientist; laboratory of Anthony Monaco MD PhD, WTCHG, Oxford
	University, UK; Research Area: Genetics of language and reading disorders.
1996 Feb-Oct	Post-doctoral research scientist; laboratory of Adrian Hill MD PhD, WTCHG, Oxford University, UK;
	Research Area: Identification of genetic loci conferring susceptibility to leprosy infection in humans.
1991-1996	Doctoral research; Supervisor: Ian Craig PhD, Genetics Lab, Biochemistry Dept, Oxford University, UK;
	Research Area: Positional cloning of human disease genes of the X chromosome.
1991	Undergraduate research; Supervisor: Michael Akam PhD, Genetics Dept, Cambridge University, UK.
	Research Area: Mutations of ultrabithorax gene in Drosophila melanogaster.
1990	Summer research assistant; Supervisor: Jeff Williams PhD, Clare Hall Laboratories, Imperial Cancer
	Research Fund, UK.

Other Experience and Professional Memberships

2019-present	Senior Editor of the journal <i>Neurobiology of Language</i>
2018	External Evaluation Committee for Neuroscience Department, Pasteur Institute, Paris, France
2017-present	Member of UCSD/Salk Center for Academic Research and Training in Anthropogeny (CARTA)
2015-2017	Reviewing Editor of <i>Brain and Language</i>
2014-2023	Scientific Advisory Board (Chair since 2016) of the Netherlands Institute for Neuroscience
2013-2017	External Advisory Committee for <i>University of Connecticut Interdisciplinary Training Program</i> on
	Language Plasticity: Genes, Brain, Cognition and Computation
2013	Scientific Committee for IMFAR: International Meeting For Autism Research
2011-present	Review Editor of Frontiers in Neurogenomics
2011-present	Section Editor of the European Journal of Human Genetics
2010-present	Associate Editor of the journal Frontiers in Language Sciences
2010-present	Scientific Member, Max Planck Society
2009-2010	Neuroscience Management Board (Co-leader: Genes & Dev Neurosci), Oxford University, UK
2009-2011	Royal Society Research Grants - Board F (dev. biol./genetics/immunology/microbiology)
2007-present	Member of the American Society of Human Genetics and the Society for Neuroscience

Honors

2018	Selected to deliver Norman Geschwind Memorial Lecture, International Dyslexia Association
2012	Special Presidential Lecturer at the Society for Neuroscience Annual Meeting, New Orleans
2011-present	Flected Member International Neuropsychological Symposium

2009	Awarded inaugural Eric Kandel Young Neuroscientists Prize by the Hertie Foundation, Germany
2009-present	Elected Fellow, Royal Society of Biology (UK)
2008	Chosen to deliver Nijmegen Lectures, world-leading 3-day linguistics lecture series at the Max Planck
	Institute in Nijmegen, Netherlands.
2008	Awarded Francis Crick Medal and Lecture by the Royal Society, UK
2007-2010	Appointed Isobel Laing Fellow in Biomedical Sciences, Oriel College, Oxford University, UK
2006-2010	Conferred with title of Reader in Molecular Neuroscience, Oxford University, UK
2005	Highly Commended for Young Researcher of the Year at the Times Higher Awards
2005	Merit Increment from the Royal Society, UK
2003-2006	Conferred with title of <i>University Research Lecturer</i> , Oxford University, UK
2002-2010	University Research Fellow, Royal Society, UK
1989-1990	Trinity Hall Scholar, Cambridge University, UK

C. Contributions to Science

- 1. Molecular windows into speech and language disorders. I was co-discoverer of FOXP2, the first gene to be implicated in a developmental speech and language disorder. I went on to carry out extensive functional studies of this gene, a transcription factor regulating expression of downstream targets, in cellular and animal model systems. This body of research has helped shed light on neurobiological pathways that are important for human communication skills, e.g.:
 - a. **Fisher SE**, Vargha-Khadem F, Watkins KE, Monaco AP, Pembrey ME (1998) Localisation of a gene implicated in a severe speech and language disorder. *Nature Genetics* 18: 168-70.
 - b. Lai CSL*, <u>Fisher SE</u>*, Hurst JA, Vargha-Khadem F, Monaco AP (2001) A forkhead-domain gene is mutated in a severe speech and language disorder. *Nature* 413: 519-23. *joint first authors
 - c. MacDermot KD, Bonora E, Sykes N, Coupe AM, Lai CSL Vernes SC, Vargha-Khadem F, McKenzie F, Smith RL, Monaco AP, <u>Fisher SE</u> (2005) Identification of FOXP2 truncation as a novel cause of developmental speech and language deficits. *American Journal of Human Genetics* 76:1074-80.
 - d. Groszer M, Keays DA, Deacon RM, de Bono JP, Prasad-Mulcare S, Gaub S, Baum MG, French CA, Nicod J, Coventry JA, Enard W, Fray M, Brown SD, Nolan PM, Pääbo S, Channon KM, Costa RM, Eilers J, Ehret G, Rawlins JN, <u>Fisher SE</u> (2008) Impaired synaptic plasticity and motor learning in mice with a point mutation implicated in human speech deficits. *Current Biology* 18: 354-62.
 - e. Vernes SC, Newbury DF, Abrahams BS, Winchester L, Nicod J, Groszer M, Alarcón M, Oliver PL, Davies KE, Geschwind DH, Monaco AP, <u>Fisher SE</u> (2008) A functional genetic link between distinct developmental language disorders. *New England Journal of Medicine* 359: 2337-45.
- 2. Evolution of human traits. I have applied innovative approaches, based on molecular genetic data, for gaining insights into the evolutionary origins of distinctive features of human biology, most recently including the first studies to integrate neuroimaging genomics with paleoarcheological data, e.g.:
 - a. Enard W, Przeworski M, <u>Fisher SE</u>, Lai CSL, Wiebe V, Kitano T, Monaco AP, Pääbo S (2002) Molecular evolution of FOXP2, a gene involved in speech and language. *Nature* 418: 869-72.
 - b. <u>Fisher SE</u>, Marcus GF (2006) The eloquent ape: genes, brains and the evolution of language. *Nature Reviews Genetics* 7: 9-20 [refereed review article].
 - c. Enard W, Gehre S, Hammerschmidt K, Hölter SM, Blass T, Somel M, Brückner MK, Schreiweis C, Winter C, Sohr R, Becker L, Wiebe V, Nickel B, Giger T, Müller U, Groszer M, Adler T, Aguilar A, Bolle I, Calzada-Wack J, Dalke C, Ehrhardt N, Favor J, Fuchs H, Gailus-Durner V, Hans W, Hölzlwimmer G, Javaheri A, Kalaydjiev S, Kallnik M, Kling E, Kunder S, Mossbrugger I, Naton B, Racz I, Rathkolb B, Rozman J, Schrewe A, Busch DH, Graw J, Ivandic B, Klingenspor M, Klopstock T, Ollert M, Quintanilla-Martinez L, Schulz H, Wolf E, Wurst W, Zimmer A, Fisher SE, Morgenstern R, Arendt T, de Angelis MH, Fischer J, Schwarz J, Pääbo S (2009) A humanized version of Foxp2 affects cortico-basal ganglia circuits in mice. *Cell* 137: 961-71.

- d. Gunz P, Tilot AK, Wittfeld K, Teumer A, Shapland CY, van Erp TGM, Dannemann M, Vernot B, Neubauer S, Guadalupe T, Fernández G, Brunner HG, Enard W, Fallon J, Hosten N, Völker U, Profico A, Di Vincenzo F, Manzi G, Kelso J, St Pourcain B, Hublin JJ, Franke B, Pääbo S, Macciardi F, Grabe HJ, Fisher SE (2019) Neandertal introgression sheds light on modern human endocranial globularity. *Current Biology* 29: 120-7.
- e. Alagöz G, Molz B, Eising E, Schijven D, Francks C, Stein JL, <u>Fisher SE</u> (2022) Using neuroimaging genomics to investigate the evolution of human brain structure. *Proc Natl Acad Sci USA* 119: e2200638119.
- **3.** Monogenic syndromes that disrupt speech and language development. Using next-generation DNA-sequencing coupled to functional assays in cellular systems, my team has identified and characterized multiple novel neurodevelopmental disorders, e.g.:
 - a. O'Roak BJ, Deriziotis P, Lee C, Vives L, Schwartz JJ, Girirajan S, Karakoc E, MacKenzie AP, Ng S, Baker C, Rieder M, Nickerson D, Bernier R, <u>Fisher SE</u>, Shendure J, Eichler EE (2011) Exome sequencing in sporadic autism spectrum disorders identifies severe de novo mutations. *Nature Genetics* 43: 585-9.
 - b. Deriziotis P, O'Roak BJ, Graham SA, Estruch SB, Dimitropoulou D, Bernier RA, Gerdts J, Shendure JA, Eichler EE, <u>Fisher SE</u> (2014) De novo TBR1 mutations in sporadic autism disrupt protein functions. *Nature Communications* 5: 4954.
 - c. Eising E, Carrion-Castillo A, Vino A, Strand EA, Jakielski KJ, Scerri TS, Hildebrand MS, Webster R, Ma A, Mazoyer B, Francks C, Bahlo M, Scheffer IE, Morgan AT, Shriberg LD, <u>Fisher SE</u> (2019) A set of regulatory genes co-expressed in embryonic human brain is implicated in disrupted speech development. *Molecular Psychiatry* 24: 1065-78.
 - d. Snijders Blok L, Rousseau J, Twist J, Ehresmann S, Takaku M, Venselaar H, Rodan LH, Nowak CB, Douglas J, Swoboda KJ, Steeves MA, Sahai I, Stumpel CTRM, Stegmann APA, Wheeler P, Willing M, Fiala E, Kochhar A, Gibson WT, Cohen ASA, Agbahovbe R, Innes AM, Au PYB, Rankin J, Anderson IJ, Skinner SA, Louie RJ, Warren HE, Afenjar A, Keren B, Nava C, Buratti J, Isapof A, Rodriguez D, Lewandowski R, Propst J, van Essen T, Choi M, Lee S, Chae JH, Price S, Schnur RE, Douglas G, Wentzensen IM, Zweier C, Reis A, Bialer MG, Moore C, Koopmans M, Brilstra EH, Monroe GR, van Gassen KLI, van Binsbergen E, Newbury-Ecob R, Bownass L, Bader I, Mayr JA, Wortmann SB, Jakielski KJ, Strand EA, Kloth K, Bierhals T; DDD study, Roberts JD, Petrovich RM, Machida S, Kurumizaka H, Lelieveld S, Pfundt R, Jansen S, Deriziotis P, Faivre L, Thevenon J, Assoum M, Shriberg L, Kleefstra T, Brunner HG, Wade PA, Fisher SE*, Campeau PM* (2018) CHD3 helicase domain mutations cause a neurodevelopmental syndrome with macrocephaly and impaired speech and language. Nature Communications 9: 4619. *joint senior authors
 - e. Snijders Blok L, Kleefstra T, Venselaar H, Maas S, Kroes HY, Lachmeijer AMA, van Gassen KLI, Firth HV, Tomkins S, Bodek S; DDD Study, Õunap K, Wojcik MH, Cunniff C, Bergstrom K, Powis Z, Tang S, Shinde DN, Au C, Iglesias AD, Izumi K, Leonard J, Abou Tayoun A, Baker SW, Tartaglia M, Niceta M, Dentici ML, Okamoto N, Miyake N, Matsumoto N, Vitobello A, Faivre L, Philippe C, Gilissen C, Wiel L, Pfundt R, Deriziotis P, Brunner HG, <u>Fisher SE</u> (2019) De novo variants disturbing the transactivation capacity of POU3F3 cause a characteristic neurodevelopmental disorder. *American Journal of Human Genetics* 105: 403-12. PMID: 31303265
- **4.** Mapping genes involved in complex developmental language and reading traits. I led the first genome-wide linkage screens in dyslexia, the first whole exome/genome sequencing of developmental speech and language disorders, and the first large-scale well-powered genome-wide association scans of language-/reading-related traits, e.g.:
 - a. <u>Fisher SE</u>, Francks C, Marlow AJ, MacPhie IL, Newbury DF, Cardon LR, Ishikawa-Brush Y, Richardson AJ, Talcott JB, Gayán J, Olson RK, Pennington BF, Smith SD, DeFries JC, Stein JF, Monaco AP (2002) Independent genome-wide scans identify a chromosome 18 quantitative-trait locus influencing dyslexia. *Nature Genetics* 30: 86-91.
 - b. <u>Fisher SE</u>, DeFries JC (2002) Developmental dyslexia: genetic dissection of a complex cognitive trait. *Nature Reviews Neuroscience* 3: 767-80 [refereed review article]
 - c. Villanueva P, Nudel R, Hoischen A, Fernández MA, Simpson NH, Gilissen C, Reader RH, Jara L, Echeverry MM, Francks C, Baird G, Conti-Ramsden G, O'Hare A, Bolton PF, Hennessy ER, the SLI Consortium, Palomino H, Carvajal-Carmona L, Veltman JA, Cazier J-B, De Barbieri Z, <u>Fisher SE</u>*, Newbury DF* (2015) Exome sequencing in an admixed isolated population indicates NFXL1 variants confer a risk for Specific Language Impairment. *PLoS Genetics* 11: e1004925. *joint senior authors

- d. Doust C, Fontanillas P, Eising E, Gordon SD, Wang Z, Alagöz G, Molz B; 23andMe Research Team; Quantitative Trait Working Group of the GenLang Consortium; Pourcain BS, Francks C, Marioni RE, Zhao J, Paracchini S, Talcott JB, Monaco AP, Stein JF, Gruen JR, Olson RK, Willcutt EG, DeFries JC, Pennington BF, Smith SD, Wright MJ, Martin NG, Auton A, Bates TC, <u>Fisher SE</u>, Luciano M (2022) Discovery of 42 genome-wide significant loci associated with dyslexia. *Nature Genetics* 54: 1621-29.
- e. Eising E, Mirza-Schreiber N, de Zeeuw EL, Wang CA, Truong DT, Allegrini AG, Shapland CY, Zhu G, Wigg KG, Gerritse ML, Molz B, Alagöz G, Gialluisi A, Abbondanza F, Rimfeld K, van Donkelaar M, Liao Z, Jansen PR, Andlauer TFM, Bates TC, Bernard M, Blokland K, Bonte M, Børglum AD, Bourgeron T, Brandeis D, Ceroni F, Csépe V, Dale PS, de Jong PF, ..., St Pourcain B, Francks C, <u>Fisher SE</u> (2022) Genome-wide analyses of individual differences in quantitatively assessed reading- and language-related skills in up to 34,000 people. *Proc Natl Acad Sci USA* 119: e2202764119.
- 5. Molecular genetics of synaesthesia and of musicality. My team is the international leader in identifying genetic factors that influence synaesthesia, a rare developmental condition involving blurring of the senses. I was involved in the first study to report elevated prevalence of this trait in autism. As well as identifying rare gene variants in multigenerational families with synaesthesia, my group has studied common polymorphisms in a unique collection of hundreds of unrelated people with the condition, the largest synaesthesia research cohort in the world. Most recently, I have co-founded a new international consortium for studying the genomics of musicality traits.
 - a. Gregersen PK, Kowalsky E, Lee A, Baron-Cohen S, <u>Fisher SE</u>, Asher JE, Ballard D, Freudenberg J, Li W (2013) Absolute pitch exhibits phenotypic and genetic overlap with synesthesia. *Human Molecular Genetics* 22: 2097-104.
 - b. Baron-Cohen S, Johnson D, Asher J, Wheelwright S, <u>Fisher SE</u>, Gregersen PK, Allison C (2013) Is synaesthesia more common in autism? *Molecular Autism* 4: 40.
 - c. Tilot AK, Kucera KS, Vino A, Asher JE, Baron-Cohen S, <u>Fisher SE</u> (2018) Rare variants in axonogenesis genes connect three families with sound-color synesthesia. *Proc Natl Acad Sci U S A* 115: 3168-73.
 - d. Tilot AK, Vino A, Kucera KS, Carmichael DA, van den Heuvel L, den Hoed J, Sidoroff-Dorso AV, Campbell A, Porteous DJ, St Pourcain B, van Leeuwen TM, Ward J, Rouw R, Simner J, <u>Fisher SE</u> (2019) Investigating genetic links between grapheme-colour synaesthesia and neuropsychiatric traits. *Philos Trans R Soc Lond B Biol Sci* 374: 20190026.
 - e. Wesseldijk LW, Henechowicz TL, Baker DJ, Bignardi G, Karlsson R, Gordon RL, Mosing MA, Ullén F, <u>Fisher SE</u> (2024) Notes from Beethoven's genome. *Current Biology* 34: R233-R234

Complete List of Published Work: https://www.mpi.nl/people/fisher-simon-e/publications