

## CURRICULUM VITAE

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**Bryan J. Traynor, MB, BCh, BAO, MMSc, MD, PhD, FANA, FRCPI, FRCP**

### DEMOGRAPHIC AND PERSONAL INFORMATION

#### Current Appointments

2009 – present	Chief, Neuromuscular Diseases Research Section, Laboratory of Neurogenetics, National Institute on Aging (NIA), National Institutes of Health (NIH), Bethesda, MD, USA
2014 – present	Senior Investigator with Tenure, NIA, NIH, Bethesda, MD, USA
2020 – present	Adjunct Professor, Department of Neurology, Johns Hopkins University School of Medicine, Baltimore, MD, USA
2020 – present	Senior Honorary Research Fellow, Queen Square Institute of Neurology, University College London, UK
2020 – present	Adjunct faculty, National Institute of Neurological Disorders and Stroke (NINDS), NIH, Bethesda, MD, USA
2021 – present	Adjunct faculty, National Center for Translational Sciences (NCATS), NIH, Rockville, MD, USA

#### Personal Data

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#### Education

##### Doctoral/Graduate

06/1993	Medical degree (M.B., B.Ch., B.A.O.), University College Dublin, Ireland; Graduated with honors and ranked 2nd in class
06/1996	Membership of the Royal College of Physicians of Ireland (MRCPI), Ireland
06/2000	Medical Doctorate (M.D.), University College Dublin, Ireland; Doctorate based on neuroepidemiology and neurogenetics
06/2004	Masters of Medical Science (M.M.Sc.), Harvard-MIT Health Sciences and Technology, MA; Drug discovery and clinical trial design
06/2012	Doctor of Philosophy (Ph.D.), University College Dublin, Ireland; Doctorate based on neurogenetics

#### Professional Experience

07/1993 – 07/1994	Intern, Internal Medicine, St Vincent's University Hospital, Dublin, Ireland
07/1994 – 07/1995	Senior House Officer, Internal Medicine, Wexford General Hospital, Ireland
07/1995 – 01/1996	Resident, Internal Medicine, Boston City Hospital and Boston University Medical Center, Boston, MA, USA
01/1996 – 07/1996	Senior House Officer, Neurology, St Vincent's University Hospital, Dublin, Ireland
07/1996 – 01/1996	Senior House Office, Endocrinology, St. Vincent's University Hospital, Dublin, Ireland
01/1996 – 07/1997	Senior House Officer, Neurology, National Neuroscience Center, Beaumont Hospital, Dublin, Ireland
07/1997 – 10/1999	Lecturer, Neurology, Royal College of Surgeons of Ireland, Dublin, Ireland
07/1997 – 10/1999	Research Registrar, Neurology, National Neuroscience Center, Beaumont Hospital, Dublin, Ireland

10/1999 – 10/2002	Resident, Neurology, Massachusetts General Hospital and Brigham and Women's Hospital, Boston
10/1999 – 10/2002	Clinical Fellow, Neurology, Harvard Medical School, Boston, MA, USA
10/2002 – 07/2004	Research Fellow, Neuromuscular diseases, Massachusetts General Hospital, Boston, MA, USA
10/2002 – 07/2004	Research Fellow, Neuromuscular diseases, Harvard Medical School, Boston, MA, USA
07/2004 – 07/2005	Staff Neurologist, Neurology Department, Massachusetts General Hospital, Boston, MA, USA
07/2004 – 07/2005	Instructor, Neurology, Harvard Medical School, Boston, MA, USA
07/2005 – 07/2007	Clinical Associate, Neuroscience, National Institute of Mental Health, Bethesda, USA
01/2006 – 01/2009	Staff Neurologist, Neurology Department, Johns Hopkins Hospital, Baltimore, MD, USA
07/2007 – 07/2009	Clinical Associate, Neuroscience, National Institute of Neurological Disorders and Stroke, Bethesda, MD, USA
07/2009 – 06/2011	Assistant Clinical Investigator, National Institute on Aging, Bethesda, MD, USA
06/2011 – 02/2014	Investigator, National Institute on Aging, Bethesda, MD, USA
01/2010 – 07/2020	Adjunct Assistant Professor, Department of Neurology, Johns Hopkins University, Baltimore, MD, USA
07/2020 – present	Adjunct Professor, Department of Neurology, Johns Hopkins University, Baltimore, MD, USA
07/2009 – present	Chief, Neuromuscular Diseases Research Section, National Institute on Aging, Bethesda, MD, USA
02/2014 – present	Senior Investigator with Tenure, National Institute on Aging, Bethesda, MD, USA
09/2020 – present	Senior Honorary Research Fellow, Queen Square Institute of Neurology, University College London
11/2020 – present	Adjunct faculty, National Institute of Neurological Disorders and Stroke, Bethesda, MD

## PUBLICATIONS

### Metrics

	All	Since 2016
Publications	217	52
Citations	33,725	19,916
h-index	82	63
i10-index	175	156

Source: <https://scholar.google.com/citations?user=-t7Y3hAAAAAJ&hl=en>

### Selected publications

1. Incidence and prevalence of ALS in Ireland, 1995-1997: a population-based study. **Traynor BJ**, Codd MB, Corr B, Forde C, Frost E, Hardiman O. *Neurology*. 1999;52:504-9. **Citations: 346**
2. Amyotrophic lateral sclerosis mimic syndromes: a population-based study. **Traynor BJ**, Codd MB, Corr B, Forde C, Frost E, Hardiman O. *Arch Neurol*. 2000;57:109-13. **Citations: 255**
3. Clinical features of amyotrophic lateral sclerosis according to the El Escorial and Airlie House diagnostic criteria: A population-based study. **Traynor BJ**, Codd MB, Corr B, Forde C, Frost E, Hardiman OM. *Arch Neurol*. 2000;57:1171-6. **Citations: 452**
4. Ethnic variation in the incidence of ALS: a systematic review. Cronin S, Hardiman O, **Traynor BJ**. *Neurology*. 2007;68:1002-7. **Citations: 318**
5. Genome-wide genotyping in amyotrophic lateral sclerosis and neurologically normal controls: first stage analysis and public release of data. Schymick JC, Scholz SW, Fung HC, Britton A, Arepalli S, Gibbs JR, Lombardo F, Matarin M, Kasperaviciute D, Hernandez DG, Crews C, Bruijn L, Rothstein J, Mora G, Restagno G, Chiò A, Singleton A, Hardy J, **Traynor BJ**. *Lancet Neurol*. 2007;6:322-8. **Citations: 249**
6. Exome sequencing reveals VCP mutations as a cause of familial ALS. Johnson JO, Mandrioli J, Benatar M, Abramzon Y, Van Deerlin VM, Trojanowski JQ, Gibbs JR, Brunetti M, Gronka S, Wu J, Ding J, McCluskey L, Martinez-Lage M, Falcone D, Hernandez DG, Arepalli S, Chong S, Schymick JC, Rothstein J, Landi F, Wang YD, Calvo A, Mora G, Sabatelli M, Monsurro MR, Battistini S, Salvi F, Spataro R, Sola P, Borghero G, Galassi G, Scholz SW, Taylor JP, Restagno G, Chiò A, **Traynor BJ**. *Neuron*. 2010;68:857-64. PMC3032425. **Citations: 1,200**

7. Chromosome 9p21 in amyotrophic lateral sclerosis in Finland: a genome-wide association study. Laaksovirta H, Peuralinna T, Schymick JC, Scholz SW, Lai SL, Myllykangas L, Sulkava R, Jansson L, Hernandez DG, Gibbs JR, Nalls MA, Heckerman D, Tienari PJ, **Traynor BJ**. *Lancet Neurol*. 2010;9:978-85. PMC2965392. **Citations: 274**
8. A hexanucleotide repeat expansion in C9ORF72 is the cause of chromosome 9p21-linked ALS-FTD. Renton AE, Majounie E, Waite A, Simon-Sanchez J, Rollinson S, Gibbs JR, Schymick JC, Laaksovirta H, Van Swieten JC, Myllykangas L, Kalimo H, Paetau A, Abramzon Y, Remes AM, Kaganovich A, Scholz SW, Duckworth J, Ding J, Harmer DW, Hernandez DG, Johnson JO, Mok K, Ryten M, Trabzuni D, Guerreiro RJ, Orrell RW, Neal J, Murray A, Pearson J, Jansen IE, Sondervan D, Seelaar H, Blake D, Young K, Halliwell N, Callister JB, Toulson G, Richardson A, Gerhard A, Snowden J, Mann D, Neary D, Nalls MA, Peuralinna T, Jansson L, Isoviiita VM, Kaivorinne AL, Holtta-Vuori M, Ikonen E, Sulkava R, Benatar M, Wu J, Chiò A, Restagno G, Borghero G, Sabatelli M, Heckerman D, Rogaeva E, Zinman L, Rothstein JD, Sendtner M, Drepper C, Eichler EE, Alkan C, Abdullaev Z, Pack SD, Dutra A, Pak E, Hardy J, Singleton A, Williams NM, Heutink P, Pickering-Brown S, Morris HR, Tienari PJ, **Traynor BJ**. *Neuron*. 2011;72:257-68. PMC3200438. **Citations: 3,614**
9. Repeat expansion in C9ORF72 in Alzheimer's disease. Majounie E, Abramzon Y, Renton AE, Perry R, Bassett SS, Pletnikova O, Troncoso JC, Hardy J, Singleton AB, **Traynor BJ**. *N Engl J Med*. 2012;366:283-4. PMC3513272. **Citations: 178**
10. Frequency of the C9orf72 hexanucleotide repeat expansion in patients with amyotrophic lateral sclerosis and frontotemporal dementia: a cross-sectional study. Majounie E, Renton AE, Mok K, Dopfer EG, Waite A, Rollinson S, Chiò A, Restagno G, Nicolaou N, Simon-Sanchez J, Van Swieten JC, Abramzon Y, Johnson JO, Sendtner M, Pamphlett R, Orrell RW, Mead S, Sidle KC, Houlden H, Rohrer JD, Morrison KE, Pall H, Talbot K, Ansorge O, Hernandez DG, Arepalli S, Sabatelli M, Mora G, Corbo M, Giannini F, Calvo A, Englund E, Borghero G, Floris GL, Remes AM, Laaksovirta H, McCluskey L, Trojanowski JQ, Van Deerlin VM, Schellenberg GD, Nalls MA, Drory VE, Lu CS, Yeh TH, Ishiura H, Takahashi Y, Tsuji S, Le Ber I, Brice A, Drepper C, Williams N, Kirby J, Shaw P, Hardy J, Tienari PJ, Heutink P, Morris HR, Pickering-Brown S, **Traynor BJ**. *Lancet Neurol*. 2012;11:323-30. PMC3322422. **Citations: 1,009**
11. Mutations in the Matrin 3 gene cause familial amyotrophic lateral sclerosis. Johnson JO, Piro EP, Boehringer A, Chia R, Feit H, Renton AE, Pliner HA, Abramzon Y, Marangi G, Winborn BJ, Gibbs JR, Nalls MA, Morgan S, Shoai M, Hardy J, Pittman A, Orrell RW, Malaspina A, Sidle KC, Fratta P, Harms MB, Baloh RH, Pestronk A, Weihl CC, Rogaeva E, Zinman L, Drory VE, Borghero G, Mora G, Calvo A, Rothstein JD, Drepper C, Sendtner M, Singleton AB, Taylor JP, Cookson MR, Restagno G, Sabatelli M, Bowser R, Chiò A, **Traynor BJ**. *Nat Neurosci*. 2014;17:664-6. PMC4000579. **Citations: 388**
12. A genome-wide association study of myasthenia gravis. Renton AE, Pliner HA, Provenzano C, Evoli A, Ricciardi R, Nalls MA, Marangi G, Abramzon Y, Arepalli S, Chong S, Hernandez DG, Johnson JO, Bartoccioni E, Scuderi F, Maestri M, Gibbs JR, Errichiello E, Chiò A, Restagno G, Sabatelli M, Macek M, Scholz SW, Corse A, Chaudhry V, Benatar M, Barohn RJ, Mcvey A, Pasnoor M, Dimachkie MM, Rowin J, Kissel J, Freimer M, Kaminski HJ, Sanders DB, Lipscomb B, Massey JM, Chopra M, Howard JF, Jr., Koopman WJ, Nicolle MW, Pascuzzi RM, Pestronk A, Wulf C, Florence J, Blackmore D, Soloway A, Siddiqi Z, Muppidi S, Wolfe G, Richman D, Mezei MM, Jiwa T, Oger J, Drachman DB, **Traynor BJ**. *JAMA Neurol*. 2015;72:396-404. PMC4856525. **Citations: 120**
13. Projected increase in amyotrophic lateral sclerosis from 2015 to 2040. Arthur KC, Calvo A, Price TR, Geiger JT, Chiò A, **Traynor BJ**. *Nat Commun*. 2016;7:12408. PMC4987527. **Citations: 226**
14. Genome-wide Analyses Identify KIF5A as a Novel ALS Gene. Nicolas A, Kenna KP, Renton AE, Ticozzi N, Faghri F, Chia R, Dominov JA, Kenna BJ, Nalls MA, Keagle P, Rivera AM, Rheenen WV, Murphy NA, Vugt JFFaV, Geiger JT, Spek RaVD, Pliner HA, Shankaracharya F, Smith BN, Marangi G, Topp SD, Abramzon Y, Gkazi AS, Eicher JD, Kenna A, Italsgen Consortium, Mora G, Calvo A, Mazzini L, Riva N, Mandrioli J, Caponnetto C, Battistini S, Volanti P, Bella VL, Conforti FL, Borghero G, Messina S, Simone IL, Trojsi F, Salvi F, Logullo FO, D'alfonso S, Corrado L, Capasso M, Ferrucci L, Genomic Translation for Als Care (Gtac) Consortium, Moreno CDaM, Kamalakaran S, Goldstein DB, The Als Sequencing Consortium, Gitler AD, Harris T, Myers RM, Nygc Als Consortium, Phatnani H, Musunuri RL, Evani US, Abhyankar A, Zody MC, Answer Als Foundation, Kaye J, Finkbeiner S, Wyman S, Lenail A, Lima L, Fraenkel E, Svendsen CN, Thompson LM, Eyk JEV, Berry JD, Miller TM, Kolb SJ, Cudkowicz M, Baxi E, Clinical Research in Als and Related Disorders for Therapeutic Development (Create) Consortium, Benatar M, Taylor JP, Rampersaud E, Wu G, Wu J, Slagen Consortium, Lauria G, Verde F, Fogh I, Tiloca C, Comi GP, Sorarù G, Cereda C, French Als Consortium, Corcia P, Laaksovirta H, Myllykangas L, Jansson L, Valori M, Ealing J, Hamdallah H, Rollinson S, Pickering-Brown S, Orrell RW, Sidle KC, Malaspina A, Hardy J, Singleton AB, Johnson JO, Arepalli S, Sapp PC, Mckenna-Yasek D, Polak M, Asress S, Al-Sarraj S, King A, Troakes C, Vance C, Bellerocche JD, Baas F, Asbroek ALT, Muñoz-Blanco JL, Hernandez DG, Ding J, Gibbs JR, Scholz SW, Floeter MK, Campbell RH, Landi F, Bowser R, Pulst SM,

- Ravits JM, Macgowan DJL, Kirby J, Pioro E, Pamphlett R, Broach J, Gerhard G, Duncley TL, Brady CB, Kowall NW, Troncoso JC, Ber IL, Mouzat K, Lumbroso S, Heiman-Patterson TD, Kamel F, Bosch LVD, Baloh RH, Strom TM, Meitinger T, Shatunov A, Eijk KRV, Carvalho MD, Kooyman M, Middelkoop B, Moisse M, McLaughlin RL, Es MaV, Weber M, Boylan KB, Blitterswijk MV, Rademakers R, Morrison KE, Basak AN, Mora JS, Drory VE, Shaw PJ, Turner MR, Talbot K, Hardiman O, Williams KL, Fifita JA, Nicholson GA, Blair IP, Rouleau GA, Esteban-Pérez J, García-Redondo A, Al-Chalabi A, Project Mine ALS Sequencing Consortium, Rogaeva E, Zinman L, Ostrow L, Maragakis NJ, Rothstein JD, Simmons Z, Cooper-Knock J, Brice A, Goutman SA, Feldman EL, Gibson SB, Taroni F, Ratti A, Gellera C, Damme PV, Robberecht W, Fratta P, Sabatelli M, Lunetta C, Ludolph AC, Andersen PM, Weishaupt JH, Camu W, Trojanowski JQ, Deerlin VMV, Robert H. Brown J, Berg LHVD, Veldink JH, Harms MB, Glass JD, Stone DJ (contributed equally), Tienari P (contributed equally), Silani V (contributed equally), Chiò A (contributed equally), Shaw CE (contributed equally), **Traynor BJ** (contributed equally), Landers JE (contributed equally). *Neuron*. 2018;97(6):1268-1283. **Citations: 281**
15. Pathogenic Huntingtin Repeat Expansions in Patients with Frontotemporal Dementia and Amyotrophic Lateral Sclerosis. Dewan R, Chia R, Ding J, Hickman RA, Stein TD, Abramzon Y, Ahmed S, Sabir MS, Portley MK, Tucci A, Ibáñez K, Shankaracharya, Keagle P, Rossi G, Caroppo P, Tagliavini F, Waldo ML, Johansson PM, Nilsson CF, The American Genome Center (TAGC), The FALS Sequencing Consortium, The Genomics England Research Consortium, The International ALS/FTD Genomics Consortium (iAFGC), The International FTD Genetics Consortium (IFGC), The International LBD Genomics Consortium (iLBDGC), The NYGC ALS Consortium, The PROSPECT Consortium, Rowe JB, Benussi L, Binetti G, Ghidoni R, Jabbari E, Viollet C, Glass JD, Singleton AB, Silani V, Ross OA, Ryten M, Torkamani A, Tanaka T, Ferrucci L, Resnick SM, Pickering-Brown S, Brady CB, Kowal N, Hardy JA, Van Deerlin V, Vonsattel JP, Harms MB (contributed equally), Morris HR (contributed equally), Ferrari R (contributed equally), Landers JE (contributed equally), Chiò A (contributed equally), Gibbs JR (contributed equally), Dalgard CL (contributed equally), Scholz SW (contributed equally), **Traynor BJ** (contributed equally). *Neuron*. 2021 Feb; 109(3):448-460.
  16. Genetic analysis of amyotrophic lateral sclerosis identifies contributing pathways and cell types. Saez-Atienzar S, Bandres-Ciga S, Langston RG, Kim JJ, Choi SW, Reynolds RH, the International ALS Genomics Consortium; ITALSGEN; Abramzon Y, Dewan R, Ahmed S, Landers JE, Chia R, Ryten M, Cookson MR, Nalls MA, Chiò A, **Traynor BJ**. *Science Advances*. 2021 Jan; 7(3):eabd9036
  17. Genome sequencing analysis identifies new loci associated with Lewy body dementia and provides insights into the complex genetic architecture. Chia R, Sabir MS, Bandres-Ciga S, Saez S, Reynolds RH, Gustavsson E, Walton RL, Ahmed S, Viollet C, Ding J, Makarios MB, Diez-Fairen M, Portley MK, Shah Z, Abramzon Y, Hernandez DG, Blauwendraat C, Stone DJ, Eicher J, Bras J, Grassano M, Calvo A, Mora G, Canosa A, Floris G, Bohannan RC, Brett F, Gan-Or Z, Geiger JT, Moore A, May P, Krüger R, Goldstein D, Lopez G, Tayebi N, Sidransky E; the Fox Investigation for New Discovery of Biomarkers; The American Genome Center; Norcliffe-Kaufmann L, Palma J-A, Kaufmann H, Shakkottai V, Perkins M, Newell KL, Gasser T, Schulte C, Landi F, Salvi E, Cusi D, Masliah E, Kim RC, Caraway CA, Monuki E, Brunetti M, Albert MS, Pletnikova O, Troncoso JC, Flanagan ME, Mao Q, Bigio EH, Rodríguez-Rodríguez E, Infante J, Lage C, González-Aramburu I, Sanchez-Juan P, Porcel-Molina L, Ghetti B, Keith J, Black SE, Masellis M, Rogaeva E, Duyckaerts C, Brice A, Lesage S, Xiromerisiou G, Barrett MJ, Tilley BS, Gentleman S, Logroscino G, Serrano GE, Beach TG, McKeith I, Thomas AJ, Attems J, Morris CM, Palmer L, Love S, Troakes C, Al-Sarraj S, Hodges AK, Aarsland D, Klein G, Kaiser SM, Woltjer R, Pastor P, Bekris LM, Leverenz J, Besser LM, Kuzma A, Renton AE, Goate A, Bennett DA, Scherzer CR, Morris HR, Ferrari R, Pickering-Brown S, Faber K, Kukull W, Lleó A, Fortea J, Alcolea D, Clarimon J, Nalls MA, Ferrucci L, Resnick SM, Tanaka T, Foroud TM, Graff-Radford NR, Wszolek ZK, Ferman T, Boeve BF, Hardy JA, Dickson D, Torkamani A, Singleton AB, Ryten M, Chiò A (contributed equally), Ross OA (contributed equally), Gibbs JR (contributed equally), Dalgard CL (contributed equally), **Traynor BJ** (contributed equally), Scholz SW (contributed equally). *Nature Genetics*. 2021 Mar; 53(3):294-303.
  18. Mutations in the SPTLC1 gene are a cause of juvenile amyotrophic lateral sclerosis. Johnson JO, Chia R, Miller DE, Li R, Kumaran R, Abramzon Y, Alahmady N, Renton AE, Topp SD, Gibbs JR, Cookson MR, Sabir MS, Dalgard CL, Troakes C, Jones AR, Shatunov A, Iacoangeli A, Al Khleifat A, Ticozzi N, Silani V, Gellera C, Blair IP, Dobson-Stone C, Kwok JB, Bonkowski ES, Palvadeau R, Tienari PJ, Morrison KE, Shaw PJ, Al-Chalabi A, Brown RH, Calvo A, Mora G, Al-Saif H, Gotkine M, Leigh F, Chang IJ, Perlman SJ, Glass I, Scott AI, Shaw CE, Basak AN (contributed equally), Landers JE (contributed equally), Chiò A (contributed equally), Crawford TO (contributed equally), Smith BN (contributed equally), and **Traynor BJ** (contributed equally) on behalf of The FALS Sequencing Consortium; The American Genome Center, The International ALS Genomics Consortium; and The ITALSGEN Consortium. *JAMA Neurology*. 2021. ePrint ahead of publication.

## Original Research

1. Service provision for patients with ALS/MND: a cost-effective multidisciplinary approach. Corr B, Frost E, **Traynor BJ**, Hardiman O. *J Neurol Sci*. 1998;160 Suppl 1:S141-5.
2. "True" sporadic ALS associated with a novel SOD-1 mutation. Alexander MD, **Traynor BJ**, Miller N, Corr B, Frost E, McQuaid S, Brett FM, Green A, Hardiman O. *Ann Neurol*. 2002;52:680-3.
3. Access to health services in Ireland for people with Multiple Sclerosis and Motor Neurone Disease. Hardiman O, Corr B, Frost E, Gibbons P, Mahon L, **Traynor BJ**. *Ir Med J*. 2003;96:200-3.
4. Effect of a multidisciplinary amyotrophic lateral sclerosis (ALS) clinic on ALS survival: a population-based study, 1996-2000. **Traynor BJ**, Alexander M, Corr B, Frost E, Hardiman O. *J Neurol Neurosurg Psychiatry*. 2003;74:1258-61. PMC1738639.
5. An outcome study of riluzole in amyotrophic lateral sclerosis--a population-based study in Ireland, 1996-2000. **Traynor BJ**, Alexander M, Corr B, Frost E, Hardiman O. *J Neurol*. 2003;250:473-9.
6. A novel candidate region for ALS on chromosome 14q11.2. Greenway MJ, Alexander MD, Ennis S, **Traynor BJ**, Corr B, Frost E, Green A, Hardiman O. *Neurology*. 2004;63:1936-8.
7. Functional outcome measures as clinical trial endpoints in ALS. **Traynor BJ**, Zhang H, Shefner JM, Schoenfeld D, Cudkowicz ME. *Neurology*. 2004;63:1933-5.
8. Tolerance of high-dose (3,000 mg/day) coenzyme Q10 in ALS. Ferrante KL, Shefner J, Zhang H, Betensky R, O'Brien M, Yu H, Fantasia M, Taft J, Beal MF, **Traynor B**, Newhall K, Donofrio P, Caress J, Ashburn C, Freiberg B, O'Neill C, Paladenech C, Walker T, Pestronk A, Abrams B, Florence J, Renna R, Schierbecker J, Malkus B, Cudkowicz M. *Neurology*. 2005;65:1834-6.
9. Analysis of IFT74 as a candidate gene for chromosome 9p-linked ALS-FTD. Momeni P, Schymick J, Jain S, Cookson MR, Cairns NJ, Greggio E, Greenway MJ, Berger S, Pickering-Brown S, Chiò A, Fung HC, Holtzman DM, Huey ED, Wassermann EM, Adamson J, Hutton ML, Rogaeva E, St George-Hyslop P, Rothstein JD, Hardiman O, Grafman J, Singleton A, Hardy J, **Traynor BJ**. *BMC Neurol*. 2006;6:44. PMC1764752.
10. Neuroprotective agents for clinical trials in ALS: a systematic assessment. **Traynor BJ**, Bruijn L, Conwit R, Beal F, O'Neill G, Fagan SC, Cudkowicz ME. *Neurology*. 2006;67:20-7.
11. Progranulin mutations and amyotrophic lateral sclerosis or amyotrophic lateral sclerosis-frontotemporal dementia phenotypes. Schymick JC, Yang Y, Andersen PM, Vonsattel JP, Greenway M, Momeni P, Elder J, Chiò A, Restagno G, Robberecht W, Dahlberg C, Mukherjee O, Goate A, Graff-Radford N, Caselli RJ, Hutton M, Gass J, Cannon A, Rademakers R, Singleton AB, Hardiman O, Rothstein J, Hardy J, **Traynor BJ**. *J Neurol Neurosurg Psychiatry*. 2007;78:754-6. PMC2117704.
12. Prevalence of SOD1 mutations in the Italian ALS population. Chiò A, **Traynor BJ**, Lombardo F, Fimognari M, Calvo A, Ghiglione P, Mutani R, Restagno G. *Neurology*. 2008;70:533-7.
13. Amyotrophic lateral sclerosis and soccer: a different epidemiological approach strengthens the previous findings. Chiò A, **Traynor BJ**, Swingle R, Mitchell D, Hardiman O, Mora G, Beghi E, Logroscino G. *J Neurol Sci*. 2008;269:187-8; author reply 8-9.
14. A genome-wide association study of sporadic ALS in a homogenous Irish population. Cronin S, Berger S, Ding J, Schymick JC, Washecka N, Hernandez DG, Greenway MJ, Bradley DG, **Traynor BJ**, Hardiman O. *Hum Mol Genet*. 2008;17:768-74.
15. TDP-43 is not a common cause of sporadic amyotrophic lateral sclerosis. Guerreiro RJ, Schymick JC, Crews C, Singleton A, Hardy J, **Traynor BJ**. *PLoS One*. 2008;3:e2450. PMC2408729.
16. Genotype, haplotype and copy-number variation in worldwide human populations. Jakobsson M, Scholz SW, Scheet P, Gibbs JR, VanLiere JM, Fung HC, Szpiech ZA, Degnan JH, Wang K, Guerreiro R, Bras JM, Schymick JC, Hernandez DG, **Traynor BJ**, Simon-Sanchez J, Matarin M, Britton A, Van De Leemput J, Rafferty I, Bucan M, Cann HM, Hardy JA, Rosenberg NA, Singleton AB. *Nature*. 2008;451:998-1003.
17. A common haplotype within the PON1 promoter region is associated with sporadic ALS. Landers JE, Shi L, Cho TJ, Glass JD, Shaw CE, Leigh PN, Diekstra F, Polak M, Rodriguez-Leyva I, Niemann S, **Traynor BJ**, McKenna-Yasek D, Sapp PC, Al-Chalabi A, Wills AM, Brown RH, Jr. *Amyotroph Lateral Scler*. 2008;9:306-14. PMC2739087.
18. Epidemiology and clinical features of amyotrophic lateral sclerosis in Ireland between 1995 and 2004. O'toole O, **Traynor BJ**, Brennan P, Sheehan C, Frost E, Corr B, Hardiman O. *J Neurol Neurosurg Psychiatry*. 2008;79:30-2.
19. Genetic variation in DPP6 is associated with susceptibility to amyotrophic lateral sclerosis. Van Es MA, Van Vught PW, Blauw HM, Franke L, Saris CG, Van Den Bosch L, De Jong SW, De Jong V, Baas F, Van't Slot R, Lemmens R, Schelhaas HJ, Birve A, Slegers K, Van Broeckhoven C, Schymick JC, **Traynor BJ**, Wokke JH, Wijmenga C, Robberecht W, Andersen PM, Veldink JH, Ophoff RA, Van Den Berg LH. *Nat Genet*. 2008;40:29-31.

20. Web-based data management for a phase II clinical trial in ALS. Buchsbaum R, Kaufmann P, Barsdorf AI, Arbing R, Montes J, Thompson JL; QALS Study Group\*. *Amyotroph Lateral Scler.* 2009;10:374-7. NIHMSID940286.
21. Two Italian kindreds with familial amyotrophic lateral sclerosis due to FUS mutation. Chiò A, Restagno G, Brunetti M, Ossola I, Calvo A, Mora G, Sabatelli M, Monsurro MR, Battistini S, Mandrioli J, Salvi F, Spataro R, Schymick J, **Traynor BJ**, La Bella V. *Neurobiol Aging.* 2009;30:1272-5. PMC2771748.
22. A two-stage genome-wide association study of sporadic amyotrophic lateral sclerosis. Chiò A, Schymick JC, Restagno G, Scholz SW, Lombardo F, Lai SL, Mora G, Fung HC, Britton A, Arepalli S, Gibbs JR, Nalls M, Berger S, Kwee LC, Oddone EZ, Ding J, Crews C, Rafferty I, Washecka N, Hernandez D, Ferrucci L, Bandinelli S, Guralnik J, Macciardi F, Torri F, Lupoli S, Chanock SJ, Thomas G, Hunter DJ, Gieger C, Wichmann HE, Calvo A, Mutani R, Battistini S, Giannini F, Caponnetto C, Mancardi GL, La Bella V, Valentino F, Monsurro MR, Tedeschi G, Marinou K, Sabatelli M, Conte A, Mandrioli J, Sola P, Salvi F, Bartolomei I, Siciliano G, Carlesi C, Orrell RW, Talbot K, Simmons Z, Connor J, Piro EP, Dunkley T, Stephan DA, Kasperaviciute D, Fisher EM, Jabonka S, Sendtner M, Beck M, Bruijn L, Rothstein J, Schmidt S, Singleton A, Hardy J, **Traynor BJ**. *Hum Mol Genet.* 2009;18:1524-32. PMC2664150.
23. Phase II trial of CoQ10 for ALS finds insufficient evidence to justify phase III. Kaufmann P, Thompson JL, Levy G, Buchsbaum R, Shefner J, Krivickas LS, Katz J, Rollins Y, Barohn RJ, Jackson CE, Tiryaki E, Lomen-Hoerth C, Armon C, Tandan R, Rudnicki SA, Rezania K, Sufit R, Pestronk A, Novella SP, Heiman-Patterson T, Kasarskis EJ, Piro EP, Montes J, Arbing R, Vecchio D, Barsdorf A, Mitsumoto H, Levin B. *Ann Neurol.* 2009;66:235-44. PMC2854625.
24. Extended tracts of homozygosity identify novel candidate genes associated with late-onset Alzheimer's disease. Nalls MA, Guerreiro RJ, Simon-Sanchez J, Bras JT, **Traynor BJ**, Gibbs JR, Launer L, Hardy J, Singleton AB. *Neurogenetics.* 2009;10:183-90. PMC2908484.
25. Ubiquitin associated protein 1 is a risk factor for frontotemporal lobar degeneration. Rollinson S, Rizzu P, Sikkink S, Baker M, Halliwell N, Snowden J, **Traynor BJ**, Ruano D, Cairns N, Rohrer JD, Mead S, Collinge J, Rossor M, Akay E, Guerreiro R, Rademakers R, Morrison KE, Pastor P, Alonso E, Martinez-Lage P, Graff-Radford N, Neary D, Heutink P, Mann DM, Van Swieten J, Pickering-Brown SM. *Neurobiol Aging.* 2009;30:656-65. PMC2753870.
26. SNCA variants are associated with increased risk for multiple system atrophy. Scholz SW, Houlden H, Schulte C, Sharma M, Li A, Berg D, Melchers A, Paudel R, Gibbs JR, Simon-Sanchez J, Paisan-Ruiz C, Bras J, Ding J, Chen H, **Traynor BJ**, Arepalli S, Zonozi RR, Revesz T, Holton J, Wood N, Lees A, Oertel W, Wullner U, Goldwurm S, Pellecchia MT, Illig T, Riess O, Fernandez HH, Rodriguez RL, Okun MS, Poewe W, Wenning GK, Hardy JA, Singleton AB, Del Sorbo F, Schneider S, Bhatia KP, Gasser T. *Ann Neurol.* 2009;65:610-4. PMC3520128.
27. Genome-wide association reveals three SNPs associated with sporadic amyotrophic lateral sclerosis through a two-locus analysis. Sha Q, Zhang Z, Schymick JC, **Traynor BJ**, Zhang S. *BMC Med Genet.* 2009;10:86. PMC2752455.
28. Genome-wide association study reveals genetic risk underlying Parkinson's disease. Simon-Sanchez J, Schulte C, Bras JM, Sharma M, Gibbs JR, Berg D, Paisan-Ruiz C, Lichtner P, Scholz SW, Hernandez DG, Kruger R, Federoff M, Klein C, Goate A, Perlmutter J, Bonin M, Nalls MA, Illig T, Gieger C, Houlden H, Steffens M, Okun MS, Racette BA, Cookson MR, Foote KD, Fernandez HH, **Traynor BJ**, Schreiber S, Arepalli S, Zonozi R, Gwinn K, Van Der Brug M, Lopez G, Chanock SJ, Schatzkin A, Park Y, Hollenbeck A, Gao J, Huang X, Wood NW, Lorenz D, Deuschl G, Chen H, Riess O, Hardy JA, Singleton AB, Gasser T. *Nat Genet.* 2009;41:1308-12. PMC2787725.
29. Amyotrophic lateral sclerosis, physical exercise, trauma and sports: results of a population-based pilot case-control study. Beghi E, Logroscino G, Chiò A, Hardiman O, Millul A, Mitchell D, Swingler R, **Traynor BJ**. *Amyotroph Lateral Scler.* 2010;11:289-92. PMC3513269.
30. Amyotrophic lateral sclerosis-frontotemporal lobar dementia in 3 families with p.Ala382Thr TARDBP mutations. Chiò A, Calvo A, Moglia C, Restagno G, Ossola I, Brunetti M, Montuschi A, Cistaro A, Ticca A, **Traynor BJ**, Schymick JC, Mutani R, Marrosu MG, Murru MR, Borghero G. *Arch Neurol.* 2010;67:1002-9. PMC3535689.
31. Abundant quantitative trait loci exist for DNA methylation and gene expression in human brain. Gibbs JR, Van Der Brug MP, Hernandez DG, **Traynor BJ**, Nalls MA, Lai SL, Arepalli S, Dillman A, Rafferty IP, Troncoso J, Johnson R, Zielke HR, Ferrucci L, Longo DL, Cookson MR, Singleton AB. *PLoS Genet.* 2010;6:e1000952. PMC2869317.
32. Incidence of amyotrophic lateral sclerosis in Europe. Logroscino G, **Traynor BJ**, Hardiman O, Chiò A, Mitchell D, Swingler RJ, Millul A, Benn E, Beghi E. *J Neurol Neurosurg Psychiatry.* 2010;81:385-90. PMC2850819.
33. Chromosome 9p21 in sporadic amyotrophic lateral sclerosis in the UK and seven other countries: a genome-wide association study. Shatunov A, Mok K, Newhouse S, Weale ME, Smith B, Vance C, Johnson L, Veldink JH, Van Es MA, Van Den Berg LH, Robberecht W, Van Damme P, Hardiman O, Farmer AE, Lewis CM, Butler AW,



- Abel O, Andersen PM, Fogh I, Silani V, Chiò A, **Traynor BJ**, Melki J, Meininger V, Landers JE, Mcguffin P, Glass JD, Pall H, Leigh PN, Hardy J, Brown RH, Jr., Powell JF, Orrell RW, Morrison KE, Shaw PJ, Shaw CE, Al-Chalabi A. *Lancet Neurol.* 2010;9:986-94. PMC3257853.
34. Kinesin-associated protein 3 (KIFAP3) has no effect on survival in a population-based cohort of ALS patients. **Traynor BJ**, Nalls M, Lai SL, Gibbs RJ, Schymick JC, Arepalli S, Hernandez D, Van Der Brug MP, Johnson JO, Dillman A, Cookson M, Moglia C, Calvo A, Restagno G, Mora G, Chiò A. *Proc Natl Acad Sci U S A.* 2010;107:12335-8. PMC2901467. **Citations: 43**
35. Tau levels do not influence human ALS or motor neuron degeneration in the SOD1G93A mouse. Taes I, Goris A, Lemmens R, Van Es MA, Van Den Berg LH, Chiò A, **Traynor BJ**, Birve A, Andersen P, Slowik A, Tomik B, Brown RH, Jr., Shaw CE, Al-Chalabi A, Boonen S, Van Den Bosch L, Dubois B, Van Damme P, Robberecht W. *Neurology.* 2010;74:1687-93. PMC2882212.
36. A two-stage meta-analysis identifies several new loci for Parkinson's disease. **International Parkinson Disease Genomics Consortium\***, WTCCC. *PLoS Genet.* 2011;7:e1002142. PMC3128098.
37. A patient carrying a homozygous p.A382T TARDBP missense mutation shows a syndrome including ALS, extrapyramidal symptoms, and FTD. Borghero G, Floris G, Cannas A, Marrosu MG, Murru MR, Costantino E, Parish LD, Pugliatti M, Ticca A, **Traynor BJ**, Calvo A, Cammarosano S, Moglia C, Cistaro A, Brunetti M, Restagno G, Chiò A. *Neurobiol Aging.* 2011;32:2327.e1-5. PMC3192246.
38. Large proportion of amyotrophic lateral sclerosis cases in Sardinia due to a single founder mutation of the TARDBP gene. Chiò A, Borghero G, Pugliatti M, Ticca A, Calvo A, Moglia C, Mutani R, Brunetti M, Ossola I, Marrosu MG, Murru MR, Floris G, Cannas A, Parish LD, Cossu P, Abramzon Y, Johnson JO, Nalls MA, Arepalli S, Chong S, Hernandez DG, **Traynor BJ**, Restagno G. *Arch Neurol.* 2011;68:594-8. PMC3513278.
39. A de novo missense mutation of the FUS gene in a "true" sporadic ALS case. Chiò A, Calvo A, Moglia C, Ossola I, Brunetti M, Sbaiz L, Lai SL, Abramzon Y, **Traynor BJ**, Restagno G. *Neurobiol Aging.* 2011;32:553.e23-6. PMC2972379.
40. No major progranulin genetic variability contribution to disease etiopathogenesis in an ALS Italian cohort. Del Bo R, Corti S, Santoro D, Ghione I, Fenoglio C, Ghezzi S, Ranieri M, Galimberti D, Mancuso M, Siciliano G, Briani C, Murri L, Scarpini E, Schymick JC, **Traynor BJ**, Bresolin N, Comi GP. *Neurobiol Aging.* 2011;32:1157-8. PMC3511779.
41. Distinct DNA methylation changes highly correlated with chronological age in the human brain. Hernandez DG, Nalls MA, Gibbs JR, Arepalli S, Van Der Brug M, Chong S, Moore M, Longo DL, Cookson MR, **Traynor BJ**, Singleton AB. *Hum Mol Genet.* 2011;20:1164-72. PMC3043665.
42. FUS mutations in sporadic amyotrophic lateral sclerosis. Lai SL, Abramzon Y, Schymick JC, Stephan DA, Dunckley T, Dillman A, Cookson M, Calvo A, Battistini S, Giannini F, Caponnetto C, Mancardi GL, Spataro R, Monsurro MR, Tedeschi G, Marinou K, Sabatelli M, Conte A, Mandrioli J, Sola P, Salvi F, Bartolomei I, Lombardo F, Mora G, Restagno G, Chiò A, **Traynor BJ**. *Neurobiol Aging.* 2011;32:550.e1-4. PMC2891336.
43. Imputation of sequence variants for identification of genetic risks for Parkinson's disease: a meta-analysis of genome-wide association studies. **International Parkinson Disease Genomics Consortium\***; Nalls MA, Plagnol V, Hernandez DG, Sharma M, Sheerin UM, Saad M, Simon-Sanchez J, Schulte C, Lesage S, Sveinbjornsdottir S, Stefansson K, Martinez M, Hardy J, Heutink P, Brice A, Gasser T, Singleton AB, Wood NW. *Lancet.* 2011;377:641-9. PMC3696507.
44. APOE and AbetaPP gene variation in cortical and cerebrovascular amyloid-beta pathology and Alzheimer's disease: a population-based analysis. Peuralinna T, Tanskanen M, Makela M, Polvikoski T, Paetau A, Kalimo H, Sulkava R, Hardy J, Lai SL, Arepalli S, Hernandez D, **Traynor BJ**, Singleton A, Tienari PJ, Myllykangas L. *J Alzheimers Dis.* 2011;26:377-85. PMC3516850.
45. Valosin-containing protein (VCP) mutations in sporadic amyotrophic lateral sclerosis. Abramzon Y, Johnson JO, Scholz SW, Taylor JP, Brunetti M, Calvo A, Mandrioli J, Benatar M, Mora G, Restagno G, Chiò A, **Traynor BJ**. *Neurobiol Aging.* 2012;33:2231.e1-e6. PMC3391327.
46. Polymorphisms in the GluR2 gene are not associated with amyotrophic lateral sclerosis. Bogaert E, Goris A, Van Damme P, Geelen V, Lemmens R, Van Es MA, Van Den Berg LH, Slegers K, Verpoorten N, Timmerman V, De Jonghe P, Van Broeckhoven C, **Traynor BJ**, Landers JE, Brown RH, Jr., Glass JD, Al-Chalabi A, Shaw CE, Birve A, Andersen PM, Slowik A, Tomik B, Melki J, Robberecht W, Van Den Bosch L. *Neurobiol Aging.* 2012;33:418-20. PMC2949683.
47. Clinical characteristics of patients with familial amyotrophic lateral sclerosis carrying the pathogenic GGGGCC hexanucleotide repeat expansion of C9ORF72. Chiò A, Borghero G, Restagno G, Mora G, Drepper C, **Traynor BJ**, Sendtner M, Brunetti M, Ossola I, Calvo A, Pugliatti M, Sotgiu MA, Murru MR, Marrosu MG, Marrosu F, Marinou K, Mandrioli J, Sola P, Caponnetto C, Mancardi G, Mandich P, La Bella V, Spataro R, Conte A, Monsurro MR, Tedeschi G, Pisano F, Bartolomei I, Salvi F, Lauria Pinter G, Simone I, Logroscino G,

- Gambardella A, Quattrone A, Lunetta C, Volanti P, Zollino M, Penco S, Battistini S, Renton AE, Majounie E, Abramzon Y, Conforti FL, Giannini F, Corbo M, Sabatelli M. *Brain*. 2012;135:784-93. PMC3286333.
48. Extensive genetics of ALS: a population-based study in Italy. Chiò A, Calvo A, Mazzini L, Cantello R, Mora G, Moglia C, Corrado L, D'alfonso S, Majounie E, Renton A, Pisano F, Ossola I, Brunetti M, **Traynor BJ**, Restagno G. *Neurology*. 2012;79:1983-9. PMC3484987.
  49. ALS/FTD phenotype in two Sardinian families carrying both C9ORF72 and TARDBP mutations. Chiò A, Restagno G, Brunetti M, Ossola I, Calvo A, Canosa A, Moglia C, Floris G, Tacconi P, Marrosu F, Marrosu MG, Murru MR, Majounie E, Renton AE, Abramzon Y, Pugliatti M, Sotgiu MA, **Traynor BJ**, Borghero G. *J Neurol Neurosurg Psychiatry*. 2012;83:730-3. PMC4568835.
  50. The RNA-binding motif 45 (RBM45) protein accumulates in inclusion bodies in amyotrophic lateral sclerosis (ALS) and frontotemporal lobar degeneration with TDP-43 inclusions (FTLD-TDP) patients. Collins M, Riascos D, Kovalik T, An J, Krupa K, Krupa K, Hood BL, Conrads TP, Renton AE, **Traynor BJ**, Bowser R. *Acta Neuropathol*. 2012;124:717-32. PMC3472056.
  51. Clinico-pathological features in amyotrophic lateral sclerosis with expansions in C9ORF72. Cooper-Knock J, Hewitt C, Highley JR, Brockington A, Milano A, Man S, Martindale J, Hartley J, Walsh T, Gelsthorpe C, Baxter L, Forster G, Fox M, Bury J, Mok K, Mcdermott CJ, **Traynor BJ**, Kirby J, Wharton SB, Ince PG, Hardy J, Shaw PJ. *Brain*. 2012;135:751-64. PMC3286332.
  52. Familial Lund frontotemporal dementia caused by C9ORF72 hexanucleotide expansion. Englund E, Gustafson L, Passant U, Majounie E, Renton AE, **Traynor BJ**, Rohrer JD, Mok K, Hardy J. *Neurobiol Aging*. 2012;33:1850.e13-6. PMC4562220.
  53. Frontotemporal dementia with psychosis, parkinsonism, visuo-spatial dysfunction, upper motor neuron involvement associated to expansion of C9ORF72: a peculiar phenotype? Floris G, Borghero G, Cannas A, Di Stefano F, Costantino E, Murru MR, Brunetti M, Restagno G, **Traynor BJ**, Marrosu MG, Chiò A, Marrosu F. *J Neurol*. 2012;259:1749-51. PMC4164047.
  54. Integration of GWAS SNPs and tissue specific expression profiling reveal discrete eQTLs for human traits in blood and brain. Hernandez DG, Nalls MA, Moore M, Chong S, Dillman A, Trabzuni D, Gibbs JR, Ryten M, Arepalli S, Weale ME, Zonderman AB, Troncoso J, O'brien R, Walker R, Smith C, Bandinelli S, **Traynor BJ**, Hardy J, Singleton AB, Cookson MR. *Neurobiol Dis*. 2012;47:20-8. PMC3358430.
  55. Using genome-wide complex trait analysis to quantify 'missing heritability' in Parkinson's disease. Keller MF, Saad M, Bras J, Bettella F, Nicolaou N, Simon-Sanchez J, Mittag F, Buchel F, Sharma M, Gibbs JR, Schulte C, Moskvina V, Durr A, Holmans P, Kilarski LL, Guerreiro R, Hernandez DG, Brice A, Ylikotila P, Stefansson H, Majamaa K, Morris HR, Williams N, Gasser T, Heutink P, Wood NW, Hardy J, Martinez M, Singleton AB, Nalls MA; **International Parkinson Disease Genomics Consortium\***. *Hum Mol Genet*. 2012;21:4996-5009. PMC3576713.
  56. A candidate gene for autoimmune myasthenia gravis. Landouere G, Knight MA, Stanescu H, Taye AA, Shi Y, Diallo O, Johnson JO, Hernandez D, **Traynor BJ**, Biesecker LG, Elkahlon A, Rinaldi C, Vincent A, Willcox N, Kleta R, Fischbeck KH, Burnett BG. *Neurology*. 2012;79:342-7. PMC3400092.
  57. Exome sequencing identifies a novel TRPV4 mutation in a CMT2C family. Landouere G, Sullivan JM, Johnson JO, Munns CH, Shi Y, Diallo O, Gibbs JR, Gaudet R, Ludlow CL, Fischbeck KH, **Traynor BJ**, Burnett BG, Sumner CJ. *Neurology*. 2012;79:192-4. PMC3390542.
  58. Large C9orf72 repeat expansions are not a common cause of Parkinson's disease. Majounie E, Abramzon Y, Renton AE, Keller MF, **Traynor BJ**, Singleton AB. *Neurobiol Aging*. 2012;33:2527.e1-2. PMC4545506.
  59. Mutational analysis of the VCP gene in Parkinson's disease. Majounie E, **Traynor BJ**, Chiò A, Restagno G, Mandrioli J, Benatar M, Taylor JP, Singleton AB. *Neurobiol Aging*. 2012;33:209.e1-2. PMC3221929.
  60. Use of support vector machines for disease risk prediction in genome-wide association studies: concerns and opportunities. Mittag F, Buchel F, Saad M, Jahn A, Schulte C, Bochdanovits Z, Simon-Sanchez J, Nalls MA, Keller M, Hernandez DG, Gibbs JR, Lesage S, Brice A, Heutink P, Martinez M, Wood NW, Hardy J, Singleton AB, Zell A, Gasser T, Sharma M; **International Parkinson Disease Genomics Consortium\***. *Hum Mutat*. 2012;33:1708-18. NIHMSID572533.
  61. Chromosome 9 ALS and FTD locus is probably derived from a single founder. Mok K, **Traynor BJ**, Schymick J, Tienari PJ, Laaksovirta H, Peuralinna T, Myllykangas L, Chiò A, Shatunov A, Boeve BF, Boxer AL, Dejesus-Hernandez M, Mackenzie IR, Waite A, Williams N, Morris HR, Simon-Sanchez J, Van Swieten JC, Heutink P, Restagno G, Mora G, Morrison KE, Shaw PJ, Rollinson PS, Al-Chalabi A, Rademakers R, Pickering-Brown S, Orrell RW, Nalls MA, Hardy J. *Neurobiol Aging*. 2012;33:209.e3-8. PMC3312749.
  62. C9ORF72 expansion in amyotrophic lateral sclerosis/frontotemporal dementia also causes parkinsonism. O'dowd S, Curtin D, Waite AJ, Roberts K, Pender N, Reid V, O'connell M, Williams NM, Morris HR, **Traynor BJ**, Lynch T. *Mov Disord*. 2012;27:1072-4. PMC3516857.



63. C9ORF72 hexanucleotide repeat expansions in the Italian sporadic ALS population. Sabatelli M, Conforti FL, Zollino M, Mora G, Monsurro MR, Volanti P, Marinou K, Salvi F, Corbo M, Giannini F, Battistini S, Penco S, Lunetta C, Quattrone A, Gambardella A, Logroscino G, Simone I, Bartolomei I, Pisano F, Tedeschi G, Conte A, Spataro R, La Bella V, Caponnetto C, Mancardi G, Mandich P, Sola P, Mandrioli J, Renton AE, Majounie E, Abramzon Y, Marrosu F, Marrosu MG, Murru MR, Sotgiu MA, Pugliatti M, Rodolico C, Moglia C, Calvo A, Ossola I, Brunetti M, **Traynor BJ**, Borghero G, Restagno G, Chiò A. *Neurobiol Aging*. 2012;33:1848.e15-20. PMC3372681.
64. Exome sequencing in an SCA14 family demonstrates its utility in diagnosing heterogeneous diseases. Sailer A, Scholz SW, Gibbs JR, Tucci A, Johnson JO, Wood NW, Plagnol V, Hummerich H, Ding J, Hernandez D, Hardy J, Federoff HJ, **Traynor BJ**, Singleton AB, Houlden H. *Neurology*. 2012;79:127-31. PMC3390538.
65. Age of onset of amyotrophic lateral sclerosis is modulated by a locus on 1p34.1. Ahmeti KB, Ajroud-Driss S, Al-Chalabi A, Andersen PM, Armstrong J, Birve A, Blauw HM, Brown RH, Bruijn L, Chen W, Chiò A, Comeau MC, Cronin S, Diekstra FP, Soraya Gkazi A, Glass JD, Grab JD, Groen EJ, Haines JL, Hardiman O, Heller S, Huang J, Hung WY, Jaworski JM, Jones A, Khan H, Landers JE, Langefeld CD, Leigh PN, Marion MC, McLaughlin RL, Meininger V, Melki J, Miller JW, Mora G, Pericak-Vance MA, Rampersaud E, Robberecht W, Russell LP, Salachas F, Saris CG, Shatunov A, Shaw CE, Siddique N, Siddique T, Smith BN, Sufit R, Topp S, **Traynor BJ**, Vance C, Van Damme P, Van Den Berg LH, Van Es MA, Van Vught PW, Veldink JH, Yang Y, Zheng JG. *Neurobiol Aging*. 2013;34:357.e7-19. PMC3839234.
66. Genome-wide meta-analysis identifies new susceptibility loci for migraine. Anttila V, Winsvold BS, Gormley P, Kurth T, Bettella F, McMahon G, Kallela M, Malik R, De Vries B, Terwindt G, Medland SE, Todt U, McArdle WL, Quaye L, Koiranen M, Ikram MA, Lehtimäki T, Stam AH, Ligthart L, Wedenoja J, Dunham I, Neale BM, Palta P, Hamalainen E, Schurks M, Rose LM, Buring JE, Ridker PM, Steinberg S, Stefansson H, Jakobsson F, Lawlor DA, Evans DM, Ring SM, Farkkila M, Artto V, Kaunisto MA, Freilinger T, Schoenen J, Frants RR, Pelzer N, Weller CM, Zielman R, Heath AC, Madden PaF, Montgomery GW, Martin NG, Borck G, Gobel H, Heinze A, Heinze-Kuhn K, Williams FMK, Hartikainen AL, Pouta A, Van Den Ende J, Uitterlinden AG, Hofman A, Amin N, Hottenga JJ, Vink JM, Heikkila K, Alexander M, Muller-Miyhok B, Schreiber S, Meitinger T, Wichmann HE, Aromaa A, Eriksson JG, **Traynor B**, Trabzuni D, Rossin E, Lage K, Jacobs SBR, Gibbs JR, Birney E, Kaprio J, Penninx BW, Boomsma DI, Van Duijn C, Raitakari O, Jarvelin MR, Zwart JA, Cherkas L, Strachan DP, Kubisch C, Ferrari MD, Van Den Maagdenberg A, Dichgans M, Wessman M, Smith GD, Stefansson K, Daly MJ, Nyholt DR, Chasman D, Palotie A. *Nat Genet*. 2013;45:912-7. PMC4041123.
67. The p.A382T TARDBP gene mutation in Sardinian patients affected by Parkinson's disease and other degenerative parkinsonisms. Cannas A, Borghero G, Floris GL, Solla P, Chiò A, **Traynor BJ**, Calvo A, Restagno G, Majounie E, Costantino E, Piras V, Lavra L, Pani C, Orofino G, Di Stefano F, Tacconi P, Mascia MM, Muroli A, Murru MR, Tranquilli S, Corongiu D, Rolesu M, Cuccu S, Marrosu F, Marrosu MG. *Neurogenetics*. 2013;14:161-6. PMC3661017.
68. Global epidemiology of amyotrophic lateral sclerosis: a systematic review of the published literature. Chiò A, Logroscino G, **Traynor BJ**, Collins J, Simeone JC, Goldstein LA, White LA. *Neuroepidemiology*. 2013;41:118-30. PMC4049265.
69. UNC13A influences survival in Italian amyotrophic lateral sclerosis patients: a population-based study. Chiò A, Mora G, Restagno G, Brunetti M, Ossola I, Barberis M, Ferrucci L, Canosa A, Manera U, Moglia C, Fuda G, **Traynor BJ**, Calvo A. *Neurobiol Aging*. 2013;34:357.e1-5. PMC3483408.
70. RNA toxicity from the ALS/FTD C9ORF72 expansion is mitigated by antisense intervention. Donnelly CJ, Zhang PW, Pham JT, Haeusler AR, Mistry NA, Vidensky S, Daley EL, Poth EM, Hoover B, Fines DM, Maragakis N, Tienari PJ, Petrucelli L, **Traynor BJ**, Wang J, Rigo F, Bennett CF, Blackshaw S, Sattler R, Rothstein JD. *Neuron*. 2013;80:415-28. PMC4098943.
71. A pathway-based analysis provides additional support for an immune-related genetic susceptibility to Parkinson's disease. Holmans P, Moskvina V, Jones L, Sharma M; **International Parkinson Disease Genomics Consortium\***, Vedernikov A, Buchel F, Saad M, Bras JM, Bettella F, Nicolaou N, Simon-Sanchez J, Mittag F, Gibbs JR, Schulte C, Durr A, Guerreiro R, Hernandez D, Brice A, Stefansson H, Majamaa K, Gasser T, Heutink P, Wood NW, Martinez M, Singleton AB, Nalls MA, Hardy J, Morris HR, Williams NM. *Hum Mol Genet*. 2013;22:1039-49. PMC3561909.
72. Clinical Characteristics of C9ORF72-Linked Frontotemporal Lobar Degeneration. Kaivorinne AL, Bode MK, Paavola L, Tuominen H, Kallio M, Renton AE, **Traynor BJ**, Moilanen V, Remes AM. *Dement Geriatr Cogn Dis Extra*. 2013;3:251-62. PMC3776392.
73. Mutations in prion-like domains in hnRNPA2B1 and hnRNPA1 cause multisystem proteinopathy and ALS. Kim HJ, Kim NC, Wang YD, Scarborough EA, Moore J, Diaz Z, Maclea KS, Freibaum B, Li S, Molliex A, Kanagaraj AP, Carter R, Boylan KB, Wojtas AM, Rademakers R, Pinkus JL, Greenberg SA, Trojanowski JQ, **Traynor BJ**,

- Smith BN, Topp S, Gkazi AS, Miller J, Shaw CE, Kottlors M, Kirschner J, Pestronk A, Li YR, Ford AF, Gitler AD, Benatar M, King OD, Kimonis VE, Ross ED, Wehl CC, Shorter J, Taylor JP. *Nature*. 2013;495:467-73. PMC3756911.
74. The Val158Met COMT polymorphism is a modifier of the age at onset in Parkinson's disease with a sexual dimorphism. Klebe S, Golmard JL, Nalls MA, Saad M, Singleton AB, Bras JM, Hardy J, Simon-Sanchez J, Heutink P, Kuhlenbaumer G, Charfi R, Klein C, Hagenah J, Gasser T, Wurster I, Lesage S, Lorenz D, Deuschl G, Durif F, Pollak P, Damier P, Tison F, Durr A, Amouyel P, Lambert JC, Tzourio C, Maubaret C, Charbonnier-Beaupel F, Tahiri K, Vidailhet M, Martinez M, Brice A, Corvol JC; French Parkinson's Disease Genetics Study Group; **International Parkinson's Disease Genomics Consortium\***. *J Neurol Neurosurg Psychiatry*. 2013;84:666-73. PMC3646288.
75. Age-associated changes in gene expression in human brain and isolated neurons. Kumar A, Gibbs JR, Beilina A, Dillman A, Kumaran R, Trabzuni D, Ryten M, Walker R, Smith C, **Traynor BJ**, Hardy J, Singleton AB, Cookson MR. *Neurobiol Aging*. 2013;34:1199-209. PMC3545059.
76. Hereditary spastic paraplegia type 43 (SPG43) is caused by mutation in C19orf12. Landouere G, Zhu PP, Lourenco CM, Johnson JO, Toro C, Bricceno KV, Rinaldi C, Meilleur KG, Sangare M, Diallo O, Pierson TM, Ishiura H, Tsuji S, Hein N, Fink JK, Stoll M, Nicholson G, Gonzalez MA, Speziani F, Durr A, Stevanin G, Biesecker LG, Accardi J, Landis DM, Gahl WA, **Traynor BJ**, Marques W, Jr., Zuchner S, Blackstone C, Fischbeck KH, Burnett BG. *Hum Mutat*. 2013;34:1357-60. PMC3819934.
77. Frontotemporal dementia with a C9ORF72 expansion in a Swedish family: clinical and neuropathological characteristics. Landqvist Waldo M, Gustafson L, Nilsson K, **Traynor BJ**, Renton AE, Englund E, Passant U. *Am J Neurodegener Dis*. 2013;2:276-86. PMC3852567.
78. Homozygosity analysis in amyotrophic lateral sclerosis. Mok K, Laaksovirta H, Tienari PJ, Peuralinna T, Myllykangas L, Chiò A, **Traynor BJ**, Nalls MA, Guranlian N, Shatunov A, Restagno G, Mora G, Nigel Leigh P, Shaw CE, Morrison KE, Shaw PJ, Al-Chalabi A, Hardy J, Orrell RW. *Eur J Hum Genet*. 2013;21:1429-35. PMC3829775.
79. Analysis of genome-wide association studies of Alzheimer disease and of Parkinson disease to determine if these 2 diseases share a common genetic risk. Moskvina V, Harold D, Russo G, Vedernikov A, Sharma M, Saad M, Holmans P, Bras JM, Bettella F, Keller MF, Nicolaou N, Simon-Sanchez J, Gibbs JR, Schulte C, Durr A, Guerreiro R, Hernandez D, Brice A, Stefansson H, Majamaa K, Gasser T, Heutink P, Wood N, Martinez M, Singleton AB, Nalls MA, Hardy J, Owen MJ, O'donovan MC, Williams J, Morris HR, Williams NM; **International Parkinson Disease Genomics Consortium\***; GERAD. *JAMA Neurol*. 2013;70:1268-76. NIHMSID573448.
80. Serum iron levels and the risk of Parkinson disease: a Mendelian randomization study. Pichler I, Del Greco MF, Gogele M, Lill CM, Bertram L, Do CB, Eriksson N, Foroud T, Myers RH, Nalls M, Keller MF, **International Parkinson's Disease Genomics Consortium\***; Wellcome Trust Case Control Consortium 2; Benyamin B, Whitfield JB, Pramstaller PP, Hicks AA, Thompson JR, Minelli C. *PLoS Med*. 2013;10:e1001462. PMC3672214.
81. Resolving the polymorphism-in-probe problem is critical for correct interpretation of expression QTL studies. Ramasamy A, Trabzuni D, Gibbs JR, Dillman A, Hernandez DG, Arepalli S, Walker R, Smith C, Ilori GP, Shabalina AA, Li Y, Singleton AB, Cookson MR, **North American Brain Expression Consortium\***; Hardy J, Ryten M, Weale ME. *Nucleic Acids Res*. 2013;41:e88. PMC3627570.
82. GroupICA dual regression analysis of resting state networks in a behavioral variant of frontotemporal dementia. Rytty R, Nikkinen J, Paavola L, Abou Elseoud A, Moilanen V, Visuri A, Tervonen O, Renton AE, **Traynor BJ**, Kiviniemi V, Remes AM. *Front Hum Neurosci*. 2013;7:461. PMC3752460.
83. Genome-wide association study of Tourette's syndrome. Scharf JM, Yu D, Mathews CA, Neale BM, Stewart SE, Fagerness JA, Evans P, Gamazon E, Edlund CK, Service SK, Tikhomirov A, Osiecki L, Illmann C, Pluzhnikov A, Konkashbaev A, Davis LK, Han B, Crane J, Moorjani P, Crenshaw AT, Parkin MA, Reus VI, Lowe TL, Rangel-Lugo M, Chouinard S, Dion Y, Girard S, Cath DC, Smit JH, King RA, Fernandez TV, Leckman JF, Kidd KK, Kidd JR, Pakstis AJ, State MW, Herrera LD, Romero R, Fournier E, Sandor P, Barr CL, Phan N, Gross-Tsur V, Benarroch F, Pollak Y, Budman CL, Bruun RD, Erenberg G, Naarden AL, Lee PC, Weiss N, Kremeyer B, Berrio GB, Campbell DD, Cardona Silgado JC, Ochoa WC, Mesa Restrepo SC, Muller H, Valencia Duarte AV, Lyon GJ, Leppert M, Morgan J, Weiss R, Grados MA, Anderson K, Davarya S, Singer H, Walkup J, Jankovic J, Tischfield JA, Heiman GA, Gilbert DL, Hoekstra PJ, Robertson MM, Kurlan R, Liu C, Gibbs JR, Singleton A, **North American Brain Expression Consortium\***; Hardy J, Strengman E, Ophoff RA, Wagner M, Moessner R, Mirel DB, Posthuma D, Sabatti C, Eskin E, Conti DV, Knowles JA, Ruiz-Linares A, Rouleau GA, Purcell S, Heutink P, Oostra BA, McMahon WM, Freimer NB, Cox NJ, Pauls DL. *Mol Psychiatry*. 2013;18:721-8. PMC3605224.
84. Genome-wide association study of obsessive-compulsive disorder. Stewart SE, Yu D, Scharf JM, Neale BM, Fagerness JA, Mathews CA, Arnold PD, Evans PD, Gamazon ER, Davis LK, Osiecki L, Mcgrath L, Haddad S,

- Crane J, Hezel D, Illman C, Mayerfeld C, Konkashbaev A, Liu C, Pluzhnikov A, Tikhomirov A, Edlund CK, Rauch SL, Moessner R, Falkai P, Maier W, Ruhrmann S, Grabe HJ, Lennertz L, Wagner M, Bellodi L, Cavallini MC, Richter MA, Cook EH, Jr., Kennedy JL, Rosenberg D, Stein DJ, Hemmings SM, Lochner C, Azzam A, Chavira DA, Fournier E, Garrido H, Sheppard B, Umama P, Murphy DL, Wendland JR, Veenstra-Vanderweele J, Denys D, Blom R, Deforce D, Van Nieuwerburgh F, Westenberg HG, Walitz S, Egberts K, Renner T, Miguel EC, Cappi C, Hounie AG, Conceicao Do Rosario M, Sampaio AS, Vallada H, Nicolini H, Lanzagorta N, Camarena B, Delorme R, Leboyer M, Pato CN, Pato MT, Voyiaziakis E, Heutink P, Cath DC, Posthuma D, Smit JH, Samuels J, Bienvenu OJ, Cullen B, Fyer AJ, Grados MA, Greenberg BD, Mccracken JT, Riddle MA, Wang Y, Coric V, Leckman JF, Bloch M, Pittenger C, Eapen V, Black DW, Ophoff RA, Strengman E, Cusi D, Turiel M, Frau F, Macciardi F, Gibbs JR, Cookson MR, Singleton A, **North American Brain Expression Consortium\***; Hardy J, Crenshaw AT, Parkin MA, Mirel DB, Conti DV, Purcell S, Nestadt G, Hanna GL, Jenike MA, Knowles JA, Cox N, Pauls DL. *Mol Psychiatry*. 2013;18:788-98. PMC4218751.
85. Widespread sex differences in gene expression and splicing in the adult human brain. Trabzuni D, Ramasamy A, Imran S, Walker R, Smith C, Weale ME, Hardy J, Ryten M; **North American Brain Expression Consortium\***. *Nat Commun*. 2013;4:2771. PMC3868224.
86. Cis-regulatory variants affect CHRNA5 mRNA expression in populations of African and European ancestry. Wang JC, Spiegel N, Bertelsen S, Le N, Mckenna N, Budde JP, Harari O, Kapoor M, Brooks A, Hancock D, Tischfield J, Foroud T, Bierut LJ, Steinbach JH, Edenberg HJ, **Traynor BJ**, Goate AM. *PLoS One*. 2013;8:e80204. PMC3841173.
87. Screening for C9orf72 repeat expansions in parkinsonian syndromes. Yeh TH, Lai SC, Weng YH, Kuo HC, Wu-Chou YH, Huang CL, Chen RS, Chang HC, **Traynor B**, Lu CS. *Neurobiol Aging*. 2013;34:1311.e3-4. PMC4022748.
88. Unbiased screen for interactors of leucine-rich repeat kinase 2 supports a common pathway for sporadic and familial Parkinson disease. Beilina A, Rudenko IN, Kaganovich A, Civiero L, Chau H, Kalia SK, Kalia LV, Lobbstaal E, Chia R, Ndukwe K, Ding J, Nalls MA, International Parkinson's Disease Genomics Consortium; **North American Brain Expression Consortium\***; Olszewski M, Hauser DN, Kumaran R, Lozano AM, Baekelandt V, Greene LE, Taymans JM, Greggio E, Cookson MR. *Proc Natl Acad Sci U S A*. 2014;111:2626-31. PMC3932908.
89. Mutations in the CHCHD10 gene are a common cause of familial amyotrophic lateral sclerosis. Johnson JO, Glynn SM, Gibbs JR, Nalls MA, Sabatelli M, Restagno G, Drory VE, Chiò A, Rogaeva E, **Traynor BJ**. *Brain*. 2014;137:e311. PMC4240285.
90. Genetic architecture of ALS in Sardinia. Borghero G, Pugliatti M, Marrosu F, Marrosu MG, Murru MR, Floris G, Cannas A, Parish LD, Occhineri P, Cau TB, Loi D, Ticca A, Traccis S, Manera U, Canosa A, Moglia C, Calvo A, Barberis M, Brunetti M, Pliner HA, Renton AE, Nalls MA, **Traynor BJ**, Restagno G, Chiò A. *Neurobiol Aging*. 2014;35:2882.e7-.e12. PMC4252367.
91. De novo nonsense mutation of the FUS gene in an apparently familial amyotrophic lateral sclerosis case. Calvo A, Moglia C, Canosa A, Brunetti M, Barberis M, **Traynor BJ**, Carrara G, Valentini C, Restagno G, Chiò A. *Neurobiol Aging*. 2014;35:1513.e7-11. PMC3961545.
92. The metabolic signature of C9ORF72-related ALS: FDG PET comparison with nonmutated patients. Cistaro A, Pagani M, Montuschi A, Calvo A, Moglia C, Canosa A, Restagno G, Brunetti M, **Traynor BJ**, Nobili F, Carrara G, Fania P, Lopiano L, Valentini MC, Chiò A. *Eur J Nucl Med Mol Imaging*. 2014;41:844-52. NIHMSID940289.
93. C9orf72 and UNC13A are shared risk loci for amyotrophic lateral sclerosis and frontotemporal dementia: a genome-wide meta-analysis. Diekstra FP, Van Deerlin VM, Van Swieten JC, Al-Chalabi A, Ludolph AC, Weishaupt JH, Hardiman O, Landers JE, Brown RH, Jr., Van Es MA, Pasterkamp RJ, Koppers M, Andersen PM, Estrada K, Rivadeneira F, Hofman A, Uitterlinden AG, Van Damme P, Melki J, Meininger V, Shatunov A, Shaw CE, Leigh PN, Shaw PJ, Morrison KE, Fogh I, Chiò A, **Traynor BJ**, Czell D, Weber M, Heutink P, De Bakker PI, Silani V, Robberecht W, Van Den Berg LH, Veldink JH. *Ann Neurol*. 2014;76:120-33. PMC4137231.
94. Susceptibility loci for pigmentation and melanoma in relation to Parkinson's disease. Dong J, Gao J, Nalls M, Gao X, Huang X, Han J, Singleton AB, Chen H; **International Parkinson's Disease Genomics Consortium\***. *Neurobiol Aging*. 2014;35:1512.e5-e10. PMC3961492.
95. A genome-wide association meta-analysis identifies a novel locus at 17q11.2 associated with sporadic amyotrophic lateral sclerosis. Fogh I, Ratti A, Gellera C, Lin K, Tiloca C, Moskvina V, Corrado L, Soraru G, Cereda C, Corti S, Gentilini D, Calini D, Castellotti B, Mazzini L, Querin G, Gagliardi S, Del Bo R, Conforti FL, Siciliano G, Inghilleri M, Sacca F, Bongioanni P, Penco S, Corbo M, Sorbi S, Filosto M, Ferlini A, Di Blasio AM, Signorini S, Shatunov A, Jones A, Shaw PJ, Morrison KE, Farmer AE, Van Damme P, Robberecht W, Chiò A, **Traynor BJ**, Sendtner M, Melki J, Meininger V, Hardiman O, Andersen PM, Leigh NP, Glass JD, Overste D, Diekstra FP, Veldink JH, Van Es MA, Shaw CE, Weale ME, Lewis CM, Williams J, Brown RH, Landers JE, Ticozzi N, Ceroni

- M, Pegoraro E, Comi GP, D'alfonso S, Van Den Berg LH, Taroni F, Al-Chalabi A, Powell J, Silani V. *Hum Mol Genet.* 2014;23:2220-31. PMC3959809.
96. No evidence for shared genetic basis of common variants in multiple sclerosis and amyotrophic lateral sclerosis. Goris A, Van Setten J, Diekstra F, Ripke S, Patsopoulos NA, Sawcer SJ, Van Es M, Andersen PM, Melki J, Meininger V, Hardiman O, Landers JE, Brown RH, Jr., Shatunov A, Leigh N, Al-Chalabi A, Shaw CE, **Traynor BJ**, Chiò A, Restagno G, Mora G, Ophoff RA, Oksenberg JR, Van Damme P, Compston A, Robberecht W, Dubois B, Van Den Berg LH, De Jager PL, Veldink JH, De Bakker PI. *Hum Mol Genet.* 2014;23:1916-22. PMC3943520.
97. Novel TARDBP sequence variant and C9ORF72 repeat expansion in a family with frontotemporal dementia. Kaivorinne AL, Moilanen V, Kervinen M, Renton AE, **Traynor BJ**, Majamaa K, Remes AM. *Alzheimer Dis Assoc Disord.* 2014;28:190-3. PMC3511614.
98. Genome-wide analysis of the heritability of amyotrophic lateral sclerosis. Keller MF, Ferrucci L, Singleton AB, Tienari PJ, Laaksovirta H, Restagno G, Chiò A, **Traynor BJ**, Nalls MA. *JAMA Neurol.* 2014;71:1123-34. PMC4566960.
99. Large-scale meta-analysis of genome-wide association data identifies six new risk loci for Parkinson's disease. Nalls MA, Pankratz N, Lill CM, Do CB, Hernandez DG, Saad M, Destefano AL, Kara E, Bras J, Sharma M, Schulte C, Keller MF, Arepalli S, Letson C, Edsall C, Stefansson H, Liu X, Pliner H, Lee JH, Cheng R, **International Parkinson's Disease Genomics Consortium\***; Ikram MA, Ioannidis JP, Hadjigeorgiou GM, Bis JC, Martinez M, Perlmutter JS, Goate A, Marder K, Fiske B, Sutherland M, Xiromerisiou G, Myers RH, Clark LN, Stefansson K, Hardy JA, Heutink P, Chen H, Wood NW, Houlden H, Payami H, Brice A, Scott WK, Gasser T, Bertram L, Eriksson N, Foroud T, Singleton AB. *Nat Genet.* 2014;46:989-93. PMC4146673.
100. Genetic comorbidities in Parkinson's disease. Nalls MA, Saad M, Noyce AJ, Keller MF, Schrag A, Bestwick JP, **Traynor BJ**, Gibbs JR, Hernandez DG, Cookson MR, Morris HR, Williams N, Gasser T, Heutink P, Wood N, Hardy J, Martinez M, Singleton AB. *Hum Mol Genet.* 2014;23:831-41. PMC3888265.
101. Hippocampal sclerosis dementia with the C9ORF72 hexanucleotide repeat expansion. Pletnikova O, Sloane KL, Renton AE, **Traynor BJ**, Crain BJ, Reid T, Zu T, Ranum LP, Troncoso JC, Rabins PV, Onyike CU. *Neurobiol Aging.* 2014;35:2419.e17-21. PMC4087047.
102. Genetic variability in the regulation of gene expression in ten regions of the human brain. Ramasamy A, Trabzuni D, Guelfi S, Varghese V, Smith C, Walker R, De T, **North American Brain Expression Consortium\***; Coin L, De Silva R, Cookson MR, Singleton AB, Hardy J, Ryten M, Weale ME. *Nat Neurosci.* 2014;17:1418-28. PMC4208299.
103. Discovery of a Biomarker and Lead Small Molecules to Target r(GGGGCC)-Associated Defects in c9FTD/ALS. Su Z, Zhang Y, Gendron TF, Bauer PO, Chew J, Yang WY, Fostvedt E, Jansen-West K, Belzil VV, Desaro P, Johnston A, Overstreet K, Oh SY, Todd PK, Berry JD, Cudkowicz ME, Boeve BF, Dickson D, Floeter MK, **Traynor BJ**, Morelli C, Ratti A, Silani V, Rademakers R, Brown RH, Rothstein JD, Boylan KB, Petrucelli L, Disney MD. *Neuron.* 2014;84:239. NIHMSID619164.
104. Analysis of the KIFAP3 gene in amyotrophic lateral sclerosis: a multicenter survival study. Van Doormaal PT, Ticozzi N, Gellera C, Ratti A, Taroni F, Chiò A, Calvo A, Mora G, Restagno G, **Traynor BJ**, Birve A, Lemmens R, Van Es MA, Saris CG, Blauw HM, Van Vught PW, Groen EJ, Corrado L, Mazzini L, Del Bo R, Corti S, Waibel S, Meyer T, Ludolph AC, Goris A, Van Damme P, Robberecht W, Shatunov A, Fogh I, Andersen PM, D'alfonso S, Hardiman O, Cronin S, Rujescu D, Al-Chalabi A, Landers JE, Silani V, Van Den Berg LH, Veldink JH. *Neurobiol Aging.* 2014;35:2420.e13-4. PMC5496711.
105. ATXN2 is a modifier of phenotype in ALS patients of Sardinian ancestry. Borghero G, Pugliatti M, Marrosu F, Marrosu MG, Murru MR, Floris G, Cannas A, Parish LD, Cau TB, Loi D, Ticca A, Traccis S, Manera U, Canosa A, Moglia C, Calvo A, Barberis M, Brunetti M, Renton AE, Nalls MA, **Traynor BJ**, Restagno G, Chiò A. *Neurobiol Aging.* 2015;36:2906.e1-5. PMC5193218.
106. HFE p.H63D polymorphism does not influence ALS phenotype and survival. Chiò A, Mora G, Sabatelli M, Caponnetto C, Lunetta C, **Traynor BJ**, Johnson JO, Nalls MA, Calvo A, Moglia C, Borghero G, Monsurro MR, La Bella V, Volanti P, Simone I, Salvi F, Logullo FO, Nilo R, Giannini F, Mandrioli J, Tanel R, Murru MR, Mandich P, Zollino M, Conforti FL, Penco S, Brunetti M, Barberis M, Restagno G. *Neurobiol Aging.* 2015;36:2906.e7-11. PMC5183653.
107. CHCH10 mutations in an Italian cohort of familial and sporadic amyotrophic lateral sclerosis patients. Chiò A, Mora G, Sabatelli M, Caponnetto C, **Traynor BJ**, Johnson JO, Nalls MA, Calvo A, Moglia C, Borghero G, Monsurro MR, La Bella V, Volanti P, Simone I, Salvi F, Logullo FO, Nilo R, Battistini S, Mandrioli J, Tanel R, Murru MR, Mandich P, Zollino M, Conforti FL, Brunetti M, Barberis M, Restagno G, Penco S, Lunetta C. *Neurobiol Aging.* 2015;36:1767.e3-.e6. PMC4380794.
108. Genome-wide meta-analysis identifies six novel loci associated with habitual coffee consumption. Cornelis MC,

- Byrne EM, Esko T, Nalls MA, Ganna A, Paynter N, Monda KL, Amin N, Fischer K, Renstrom F, Ngwa JS, Huikari V, Cavadino A, Nolte IM, Teumer A, Yu K, Marques-Vidal P, Rawal R, Manichaikul A, Wojczynski MK, Vink JM, Zhao JH, Burlutsky G, Lahti J, Mikkila V, Lemaitre RN, Eriksson J, Musani SK, Tanaka T, Geller F, Luan J, Hui J, Magi R, Dimitriou M, Garcia ME, Ho WK, Wright MJ, Rose LM, Magnusson PK, Pedersen NL, Couper D, Oostra BA, Hofman A, Ikram MA, Tiemeier HW, Uitterlinden AG, Van Rooij FJ, Barroso I, Johansson I, Xue L, Kaakinen M, Milani L, Power C, Snieder H, Stolk RP, Baumeister SE, Biffar R, Gu F, Bastardot F, Kutalik Z, Jacobs DR, Jr., Forouhi NG, Mihailov E, Lind L, Lindgren C, Michaelsson K, Morris A, Jensen M, Khaw KT, Luben RN, Wang JJ, Mannisto S, Perala MM, Kahonen M, Lehtimaki T, Viikari J, Mozaffarian D, Mukamal K, Psaty BM, Doring A, Heath AC, Montgomery GW, Dahmen N, Carithers T, Tucker KL, Ferrucci L, Boyd HA, Melbye M, Treur JL, Mellstrom D, Hottenga JJ, Prokopenko I, Tonjes A, Deloukas P, Kanoni S, Lorentzon M, Houston DK, Liu Y, Danesh J, Rasheed A, Mason MA, Zonderman AB, Franke L, Kristal BS, **International Parkinson's Disease Genomics Consortium (IPDGC)\*; North American Brain Expression Consortium (NABEC)\***; Karjalainen J, Reed DR, Westra HJ, Evans MK, Saleheen D, Harris TB, Dedoussis G, Curhan G, Stumvoll M, Beilby J, Pasquale LR, Feenstra B, Bandinelli S, Ordovas JM, Chan AT, Peters U, Ohlsson C, Gieger C, Martin NG, Waldenberger M, Siscovick DS, Raitakari O, Eriksson JG, Mitchell P, Hunter DJ, Kraft P, Rimm EB, Boomsma DI, Borecki IB, Loos RJ, Wareham NJ, Vollenweider P, Caporaso N, Grabe HJ, Neuhaus ML, Wolfenbutter BH, Hu FB, Hypponen E, Jarvelin MR, Cupples LA, Franks PW, Ridker PM, Van Duijn CM, Heiss G, Metspalu A, North KE, Ingelsson E, Nettleton JA, Van Dam RM, Chasman DI. *Mol Psychiatry*. 2015;20:647-56. PMC4388784.
109. Screening a UK amyotrophic lateral sclerosis cohort provides evidence of multiple origins of the C9orf72 expansion. Fratta P, Polke JM, Newcombe J, Mizielinska S, Lashley T, Poulter M, Beck J, Preza E, Devoy A, Sidle K, Howard R, Malaspina A, Orrell RW, Clarke J, Lu CH, Mok K, Collins T, Shoai M, Nanji T, Wray S, Adamson G, Pittman A, Renton AE, **Traynor BJ**, Sweeney MG, Revesz T, Houlden H, Mead S, Isaacs AM, Fisher EM. *Neurobiol Aging*. 2015;36:546.e1-7. PMC4270445.
110. Common genetic variants influence human subcortical brain structures. Hibar DP, Stein JL, Renteria ME, Arias-Vasquez A, Desrivieres S, Jahanshad N, Toro R, Wittfeld K, Abramovic L, Andersson M, Aribisala BS, Armstrong NJ, Bernard M, Bohlken MM, Boks MP, Bralten J, Brown AA, Chakravarty MM, Chen Q, Ching CR, Cuellar-Partida G, Den Braber A, Giddaluru S, Goldman AL, Grimm O, Guadalupe T, Hass J, Woldehawariat G, Holmes AJ, Hoogman M, Janowitz D, Jia T, Kim S, Klein M, Kraemer B, Lee PH, Olde Loohuis LM, Luciano M, Macare C, Mather KA, Mattheisen M, Milanese Y, Nho K, Pappmeyer M, Ramasamy A, Risacher SL, Roiz-Santianez R, Rose EJ, Salami A, Samann PG, Schmaal L, Schork AJ, Shin J, Strike LT, Teumer A, Van Donkelaar MM, Van Eijk KR, Walters RK, Westlye LT, Whelan CD, Winkler AM, Zwiers MP, Alhusaini S, Athanasiu L, Ehrlich S, Hakobyan MM, Hartberg CB, Haukvik UK, Heister AJ, Hoehn D, Kasperaviciute D, Liewald DC, Lopez LM, Makkinje RR, Matarin M, Naber MA, McKay DR, Needham M, Nugent AC, Putz B, Royle NA, Shen L, Sprooten E, Trabzuni D, Van Der Marel SS, Van Hulzen KJ, Walton E, Wolf C, Almasy L, Ames D, Arepalli S, Assareh AA, Bastin ME, Brodaty H, Bulayeva KB, Carless MA, Cichon S, Corvin A, Curran JE, Czisch M, De Zubicaray GI, Dillman A, Duggirala R, Dyer TD, Erk S, Fedko IO, Ferrucci L, Foroud TM, Fox PT, Fukunaga M, Gibbs JR, Goring HH, Green RC, Guelfi S, Hansell NK, Hartman CA, Hegenscheid K, Heinz A, Hernandez DG, Heslenfeld DJ, Hoekstra PJ, Holsboer F, Homuth G, Hottenga JJ, Ikeda M, Jack CR, Jr., Jenkinson M, Johnson R, Kanai R, Keil M, Kent JW, Jr., Kochunov P, Kwok JB, Lawrie SM, Liu X, Longo DL, McMahon KL, Meisenzahl E, Melle I, Mohnke S, Montgomery GW, Mostert JC, Muhleisen TW, Nalls MA, Nichols TE, Nilsson LG, Nothen MM, Ohi K, Olvera RL, Perez-Iglesias R, Pike GB, Potkin SG, Reinvang I, Reppermund S, Rietschel M, Romanczuk-Seiferth N, Rosen GD, Rujescu D, Schnell K, Schofield PR, Smith C, Steen VM, Sussmann JE, Thalamuthu A, Toga AW, **Traynor BJ**, Troncoso J, Turner JA, Valdes Hernandez MC, Van 'T Ent D, Van Der Brug M, Van Der Wee NJ, Van Tol MJ, Veltman DJ, Wassink TH, Westman E, Zielke RH, Zonderman AB, Ashbrook DG, Hager R, Lu L, McMahon FJ, Morris DW, Williams RW, Brunner HG, Buckner RL, Buitelaar JK, Cahn W, Calhoun VD, Cavalleri GL, Crespo-Facorro B, Dale AM, Davies GE, Delanty N, Depondt C, Djurovic S, Drevets WC, Espeseth T, Gollub RL, Ho BC, Hoffmann W, Hosten N, Kahn RS, Le Hellard S, Meyer-Lindenberg A, Muller-Myhsok B, Nauck M, Nyberg L, Pandolfo M, Penninx BW, Roffman JL, Sisodiya SM, Smoller JW, Van Bokhoven H, Van Haren NE, Volzke H, Walter H, Weiner MW, Wen W, White T, Agartz I, Andreassen OA, Blangero J, Boomsma DI, Brouwer RM, Cannon DM, Cookson MR, De Geus EJ, Deary IJ, Donohoe G, Fernandez G, Fisher SE, Francks C, Glahn DC, Grabe HJ, Gruber O, Hardy J, Hashimoto R, Hulshoff Pol HE, Jonsson EG, Kloszewska I, Lovestone S, Mattay VS, Mecocci P, McDonald C, McIntosh AM, Ophoff RA, Paus T, Pausova Z, Ryten M, Sachdev PS, Saykin AJ, Simmons A, Singleton A, Soininen H, Wardlaw JM, Weale ME, Weinberger DR, Adams HH, Launer LJ, Seiler S, Schmidt R, Chauhan G, Satizabal CL, Becker JT, Yanek L, Van Der Lee SJ, Ebling M, Fischl B, Longstreth WT, Jr., Greve D, Schmidt H, Nyquist P, Vinke LN, Van Duijn CM, Xue L, Mazoyer B, Bis JC, Gudnason V, Seshadri S, Ikram MA, Martin NG, Wright MJ,

- Schumann G, Franke B, Thompson PM, Medland SE. *Nature*. 2015;520:224-9. PMC4393366.
111. NeuroX, a fast and efficient genotyping platform for investigation of neurodegenerative diseases. Nalls MA, Bras J, Hernandez DG, Keller MF, Majounie E, Renton AE, Saad M, Jansen I, Guerreiro R, Lubbe S, Plagnol V, Gibbs JR, Schulte C, Pankratz N, Sutherland M, Bertram L, Lill CM, Destefano AL, Faroud T, Eriksson N, Tung JY, Edsall C, Nichols N, Brooks J, Arepalli S, Pliner H, Letson C, Heutink P, Martinez M, Gasser T, **Traynor BJ**, Wood N, Hardy J, Singleton AB. *Neurobiol Aging*. 2015;36:1605.e7-12. PMC4317375.
112. The transcriptional landscape of age in human peripheral blood. Peters MJ, Joehanes R, Pilling LC, Schurmann C, Conneely KN, Powell J, Reinmaa E, Sutphin GL, Zhernakova A, Schramm K, Wilson YA, Kobes S, Tukiainen T, **North American Brain Expression Consortium\***; Ramos YF, Goring HH, Fornage M, Liu Y, Gharib SA, Stranger BE, De Jager PL, Aviv A, Levy D, Murabito JM, Munson PJ, Huan T, Hofman A, Uitterlinden AG, Rivadeneira F, Van Rooij J, Stolk L, Broer L, Verbiest MM, Jhamai M, Arp P, Metspalu A, Tserel L, Milani L, Samani NJ, Peterson P, Kasela S, Codd V, Peters A, Ward-Caviness CK, Herder C, Waldenberger M, Roden M, Singmann P, Zeilinger S, Illig T, Homuth G, Grabe HJ, Volzke H, Steil L, Kocher T, Murray A, Melzer D, Yaghootkar H, Bandinelli S, Moses EK, Kent JW, Curran JE, Johnson MP, Williams-Blangero S, Westra HJ, Mcrae AF, Smith JA, Kardia SL, Hovatta I, Perola M, Ripatti S, Salomaa V, Henders AK, Martin NG, Smith AK, Mehta D, Binder EB, Nylocks KM, Kennedy EM, Klengel T, Ding J, Suchy-Dacey AM, Enquobahrie DA, Brody J, Rotter JI, Chen YD, Houwing-Duistermaat J, Kloppenburg M, Slagboom PE, Helmer Q, Den Hollander W, Bean S, Raj T, Bakhshi N, Wang QP, Oyston LJ, Psaty BM, Tracy RP, Montgomery GW, Turner ST, Blangero J, Meulenbelt I, Ressler KJ, Yang J, Franke L, Kettunen J, Visscher PM, Neely GG, Korstanje R, Hanson RL, Prokisch H, Ferrucci L, Esko T, Teumer A, Van Meurs JB, Johnson AD. *Nat Commun*. 2015;6:8570. PMC4639797.
113. Genome-wide association study of neocortical Lewy-related pathology. Peuralinna T, Myllykangas L, Oinas M, Nalls MA, Keage HA, Isoviita VM, Valori M, Polvikoski T, Paetau A, Sulkava R, Ince PG, Zaccai J, Brayne C, **Traynor BJ**, Hardy J, Singleton AB, Tienari PJ. *Ann Clin Transl Neurol*. 2015;2:920-31. PMC4574809.
114. Mutation in CPT1C Associated With Pure Autosomal Dominant Spastic Paraplegia. Rinaldi C, Schmidt T, Situ AJ, Johnson JO, Lee PR, Chen KL, Bott LC, Fado R, Harmison GH, Parodi S, Grunseich C, Renvoise B, Biesecker LG, De Michele G, Santorelli FM, Filla A, Stevanin G, Durr A, Brice A, Casals N, **Traynor BJ**, Blackstone C, Ulmer TS, Fischbeck KH. *JAMA Neurol*. 2015;72:561-70. PMC5612424.
115. Small deletion in C9orf72 hides a proportion of expansion carriers in FTLD. Rollinson S, Bennion Callister J, Young K, Ryan SJ, Drueyeh R, Rohrer JD, Snowden J, Richardson A, Jones M, Harris J, Davidson Y, Robinson A, Ealing J, Johnson JO, **Traynor B**, Mead S, Mann D, Pickering-Brown SM. *Neurobiol Aging*. 2015;36:1601.e1-5. PMC4353501.
116. The Phenotype of the C9ORF72 Expansion Carriers According to Revised Criteria for bvFTD. Solje E, Aaltokallio H, Koivumaa-Honkanen H, Suhonen NM, Moilanen V, Kiviharju A, **Traynor B**, Tienari PJ, Hartikainen P, Remes AM. *PLoS One*. 2015;10:e0131817. PMC4493025.
117. Mutation analysis of CHCHD10 in different neurodegenerative diseases. Zhang M, Xi Z, Zinman L, Bruni AC, Maletta RG, Curcio SA, Rainero I, Rubino E, Pinessi L, Nacmias B, Sorbi S, Galimberti D, Lang AE, Fox S, Surace EI, Ghani M, Guo J, Sato C, Moreno D, Liang Y, Keith J, **Traynor BJ**, St George-Hyslop P, Rogava E. *Brain*. 2015;138:e380. PMC4547051.
118. Association of a Novel ACTA1 Mutation With a Dominant Progressive Scapuloperoneal Myopathy in an Extended Family. Zukosky K, Meilleur K, **Traynor BJ**, Dastgir J, Medne L, Devoto M, Collins J, Rooney J, Zou Y, Yang ML, Gibbs JR, Meier M, Stetefeld J, Finkel RS, Schessl J, Elman L, Felice K, Ferguson TA, Ceyhan-Birsoy O, Beggs AH, Tennekoon G, Johnson JO, Bonnemann CG. *JAMA Neurol*. 2015;72:689-98. PMC4461456.
119. Novel genetic loci underlying human intracranial volume identified through genome-wide association. Adams HH, Hibar DP, Chouraki V, Stein JL, Nyquist PA, Renteria ME, Trompet S, Arias-Vasquez A, Seshadri S, Desrivieres S, Beecham AH, Jahanshad N, Wittfeld K, Van Der Lee SJ, Abramovic L, Alhusaini S, Amin N, Andersson M, Arfanakis K, Aribisala BS, Armstrong NJ, Athanasiu L, Axelsson T, Beiser A, Bernard M, Bis JC, Blanken LM, Blanton SH, Bohlken MM, Boks MP, Bralten J, Brickman AM, Carmichael O, Chakravarty MM, Chauhan G, Chen Q, Ching CR, Cuellar-Partida G, Braber AD, Doan NT, Ehrlich S, Filippi I, Ge T, Giddaluru S, Goldman AL, Gottesman RF, Greven CU, Grimm O, Griswold ME, Guadalupe T, Hass J, Haukvik UK, Hilal S, Hofer E, Hoehn D, Holmes AJ, Hoogman M, Janowitz D, Jia T, Kasperaviciute D, Kim S, Klein M, Kraemer B, Lee PH, Liao J, Liewald DC, Lopez LM, Luciano M, Macare C, Marquand A, Matarin M, Mather KA, Mattheisen M, Mazoyer B, McKay DR, McWhirter R, Milaneschi Y, Mirza-Schreiber N, Muetzel RL, Maniega SM, Nho K, Nugent AC, Loohuis LM, Oosterlaan J, Pappmeyer M, Pappa I, Pirpamer L, Pudas S, Putz B, Rajan KB, Ramasamy A, Richards JS, Risacher SL, Roiz-Santanez R, Rommelse N, Rose EJ, Royle NA, Rundek T, Samann PG, Satizabal CL, Schmaal L, Schork AJ, Shen L, Shin J, Shumskaya E, Smith AV, Sprooten E, Strike LT, Teumer A, Thomson R, Tordesillas-Gutierrez D, Toro R, Trabzuni D, Vaidya D, Van Der Grond J, Van Der Meer D,



- Van Donkelaar MM, Van Eijk KR, Van Erp TG, Van Rooij D, Walton E, Westlye LT, Whelan CD, Windham BG, Winkler AM, Woldehawariat G, Wolf C, Wolfers T, Xu B, Yanek LR, Yang J, Zijdenbos A, Zwiers MP, Agartz I, Aggarwal NT, Almasy L, Ames D, Amouyel P, Andreassen OA, Arepalli S, Assareh AA, Barral S, Bastin ME, Becker DM, Becker JT, Bennett DA, Blangero J, Van Bokhoven H, Boomsma DI, Brodaty H, Brouwer RM, Brunner HG, Buckner RL, Buitelaar JK, Bulayeva KB, Cahn W, Calhoun VD, Cannon DM, Cavalleri GL, Chen C, Cheng CY, Cichon S, Cookson MR, Corvin A, Crespo-Facorro B, Curran JE, Czisch M, Dale AM, Davies GE, De Geus EJ, De Jager PL, De Zubicaray GI, Delanty N, Depondt C, Destefano AL, Dillman A, Djurovic S, Donohoe G, Drevets WC, Duggirala R, Dyer TD, Erk S, Espeseth T, Evans DA, Fedko IO, Fernandez G, Ferrucci L, Fisher SE, Fleischman DA, Ford I, Foroud TM, Fox PT, Francks C, Fukunaga M, Gibbs JR, Glahn DC, Gollub RL, Goring HH, Grabe HJ, Green RC, Gruber O, Gudnason V, Guelfi S, Hansell NK, Hardy J, Hartman CA, Hashimoto R, Hegenscheid K, Heinz A, Le Hellard S, Hernandez DG, Heslenfeld DJ, Ho BC, Hoekstra PJ, Hoffmann W, Hofman A, Holsboer F, Homuth G, Hosten N, Hottenga JJ, Hulshoff Pol HE, Ikeda M, Ikram MK, Jack CR, Jr., Jenkinson M, Johnson R, Jonsson EG, Jukema JW, Kahn RS, Kanai R, Kloszewska I, Knopman DS, Kochunov P, Kwok JB, Lawrie SM, Lemaitre H, Liu X, Longo DL, Longstreth WT, Jr., Lopez OL, Lovestone S, Martinez O, Martinot JL, Mattay VS, McDonald C, McIntosh AM, McMahon KL, McMahon FJ, Mecocci P, Melle I, Meyer-Lindenberg A, Mohnke S, Montgomery GW, Morris DW, Mosley TH, Muhleisen TW, Muller-Myhsok B, Nalls MA, Nauck M, Nichols TE, Niessen WJ, Nothen MM, Nyberg L, Ohi K, Olvera RL, Ophoff RA, Pandolfo M, Paus T, Pausova Z, Penninx BW, Pike GB, Potkin SG, Psaty BM, Reppermund S, Rietschel M, Roffman JL, Romanczuk-Seiferth N, Rotter JI, Ryten M, Sacco RL, Sachdev PS, Saykin AJ, Schmidt R, Schofield PR, Sigurdsson S, Simmons A, Singleton A, Sisodiya SM, Smith C, Smoller JW, Soininen H, Srikanth V, Steen VM, Stott DJ, Sussmann JE, Thalamuthu A, Tiemeier H, Toga AW, **Traynor BJ**, Troncoso J, Turner JA, Tzourio C, Uitterlinden AG, Hernandez MC, Van Der Brug M, Van Der Lugt A, Van Der Wee NJ, Van Duijn CM, Van Haren NE, Van TED, Van Tol MJ, Vardarajan BN, Veltman DJ, Vernooij MW, Volzke H, Walter H, Wardlaw JM, Wassink TH, Weale ME, Weinberger DR, Weiner MW, Wen W, Westman E, White T, Wong TY, Wright CB, Zielke HR, Zonderman AB, Deary IJ, Decarli C, Schmidt H, Martin NG, De Craen AJ, Wright MJ, Launer LJ, Schumann G, Fornage M, Franke B, Dobbins S, Medland SE, Ikram MA, Thompson PM. *Nat Neurosci.* 2016;19:1569-82. PMC5227112.
120. DNA repair pathways underlie a common genetic mechanism modulating onset in polyglutamine diseases. Bettencourt C, Hensman-Moss D, Flower M, Wiethoff S, Brice A, Goizet C, Stevanin G, Koutsis G, Karadima G, Panas M, Yescas-Gomez P, Garcia-Velazquez LE, Alonso-Vilatela ME, Lima M, Raposo M, **Traynor B**, Sweeney M, Wood N, Giunti P, Durr A, Holmans P, Houlden H, Tabrizi SJ, Jones L. *Ann Neurol.* 2016;79:983-90. PMC4914895.
121. TBK1 is associated with ALS and ALS-FTD in Sardinian patients. Borghero G, Pugliatti M, Marrosu F, Marrosu MG, Murru MR, Floris G, Cannas A, Occhineri P, Cau TB, Loi D, Ticca A, Traccis S, Manera U, Canosa A, Moglia C, Calvo A, Barberis M, Brunetti M, Gibbs JR, Renton AE, Errichiello E, Zoledziewska M, Mulas A, Qian Y, Din J, Pliner HA, **Traynor BJ**, Chiò A. *Neurobiol Aging.* 2016;43:180.e1-5. NIHMSID940290.
122. ATNX2 is not a regulatory gene in Italian amyotrophic lateral sclerosis patients with C9ORF72 GGGGCC expansion. Chiò A, Mora G, Sabatelli M, Caponnetto C, Lunetta C, **Traynor BJ**, Johnson JO, Nalls MA, Calvo A, Moglia C, Borghero G, Trojsi F, La Bella V, Volanti P, Simone I, Salvi F, Logullo FO, Riva N, Carrera P, Giannini F, Mandrioli J, Tanel R, Capasso M, Tremolizzo L, Battistini S, Murru MR, Origone P, Zollino M, Penco S, Mazzini L, D'Alfonso S, Restagno G, Brunetti M, Barberis M, Conforti FL. *Neurobiol Aging.* 2016;39:218.e5-8. PMC4775342.
123. Longitudinal imaging in C9orf72 mutation carriers: Relationship to phenotype. Floeter MK, Bageac D, Danielian LE, Braun LE, **Traynor BJ**, Kwan JY. *Neuroimage Clin.* 2016;12:1035-43. PMC5153604.
124. Association of a Locus in the CAMTA1 Gene With Survival in Patients With Sporadic Amyotrophic Lateral Sclerosis. Fogh I, Lin K, Tiloca C, Rooney J, Gellera C, Diekstra FP, Ratti A, Shatunov A, Van Es MA, Proitsi P, Jones A, Sproviero W, Chiò A, McLaughlin RL, Soraru G, Corrado L, Stahl D, Del Bo R, Cereda C, Castellotti B, Glass JD, Newhouse S, Dobson R, Smith BN, Topp S, Van Rheenen W, Meininger V, Melki J, Morrison KE, Shaw PJ, Leigh PN, Andersen PM, Comi GP, Ticozzi N, Mazzini L, D'Alfonso S, **Traynor BJ**, Van Damme P, Robberecht W, Brown RH, Landers JE, Hardiman O, Lewis CM, Van Den Berg LH, Shaw CE, Veldink JH, Silani V, Al-Chalabi A, Powell J. *JAMA Neurol.* 2016;73:812-20. PMC5556366.
125. Genetic influences on schizophrenia and subcortical brain volumes: large-scale proof of concept. Franke B, Stein JL, Ripke S, Anttila V, Hibar DP, Van Hulzen KJE, Arias-Vasquez A, Smoller JW, Nichols TE, Neale MC, McIntosh AM, Lee P, McMahon FJ, Meyer-Lindenberg A, Mattheisen M, Andreassen OA, Gruber O, Sachdev PS, Roiz-Santianez R, Saykin AJ, Ehrlich S, Mather KA, Turner JA, Schwarz E, Thalamuthu A, Shugart YY, Ho YY, Martin NG, Wright MJ, Schizophrenia Working Group of the Psychiatric Genomics Consortium, Enigma Consortium, O'Donovan MC, Thompson PM, Neale BM, Medland SE, Sullivan PF. *Nat Neurosci.* 2016;19:420-

31. PMC4852730.
126. Mutation analysis of the MS4A and TREM gene clusters in a case-control Alzheimer's disease data set. Ghani M, Sato C, Kakhki EG, Gibbs JR, **Traynor B**, St George-Hyslop P, Rogaeva E. *Neurobiol Aging*. 2016;42:217.e7-.e13. NIHMSID940294.
127. OPTN 691\_692insAG is a founder mutation causing recessive ALS and increased risk in heterozygotes. Goldstein O, Nayshool O, Nefussy B, **Traynor BJ**, Renton AE, Gana-Weisz M, Drory VE, Orr-Urtreger A. *Neurology*. 2016;86:446-53. PMC4773945.
128. Loss of VPS13C Function in Autosomal-Recessive Parkinsonism Causes Mitochondrial Dysfunction and Increases PINK1/Parkin-Dependent Mitophagy. Lesage S, Drouet V, Majounie E, Deramecourt V, Jacoupy M, Nicolas A, Cormier-Dequaire F, Hassoun SM, Pujol C, Ciura S, Erpapazoglou Z, Usenko T, Muraige CA, Sahbatou M, Liebau S, Ding J, Bilgic B, Emre M, Erginel-Unaltuna N, Guven G, Tison F, Tranchant C, Vidailhet M, Corvol JC, Krack P, Leutenegger AL, Nalls MA, Hernandez DG, Heutink P, Gibbs JR, Hardy J, Wood NW, Gasser T, Durr A, Deleuze JF, Tazir M, Destee A, Lohmann E, Kabashi E, Singleton A, Corti O, Brice A, French Parkinson's Disease Genetics Study (Pdg); **International Parkinson's Disease Genomics Consortium\*** (Ipdgc). *Am J Hum Genet*. 2016;98:500-13. PMC4800038.
129. A genome-wide association study in multiple system atrophy. Sailer A, Scholz SW, Nalls MA, Schulte C, Federoff M, Price TR, Lees A, Ross OA, Dickson DW, Mok K, Mencacci NE, Schottlaender L, Chelban V, Ling H, O'sullivan SS, Wood NW, **Traynor BJ**, Ferrucci L, Federoff HJ, Mhyre TR, Morris HR, Deuschl G, Quinn N, Widner H, Albanese A, Infante J, Bhatia KP, Poewe W, Oertel W, Hoglinger GU, Wullner U, Goldwurm S, Pellecchia MT, Ferreira J, Tolosa E, Bloem BR, Rascol O, Meissner WG, Hardy JA, Revesz T, Holton JL, Gasser T, Wenning GK, Singleton AB, Houlden H. *Neurology*. 2016;87:1591-8. PMC5067544.
130. Cortical hyperexcitability in patients with C9ORF72 mutations: Relationship to phenotype. Schanz O, Bageac D, Braun L, **Traynor BJ**, Lehky TJ, Floeter MK. *Muscle Nerve*. 2016;54:264-9. PMC4940214.
131. Use of Genetic Testing in Amyotrophic Lateral Sclerosis by Neurologists. Arthur KC, Doyle C, Chiò A, **Traynor BJ**. *JAMA Neurol*. 2017;74:125-6. NIHMSID940322.
132. C9orf72 hexanucleotide repeat expansions are not a common cause of obsessive-compulsive disorder. Arthur KC, Rivera AM, Samuels J, Wang Y, Grados M, Goes FS, Maher B, Nestadt G, **Traynor BJ**. *J Neurol Sci*. 2017;375:71-2. NIHMSID845348.
133. NeuroChip, an updated version of the NeuroX genotyping platform to rapidly screen for variants associated with neurological diseases. Blauwendraat C, Faghri F, Pihlstrom L, Geiger JT, Elbaz A, Lesage S, Corvol JC, May P, Nicolas A, Abramzon Y, Murphy NA, Gibbs JR, Ryten M, Ferrari R, Bras J, Guerreiro R, Williams J, Sims R, Lubbe S, Hernandez DG, Mok KY, Robak L, Campbell RH, Rogaeva E, **Traynor BJ**, Chia R, Chung SJ, Hardy JA, Brice A, Wood NW, Houlden H, Shulman JM, Morris HR, Gasser T, Kruger R, Heutink P, Sharma M, Simon-Sanchez J, Nalls MA, Singleton AB, Scholz SW. *Neurobiol Aging*. 2017;57:247.e9-.e13. PMC5534378.
134. Exome sequencing establishes a gelsolin mutation as the cause of inherited bulbar-onset neuropathy. Caress JB, Johnson JO, Abramzon YA, Hawkins GA, Gibbs JR, Sullivan EA, Chahal CS, **Traynor BJ**. *Muscle Nerve*. 2017;56:1001-5. PMC5494018.
135. A data-driven approach links microglia to pathology and prognosis in amyotrophic lateral sclerosis. Cooper-Knock J, Green C, Altschuler G, Wei W, Bury JJ, Heath PR, Wyles M, Gelsthorpe C, Highley JR, Lorente-Pons A, Beck T, Doyle K, Otero K, **Traynor B**, Kirby J, Shaw PJ, Hide W. *Acta Neuropathol Commun*. 2017;5:23. PMC5353945.
136. Transcriptomic profiling of the human brain reveals that altered synaptic gene expression is associated with chronological aging. Dillman AA, Majounie E, Ding J, Gibbs JR, Hernandez D, Arepalli S, **Traynor BJ**, Singleton AB, Galter D, Cookson MR. *Sci Rep*. 2017;7:16890. PMC5715102.
137. Disease progression in C9orf72 mutation carriers. Floeter MK, **Traynor BJ**, Farren J, Braun LE, Tierney M, Wiggs EA, Wu T. *Neurology*. 2017;89:234-41. PMC5513817.
138. No genetic association between attention-deficit/hyperactivity disorder (ADHD) and Parkinson's disease in nine ADHD candidate SNPs. Geissler JM, **International Parkinson Disease Genomics Consortium\***; Romanos M, Gerlach M, Berg D, Schulte C. *Atten Defic Hyperact Disord*. 2017;9:121-7.
139. Poly(GP) proteins are a useful pharmacodynamic marker for C9ORF72-associated amyotrophic lateral sclerosis. Gendron TF, Chew J, Stankowski JN, Hayes LR, Zhang YJ, Prudencio M, Carlomagno Y, Daugherty LM, Jansen-West K, Perkerson EA, O'raw A, Cook C, Pregent L, Belzil V, Van Blitterswijk M, Tabassian LJ, Lee CW, Yue M, Tong J, Song Y, Castanedes-Casey M, Rousseau L, Phillips V, Dickson DW, Rademakers R, Fryer JD, Rush BK, Pedraza O, Caputo AM, Desaro P, Palmucci C, Robertson A, Heckman MG, Diehl NN, Wiggs E, Tierney M, Braun L, Farren J, Lacomis D, Ladha S, Fournier CN, Mccluskey LF, Elman LB, Toledo JB, McBride JD, Tiloca C, Morelli C, Poletti B, Solca F, Prellè A, Wu J, Jockel-Balsarotti J, Rigo F, Ambrose C, Datta A, Yang W, Raitcheva D, Antognetti G, Mccampbell A, Van Swieten JC, Miller BL, Boxer AL, Brown RH, Bowser R, Miller

- TM, Trojanowski JQ, Grossman M, Berry JD, Hu WT, Ratti A, **Traynor BJ**, Disney MD, Benatar M, Silani V, Glass JD, Floeter MK, Rothstein JD, Boylan KB, Petrucelli L. *Sci Transl Med.* 2017;9:pii: eaai7866. PMC5576451.
140. High frequency of C9orf72 hexanucleotide repeat expansion in amyotrophic lateral sclerosis patients from two founder populations sharing the same risk haplotype. Goldstein O, Gana-Weisz M, Nefussy B, Vainer B, Nayshool O, Bar-Shira A, **Traynor BJ**, Drory VE, Orr-Urtreger A. *Neurobiol Aging.* 2017; [Epub ahead of print]. NIHMSID940312.
141. Genetic variants associated with physical performance and anthropometry in old age: a genome-wide association study in the iSIRENTE cohort. Heckerman D, **Traynor BJ**, Picca A, Calvani R, Marzetti E, Hernandez D, Nalls M, Arepali S, Ferrucci L, Landi F. *Sci Rep.* 2017;7:15879. PMC5696534.
142. Novel genetic loci associated with hippocampal volume. Hibar DP, Adams HHH, Jahanshad N, Chauhan G, Stein JL, Hofer E, Renteria ME, Bis JC, Arias-Vasquez A, Ikram MK, Desrivieres S, Vernooij MW, Abramovic L, Alhusaini S, Amin N, Andersson M, Arfanakis K, Aribisala BS, Armstrong NJ, Athanasiu L, Axelsson T, Beecham AH, Beiser A, Bernard M, Blanton SH, Bohlken MM, Boks MP, Bralten J, Brickman AM, Carmichael O, Chakravarty MM, Chen Q, Ching CRK, Chouraki V, Cuellar-Partida G, Crivello F, Den Braber A, Doan NT, Ehrlich S, Giddaluru S, Goldman AL, Gottesman RF, Grimm O, Griswold ME, Guadalupe T, Gutman BA, Hass J, Haukvik UK, Hoehn D, Holmes AJ, Hoogman M, Janowitz D, Jia T, Jorgensen KN, Karbalai N, Kasperaviciute D, Kim S, Klein M, Kraemer B, Lee PH, Liewald DCM, Lopez LM, Luciano M, Macare C, Marquand AF, Matarin M, Mather KA, Mattheisen M, McKay DR, Milaneschi Y, Munoz Maniega S, Nho K, Nugent AC, Nyquist P, Loohuis LMO, Oosterlaan J, Pappmeyer M, Pirpamer L, Putz B, Ramasamy A, Richards JS, Risacher SL, Roiz-Santianez R, Rommelse N, Ropele S, Rose EJ, Royle NA, Rundek T, Samann PG, Saremi A, Satizabal CL, Schmaal L, Schork AJ, Shen L, Shin J, Shumskaya E, Smith AV, Sprooten E, Strike LT, Teumer A, Tordesillas-Gutierrez D, Toro R, Trabzuni D, Trompet S, Vaidya D, Van Der Grond J, Van Der Lee SJ, Van Der Meer D, Van Donkelaar MMJ, Van Eijk KR, Van Erp TGM, Van Rooij D, Walton E, Westlye LT, Whelan CD, Windham BG, Winkler AM, Wittfeld K, Woldehawariat G, Wolf C, Wolfers T, Yanek LR, Yang J, Zijdenbos A, Zwiers MP, Agartz I, Almasy L, Ames D, Amouyel P, Andreassen OA, Arepalli S, Assareh AA, Barral S, Bastin ME, Becker DM, Becker JT, Bennett DA, Blangero J, Van Bokhoven H, Boomsma DI, Brodaty H, Brouwer RM, Brunner HG, Buckner RL, Buitelaar JK, Bulayeva KB, Cahn W, Calhoun VD, Cannon DM, Cavalleri GL, Cheng CY, Cichon S, Cookson MR, Corvin A, Crespo-Facorro B, Curran JE, Czisch M, Dale AM, Davies GE, De Craen AJM, De Geus EJC, De Jager PL, De Zubicaray GI, Deary IJ, Dobbie S, Decarli C, Delanty N, Depondt C, Destefano A, Dillman A, Djurovic S, Donohoe G, Drevets WC, Duggirala R, Dyer TD, Enzinger C, Erk S, Espeseth T, Fedko IO, Fernandez G, Ferrucci L, Fisher SE, Fleischman DA, Ford I, Fornage M, Foroud TM, Fox PT, Francks C, Fukunaga M, Gibbs JR, Glahn DC, Gollub RL, Goring HHH, Green RC, Gruber O, Gudnason V, Gueffi S, Haberg AK, Hansell NK, Hardy J, Hartman CA, Hashimoto R, Hegenscheid K, Heinz A, Le Hellard S, Hernandez DG, Heslenfeld DJ, Ho BC, Hoekstra PJ, Hoffmann W, Hofman A, Holsboer F, Homuth G, Hosten N, Hottenga JJ, Huentelman M, Hulshoff Pol HE, Ikeda M, Jack CR, Jr., Jenkinson M, Johnson R, Jonsson EG, Jukema JW, Kahn RS, Kanai R, Kloszewska I, Knopman DS, Kochunov P, Kwok JB, Lawrie SM, Lemaitre H, Liu X, Longo DL, Lopez OL, Lovestone S, Martinez O, Martinot JL, Mattay VS, McDonald C, Mcintosh AM, McMahon FJ, McMahon KL, Mecocci P, Melle I, Meyer-Lindenberg A, Mohnke S, Montgomery GW, Morris DW, Mosley TH, Muhleisen TW, Muller-Miyhok B, Nalls MA, Nauck M, Nichols TE, Niessen WJ, Nothen MM, Nyberg L, Ohi K, Olvera RL, Ophoff RA, Pandolfo M, Paus T, Pausova Z, Penninx B, Pike GB, Potkin SG, Psaty BM, Reppermund S, Rietschel M, Roffman JL, Romanczuk-Seiferth N, Rotter JJ, Ryten M, Sacco RL, Sachdev PS, Saykin AJ, Schmidt R, Schmidt H, Schofield PR, Sigursson S, Simmons A, Singleton A, Sisodiya SM, Smith C, Smoller JW, Soininen H, Steen VM, Stott DJ, Sussmann JE, Thalamuthu A, Toga AW, **Traynor BJ**, Troncoso J, Tsolaki M, Tzourio C, Uitterlinden AG, Hernandez MCV, Van Der Brug M, Van Der Lugt A, Van Der Wee NJA, Van Haren NEM, Van 'T Ent D, Van Tol MJ, Vardarajan BN, Vellas B, Veltman DJ, Volzke H, Walter H, Wardlaw JM, Wassink TH, Weale ME, Weinberger DR, Weiner MW, Wen W, Westman E, White T, Wong TY, Wright CB, Zielke RH, Zonderman AB, Martin NG, Van Duijn CM, Wright MJ, Longstreth WT, Schumann G, Grabe HJ, Franke B, Launer LJ, Medland SE, Seshadri S, Thompson PM, Ikram MA. *Nat Commun.* 2017;8:13624. PMC5253632.
143. Discovery and functional prioritization of Parkinson's disease candidate genes from large-scale whole exome sequencing. Jansen IE, Ye H, Heetveld S, Lechler MC, Michels H, Seinstra RI, Lubbe SJ, Drouet V, Lesage S, Majounie E, Gibbs JR, Nalls MA, Ryten M, Botia JA, Vandrovicova J, Simon-Sanchez J, Castillo-Lizardo M, Rizzu P, Blauwendraat C, Chouhan AK, Li Y, Yogi P, Amin N, Van Duijn CM, **International Parkinson's Disease Genetics Consortium\***, Morris HR, Brice A, Singleton AB, David DC, Nollen EA, Jain S, Shulman JM, Heutink P. *Genome Biol.* 2017;18:22. PMC5282828.
144. Tdp-43 cryptic exons are highly variable between cell types. Jeong YH, Ling JP, Lin SZ, Donde AN, Braunstein KE, Majounie E, **Traynor BJ**, Laclair KD, Lloyd TE, Wong PC. *Mol Neurodegener.* 2017;12:13. PMC5289002.

145. Age-related penetrance of the C9orf72 repeat expansion. Murphy NA, Arthur KC, Tienari PJ, Houlden H, Chiò A, **Traynor BJ**. *Sci Rep*. 2017;7:2116. PMC5437033.
146. Excessive burden of lysosomal storage disorder gene variants in Parkinson's disease. Robak LA, Jansen IE, Van Rooij J, Uitterlinden AG, Kraaij R, Jankovic J, **International Parkinson's Disease Genomics Consortium\***, Heutink P, Shulman JM. *Brain*. 2017;140:3191-203. NIHMSID940313.
147. C9ORF72 hexanucleotide repeat exerts toxicity in a stable, inducible motor neuronal cell model, which is rescued by partial depletion of Pten. Stopford MJ, Higginbottom A, Hautbergue GM, Cooper-Knock J, Mulcahy PJ, De Vos KJ, Renton AE, Pliner H, Calvo A, Chiò A, **Traynor BJ**, Azzouz M, Heath PR, Kirby J, Shaw PJ. *Hum Mol Genet*. 2017;26:1133-45. PMC5409131.
148. Common polymorphisms of chemokine (C-X3-C motif) receptor 1 gene modify amyotrophic lateral sclerosis outcome: A population-based study. Calvo A, Moglia C, Canosa A, Cammarosano S, Ilardi A, Bertuzzo D, **Traynor BJ**, Brunetti M, Barberis M, Mora G, Casale F, Chiò A. *Muscle Nerve*. 2018;57:212-6.
149. Alzheimer risk loci and associated neuropathology in a population-based study (Vantaa 85+). Makela M, Kaivola K, Valori M, Paetau A, Polvikoski T, Singleton AB, **Traynor BJ**, Stone DJ, Peuralinna T, Tienari PJ, Tanskanen M, Myllykangas L. *Neurol Genet*. 2018;4:e211. PMC5773846.
150. Melatonin receptor type 1A gene linked to Alzheimer's disease in old age. Sulkava S, Muggalla P, Sulkava R, Ollila HM, Peuralinna T, Myllykangas L, Kaivola K, Stone DJ, **Traynor BJ**, Renton AE, Rivera AM, Helisalmi S, Soinen H, Polvikoski T, Hiltunen M, Tienari PJ, Huttunen HJ, Paunio T. *Sleep*. 2018 Jul 1;41(7). PMC6047434
151. C9orf72 hexanucleotide repeat length in older population: normal variation and effects on cognition. Kaivola K, Kiviharju A, Jansson L, Rantalainen V, Eriksson JG, Strandberg TE, Laaksovirta H, Renton AE, **Traynor BJ**, Myllykangas L, Tienari PJ. *Neurobiol Aging*. 2019 Mar 11. pii: S0197-4580(19)30076-4.
152. Shared polygenic risk and causal inferences in amyotrophic lateral sclerosis. Bandres-Ciga S, Noyce AJ, Hemani G, Nicolas A, Calvo A, Mora G; ITALSGEN Consortium; International ALS Genomics Consortium, Tienari PJ, Stone DJ, Nalls MA, Singleton AB, Chiò A, **Traynor BJ**. *Ann Neurol*. 2019 Feb 5. [Epub ahead of print]
153. Oligogenic basis of sporadic ALS: The example of SOD1 p.Ala90Val mutation. *Neurol Genet*. Kuuluvainen L, Kaivola K, Mönkäre S, Laaksovirta H, Jokela M, Udd B, Valori M, Pasanen P, Paetau A, **Traynor BJ**, Stone DJ, Schleutker J, Pöyhönen M, Tienari PJ, Myllykangas L. 2019 Apr 23;5(3):e335.
154. Recurrent homozygous damaging mutation in TMX2 encoding a protein disulfide isomerase, in four families with microlissencephaly Ghosh SG, Wang L, Breuss MW, Green JD, Stanley V, Yang X, Ross D, **Traynor BJ**, Alhashem AM, Azam M, Selim L, Bastaki L, Elbastawisy HI, Temtamy S, Zaki M, Gleeson JG. *J Med Genet*. 2019 Oct 5.
155. Dominant mutations of the Notch ligand Jagged1 cause peripheral neuropathy. Sullivan JM, Motley WW, Johnson JO, Aisenberg WH, Marshall KL, Barwick KE, Kong L, Huh JS, Saavedra-Rivera PC, McEntagart MM, Marion MH, Hicklin LA, Modarres H, Baple EL, Farah MH, Zuberi AR, Lutz CM, Gaudet R, **Traynor BJ**, Crosby AH, Sumner CJ. *J Clin Invest*. 2020 Mar 2;130(3):1506-1512. PMC7269582.
156. Epidemiological evidence for a hereditary contribution to myasthenia gravis: a retrospective cohort study of patients from North America. Green JD, Barohn RJ, Bartoccion E, Benatar M, Blackmore D, Chaudhry V, Chopra M, Corse A, Dimachkie MM, Evoli A, Florence J, Freimer M, Howard JF, Jiwa T, Kaminski HJ, Kissel JT, Koopman WJ, Lipscomb B, Maestri M, Marino M, Massey JM, McVey A, Mezei MM, Muppidi S, Nicolle MW, Oger J, Pascuzzi RM, Pasnoor M, Pestronk A, Provenzano C, Ricciardi R, Richman DP, Rowin J, Sanders DB, Siddiqi Z, Soloway A, Wolfe GI, Wulf C, Drachman DB, **Traynor BJ**. *BMJ Open*. 2020 Sep 18;10(9):e037909. PMC7511637.
157. Identification of a pathogenic intronic KIF5A mutation in an ALS-FTD kindred. Saez-Atienzar S, Dalgard CL, Ding J, Chio A, Alba C, Hupalò DN, Wilkerson MD, Bowser R, Pioro EP, Bedlack R, **Traynor BJ**. *Neurology*. 2020 Dec; 95(22):1015-1018.
158. Mutational analysis of known ALS genes in an Italian population-based cohort. Grassano M, Calvo A, Moglia C, Brunetti M, Barberis M, Sbaiz L, Canosa A, Manera U, Vasta R, Corrado L, D'Alfonso S, Mazzini L, Scholz SW, Dalgard C, Ding J, Gibbs RJ, Chia R, **Traynor BJ**, Chiò A; American Genomic Center. *Neurology*. 2021 Jan; 96(4):e600-e609.

\*member of the consortium

### Review Articles

1. Hardiman O, **Traynor BJ**, Corr B, Frost E. Models of care for motor neuron disease: setting standards. *Amyotroph Lateral Scler Other Motor Neuron Disord*. 2002;3:182-5.
2. Nogueira RG, **Traynor BJ**. The neurology of varicella-zoster virus: a historical perspective. *Arch Neurol*. 2004;61:1974-7.
3. Beghi E, Logroscino G, Chiò A, Hardiman O, Mitchell D, Swingler R, **Traynor BJ**. The epidemiology of ALS

- and the role of population-based registries. *Biochim Biophys Acta*. 2006;1762:1150-7.
4. Hardy J, Momeni P, **Traynor BJ**. Frontal temporal dementia: dissecting the aetiology and pathogenesis. *Brain*. 2006;129:830-1.
  5. Bedlack RS, **Traynor BJ**, Cudkovicz ME. Emerging disease-modifying therapies for the treatment of motor neuron disease/amyotrophic lateral sclerosis. *Expert Opin Emerg Drugs*. 2007;12:229-52.
  6. Beghi E, Mennini T, Bendotti C, Bigini P, Logroscino G, Chiò A, Hardiman O, Mitchell D, Swingler R, **Traynor BJ**, Al-Chalabi A. The heterogeneity of amyotrophic lateral sclerosis: a possible explanation of treatment failure. *Curr Med Chem*. 2007;14:3185-200.
  7. Gwinn K, Corriveau RA, Mitsumoto H, Bednarz K, Brown RH, Jr., Cudkovicz M, Gordon PH, Hardy J, Kasarskis EJ, Kaufmann P, Miller R, Sorenson E, Tandan R, **Traynor BJ**, Nash J, Sherman A, Mailman MD, Ostell J, Bruijn L, Cwik V, Rich SS, Singleton A, Refolo L, Andrews J, Zhang R, Conwit R, Keller MA. Amyotrophic lateral sclerosis: an emerging era of collaborative gene discovery. *PLoS One*. 2007;2:e1254. PMC2100166.
  8. Schymick JC, Talbot K, **Traynor BJ**. Genetics of sporadic amyotrophic lateral sclerosis. *Hum Mol Genet*. 2007;16 Spec No. 2:R233-42.
  9. Logroscino G, **Traynor BJ**, Hardiman O, Chiò A, Couratier P, Mitchell JD, Swingler RJ, Beghi E. Descriptive epidemiology of amyotrophic lateral sclerosis: new evidence and unsolved issues. *J Neurol Neurosurg Psychiatry*. 2008;79:6-11.
  10. Ravits J, **Traynor BJ**. Current and future directions in genomics of amyotrophic lateral sclerosis.. *Phys Med Rehabil Clin N Am*. 2008;19:461-77, viii. PMC3524513.
  11. Chiò A, Logroscino G, Hardiman O, Swingler R, Mitchell D, Beghi E, **Traynor B**. Prognostic factors in ALS: A critical review. *Amyotroph Lateral Scler*. 2009;10:310-23. PMC3515205.
  12. **Traynor BJ**. The era of genomic epidemiology. *Neuroepidemiology*. 2009;33:276-9. PMC2826447.
  13. Singleton AB, Hardy J, **Traynor BJ**, Houlden H. Towards a complete resolution of the genetic architecture of disease. *Trends Genet*. 2010;26:438-42. PMC2943029.
  14. **Traynor BJ**, Singleton AB. Nature versus nurture: death of a dogma, and the road ahead. *Neuron*. 2010;68:196-200. PMC2974322.
  15. Beghi E, Chiò A, Couratier P, Esteban J, Hardiman O, Logroscino G, Millul A, Mitchell D, Preux PM, Pupillo E, Stevic Z, Swingler R, **Traynor BJ**, Van Den Berg LH, Veldink JH, Zoccolella S. The epidemiology and treatment of ALS: focus on the heterogeneity of the disease and critical appraisal of therapeutic trials.. *Amyotroph Lateral Scler*. 2011;12:1-10. PMC3513399.
  16. Schymick JC, **Traynor BJ**. Expanding the genetics of amyotrophic lateral sclerosis and frontotemporal dementia. *Alzheimers Res Ther*. 2012;4:30. PMC3506944.
  17. Controversies and priorities in amyotrophic lateral sclerosis. Turner MR, Hardiman O, Benatar M, Brooks BR, Chiò A, De Carvalho M, Ince PG, Lin C, Miller RG, Mitsumoto H, Nicholson G, Ravits J, Shaw PJ, Swash M, Talbot K, **Traynor BJ**, Van Den Berg LH, Veldink JH, Vucic S, Kiernan MC. *Lancet Neurol*. 2013;12:310-22. PMC4565161.
  18. Searching for Grendel: origin and global spread of the C9ORF72 repeat expansion. Pliner HA, Mann DM, **Traynor BJ**. *Acta Neuropathol*. 2014;127:391-6. PMC4545603.
  19. Renton AE, Chiò A, **Traynor BJ**. State of play in amyotrophic lateral sclerosis genetics. *Nat Neurosci*. 2014;17:17-23. PMC4544832.
  20. **Traynor BJ**. A roadmap for genetic testing in ALS. *J Neurol Neurosurg Psychiatry*. 2014;85:476. PMC4568817.
  21. Chiò A, **Traynor BJ**. Motor neuron disease in 2014. Biomarkers for ALS--in search of the Promised Land. *Nat Rev Neurol*. 2015;11:72-4. PMC5185468.
  22. Marangi G, **Traynor BJ**. Genetic causes of amyotrophic lateral sclerosis: new genetic analysis methodologies entailing new opportunities and challenges. *Brain Res*. 2015;1607:75-93. PMC5916786.
  23. Singleton AB, **Traynor BJ**. Genetics. For complex disease genetics, collaboration drives progress. *Science*. 2015;347:1422-3. PMC5810558.
  24. Bradley WG, Andrew AS, **Traynor BJ**, Chiò A, Butt TH, Stommel EW. Gene-Environment-Time Interactions in Neurodegenerative Diseases: Hypotheses and Research Approaches. *Ann Neurosci*. 2018 Dec;25(4):261-267. PMC6470336.
  25. Chia R, Chiò A, **Traynor BJ**. Novel genes associated with amyotrophic lateral sclerosis: diagnostic and clinical implications. *Lancet Neurol*. 2018;17:94-102. PMC5901717.
  26. The Overlapping Genetics of Amyotrophic Lateral Sclerosis and Frontotemporal Dementia. Abramzon YA, Fratta P, **Traynor BJ**, Chia R. *Front Neurosci*. 2020 Feb 5;14:42. PMC7012787.
  27. The NGS technology for the identification of genes associated with the ALS. A systematic review. Pecoraro V, Mandrioli J, Carone C, Chiò A, Traynor BJ, Trenti T. *Eur J Clin Invest*. 2020 May;50(5):e13228.



28. C9orf72 Hexanucleotide Repeat in Huntington-Like Patients: Systematic Review and Meta-Analysis. Alva-Diaz C, Alarcon-Ruiz CA, Pacheco-Barrios K, Mori N, Pacheco-Mendoza J, **Traynor BJ**, Rivera-Valdivia A, Lertwilaiwittaya P, Bird TD, Cornejo-Olivas M. *Front Genet.* 2020 Nov 2;11:551780.

#### Editorials

1. **Traynor BJ**, Singleton A. Genome-wide association studies and ALS: are we there yet? *Lancet Neurol.* 2007;6:841-3.
2. **Traynor BJ**, Singleton AB. What's the FUS! *Lancet Neurol.* 2009;8:418-9. NIHMSID431035.
3. **Traynor BJ**. Road to the chromosome 9p-linked ALS/FTD locus. *J Neurol Neurosurg Psychiatry.* 2012;83:356-7. PMC3513280.
4. Renton AE, **Traynor BJ**. CRESTing the ALS mountain. *Nat Neurosci.* 2013;16:774-5. PMC4565517.
5. **Traynor BJ**, Cleveland DW. Special Issue on amyotrophic lateral sclerosis. *Exp Neurol.* 2014;262 Pt B:73-4. PMC4559336.
6. **Traynor BJ**, Angelini C. [An] enumeration shall be made. *Neurology.* 2015;85:1191-2. NIHMSID940478.
7. **Traynor BJ**, Renton AE. Exploring the epigenetics of Alzheimer disease. *JAMA Neurol.* 2015;72:8-9. NIHMSID940621.
8. **Traynor BJ**, Abramzon YA. To Dement or Not to Dement, That Is the Question. *JAMA Neurol.* 2016;73:383-4. NIHMSID940630.
9. **Traynor BJ**, Rademakers R. Dementia Research-A Roadmap for the Next Decade. *JAMA Neurol.* 2017;74:141-2. NIHMSID940136.
10. Armon C and **Traynor BJ**. High BMI is associated with low ALS risk: What does it mean? *Neurology.* 2019 In Press.
11. Bandres-Ciga S, Noyce AJ, **Traynor BJ**. Mendelian Randomization - A Journey From Obscurity to Center Stage With a Few Potholes Along the Way. *JAMA Neurol.* 2019 Oct 14. PMID: 31609383.
12. Dewan R, **Traynor BJ**. Plasma microRNA signature as biomarker for disease progression in frontotemporal dementia and amyotrophic lateral sclerosis. *J Neurol Neurosurg Psychiatry.* 2021 May;92(5):458. PMID: 33722821.
13. Chiò A, Calvo A, **Traynor BJ**. Nature meets nurture in amyotrophic lateral sclerosis. *Lancet Neurol.* 2021 May; 20(5):332-333. PMID: 33894186.
14. **Traynor BJ** and Gottesman RF. The value of studying rare genetic variants and other emerging themes in neurogenetics. *Brain.* 2021. In press.

#### Book Chapters

1. Hardy J, Momeni P, Myers A, and **Traynor BJ** (2006). The Genetics of Frontotemporal Dementia, in *Dementia and Motor Neuron Disease*, Strong MJ, Editor. 2006, Boca Raton: CRC Press: 201–207
2. **Traynor BJ**, and Singleton A (2008). The HapMap Project. *Encyclopedia of Life Sciences* (www.els.net); and in *Handbook of Human Molecular Evolution*. New York: Wiley, John & Sons, Inc.
3. Huey ED, Hardy J, Scholz SW, **Traynor BJ**, Small SA. Neurobiology of Dementia and Other Neurodegenerative Disorders, in *Tasman's Psychiatry*, fifth edition, Tasman A, Riba MB, Alarcon RD, Alfonso CA, Kanba S, Ndeti DM, Ng CH, Schulze TG, Lecic-Tosevski D, Editors. 2023, Cham: Springer Nature Switzerland.

#### Letters, Correspondence

1. **Traynor BJ**, Hardiman O. Status epilepticus. *N Engl J Med.* 1998;339:410.
2. Logroscino G, Beghi E, Hardiman O, Chiò A, Mitchell JD, Swingler RJ, **Traynor B**. Effect of referral bias on assessing survival in ALS. *Neurology.* 2007;69:939.

#### Proceedings Reports

1. 148th ENMC international workshop on the scientific contributions of the EURALS consortium on amyotrophic lateral sclerosis. Beghi E, Pupillo E, Zoccollella S. *Neuromuscul Disord.* 2009;19:379-81. NIHMSID 940286.

#### Media Releases or Interviews

1. Ruth Williams. Bryan Traynor: serious science from a funny fellow. *Lancet Neurology.* 2019; 18:720 ([https://doi.org/10.1016/S1474-4422\(18\)30286-2](https://doi.org/10.1016/S1474-4422(18)30286-2))

Case Reports                      None

Books, Textbooks                None



## FUNDING

Summary: \$14.3M in Extramural and Intramural Principal Investigator funding since 2005

### EXTRAMURAL FUNDING

#### Current

2019 – 2022      Establishing a Central Repository for Whole Genome Sequencing of ALS Patients  
 Funded by the ALS Association  
 \$3,327,053  
 Principal investigator: John Landers, Ph.D.  
 Role: Co-investigator

#### Previous

2002 – 2004      Role of start codon mutations in the pathogenesis of ALS  
 Funded by the Clinical Investigator Training program.  
 \$114,000  
 Role: Co-investigator

2004 – 2005      Role of start codon mutations in the pathogenesis of ALS  
 Funded by the Muscular Dystrophy Association.  
 \$90,000  
 Role: Co-investigator

2004 – 2005      Genetic epidemiology of ALS in Ireland  
 Funded by the ALS Association.  
 \$160,000  
 Role: Co-investigator

2006 – 2007      Genome wide association study of sporadic ALS  
 Funded by the ALS Association  
 \$450,000  
 Role: Principal Investigator

2006 – 2007      Genome wide association study of sporadic ALS  
 Funded by the Robert Packard Center for ALS Research  
 \$200,000  
 Role: Principal Investigator

2007 – 2008      Follow-up genetic association study of sporadic ALS using the Illumina iSelect custom SNP  
 chip  
 Funded by the ALS Association.  
 \$136,375  
 Role: Principal Investigator

2007 – 2008      Follow-up genetic association study of sporadic ALS using the Illumina iSelect custom SNP  
 chip  
 Funded by the Robert Packard Center for ALS Research.  
 \$142,481  
 Role: Principal Investigator

2007 – 2009      A population-based survey of cognitive decline in ALS  
 Funded by the ALS Association. \$184,000  
 Role: Co-principal investigator

2008 – 2011      An International Case-Control Study on Genetic and Environmental Risk Factors  
 Funded by the ALS Association  
 \$240,000  
 Role: Co-investigator (EURALS)

2008 – 2009      A genome wide association study of ALS in the Finnish population  
 Funded by Microsoft Research  
 \$250,000  
 Role: Principal Investigator

2008 – 2009	A genome wide association study of ALS in the Finnish population Funded by the ALS Association \$85,000 Role: Principal Investigator
2008 – 2009	A map of the epigenome in human spinal cord Funded by the Robert Packard Center for ALS Research \$160,000 Role: Principal Investigator
2008 – 2009	Whole genome association study: ilSirente cohort of persons aged 80 years and older Funded by Microsoft Research \$216,000 Role: Principal Investigator
2009 – 2011	Whole genome association study of myasthenia gravis Funded by The Myasthenia Gravis Foundation \$362,000 Role: Principal Investigator
2010 – 2011	ALS gene discovery using exome sequencing Funded by the Robert Packard Center for ALS Research \$50,000 Role: Principal Investigator
2011 – 2012	Identification of familial ALS genes by exome sequencing Funded by the ALS Association \$100,000 Role: Principal Investigator
2009 – 2011	Whole genome association study of myasthenia gravis Funded by The Myasthenia Gravis Foundation \$243,000 Role: Principal Investigator
2011 – 2012	Genetic studies of ALS Funded by the Italian Football Federation (FIGC), €300,000 Role: Principal Investigator
2012 – 2013	Large scale genome-wide association study of amyotrophic lateral sclerosis Funded by the Center for Disease Control and Prevention \$500,000 Role: Principal Investigator
2013 – 2014	Exome sequencing of 1000 sporadic ALS patients Funded by the Muscular Dystrophy Association \$400,000 Role: Principal Investigator
2014	Whole genome sequencing of Finnish ALS cases and controls Funded by Merck Inc. \$1,300,000 Role: Principal Investigator
2015	Cloud-based analysis of exome sequencing data Funded by Microsoft Research \$20,000 Role: Principal Investigator
2015	Replication of genome-wide association hits Funded by Merck Inc. \$750,000 Role: Principal Investigator
2016 – 2020	Genome-wide association study of myasthenia gravis Funded by The Myasthenia Gravis Foundation \$110,000 Role: Principal Investigator
2019 – 2020	Replication of genome-wide association hits Funded by Merck Inc.

\$250,000  
Role: Principal Investigator

## INTRAMURAL FUNDING

### Previous

2002 – 2004	Role of start codon mutations in the pathogenesis of ALS Funded by the Clinical Investigator Training program. \$114,000 Role: Co-investigator
2006 – 2007	Genome wide association study of sporadic ALS Funded by NINDS intramural funds \$343,920 Role: Principal Investigator
2017 – 2020	Genome sequencing of Lewy body dementia and frontotemporal dementia Funded by US Congressional funds/National Institute on Aging \$8,021,290 Role: Principal Investigator

## CLINICAL ACTIVITIES

### Clinical Focus

I am extensively involved in the management of amyotrophic lateral sclerosis and frontotemporal dementia. . . . .  
I am the PI of the Genetic Characterization of Movement Disorders and Dementias (03-AG-N329) protocol and the lead investigator of the Natural history and biomarkers of C9ORF72 amyotrophic lateral sclerosis and frontotemporal dementia (13-N-0188) protocol.

### Certification

#### Medical, other state/government licensure

2005 – present      Medical license, Maryland Board of Physicians (#D0063874); renewed 2019

#### Clinical (Service) Responsibilities

2005 – present      Neurologist, NIH Clinical Center, Bethesda, MD, 10%  
2009 – present      Neurologist, La Clinica del Pueblo, Washington DC, official duty, 5%

## EDUCATIONAL ACTIVITIES

### Educational Focus

My educational focus is on amyotrophic lateral sclerosis and frontotemporal dementia and the genetic causes and clinical aspects of these fatal neurodegenerative diseases.

### Teaching

#### Classroom instruction

#### JHMI/Regional

2002 – 2003	Laboratory instructor, Human Nervous System and Behavior Course, Harvard Medical School, Boston, MA, USA.
2003	Invited lecturer, Neuroepidemiology Course, Harvard School of Public Health, Boston, MA, USA.
2007	Invited lecturer, Neuroscience Graduate Program Lecture Series, National Naval Medical Center, Bethesda, MD.
2008 – 2014	Invited lecturer, Hopkins Aging, Cognition and Neurodegenerative Disorders Training Grant Graduate Course, Baltimore, MD.

- 2012 – present Invited lecturer, Hopkins Diseases and Disorders of the Nervous System Graduate Course, Baltimore, MD.  
 2020 Invited lecturer, Hopkins Course on Circuits and Brain Disorders, Baltimore, MD.

#### National

- 2013 Invited lecturer, Genetics Training Annual Retreat, University of Michigan, MI.

#### International

- 2012 – 2014 Lecturer, Molecular Neurology and Therapeutics course, Wellcome Trust Scientific Conference, Cambridge UK.  
 2020 Co-chair and organizer, “Genomics of Personalized Medicine” plenary session, 145<sup>th</sup> annual meeting of the American Neurological Association

#### Mentoring

##### Pre-doctoral Advisees / Mentees

- 2005 – 2009 Jennifer Schymick, M.D., Ph.D.; Graduate Student, Laboratory of Neurogenetics, NIA;  
 Current location: Fellow, Genetics, Stanford University, CA, USA  
 2007 Stephen Berger; Summer Post-Bac IRTA, Laboratory of Neurogenetics, NIA;  
 Current location: Graduate student, Johns Hopkins University, Baltimore, MD, USA  
 2008 – 2009 Terhi Peuralinna, PhD; Visiting Scientist, Laboratory of Neurogenetics, NIA  
 Current location: Fellow, University of Helsinki, Finland  
 2016 – present Yevgeniya Abramzon, B.S.; Graduate student at UCL, Laboratory of Neurogenetics, NIA  
 2012 – 2014 Hannah Pliner, M.A.; Post-Bac IRTA, Laboratory of Neurogenetics, NIA;  
 Current location: Graduate student, University of Washington, Seattle, WA.  
 2015 – 2016 Karissa Arthur, M.D., Medical Research Scholars Program, Laboratory of Neurogenetics, NIA  
 Current location: Neurology Resident, Johns Hopkins Hospital, Baltimore, MD  
 2014 – 2016 Alberto Rivera, B.S.; Post-Bac IRTA, Laboratory of Neurogenetics, NIA  
 Current location: Graduate student, University of Washington, Seattle, WA  
 2016 – 2018 Natalie Murphy, B.S.; Post-Bac IRTA, Laboratory of Neurogenetics, NIA  
 Current location: graduate student, Johns Hopkins University, Baltimore, MD

##### Post-doctoral Advisees / Mentees

- 2011 – 2015 Alan Renton, Ph.D.; Post-Doctoral fellow, Laboratory of Neurogenetics, NIA  
 Current location: Instructor, Mount Sinai Medical Center, New York, NY  
 2012 – 2013 Giuseppe Marangi, M.D.; Research Fellow, Laboratory of Neurogenetics, NIA  
 Current location: Faculty position, Catholic University, Rome, Italy  
 2015 – 2016 Karissa Arthur, M.D., Medical Research Scholars Program, Laboratory of Neurogenetics, NIA  
 Current location: Neurology Resident, Johns Hopkins Hospital, Baltimore, MD

##### Thesis committees

- 2017 Lanier Heyburn, Ph.D., “TDP-43 pathology: Elucidation of mechanisms and treatment with tyrosine kinase inhibition”, committee member, Georgetown University, DC, USA  
 2020 Maurizio Grassano, Ph.D., “Implementation of an integrate framework to interpret genetic variants in Amyotrophic Lateral Sclerosis”, committee member, University of Turin, Italy.

## RESEARCH ACTIVITIES

### Research Focus

Amyotrophic lateral sclerosis  
 Frontotemporal dementia  
 Myasthenia gravis  
 Neurogenetics  
 Epidemiology  
 Gene therapy

### Inventions, Patents, Copyrights

2011 US, Canadian, and European patent on the clinical testing for the hexanucleotide repeat expansion of C9orf72

## ORGANIZATIONAL ACTIVITIES

### Institutional Administrative Appointments

2010 – 2012 Member, Skin, Bone, Muscle, and Joint Working Group, PhenX Project, NHGRI  
 2010 Chairman, Genetics Subgroup Committee for the NINDS Common Data Elements Initiative  
 2010 Clinical advisor to the Genetic Testing Registry, NIH  
 2012 – 2015 Member, Earl Stadtman Tenure Track Search Committee, NIH  
 2013 ad hoc reviewer, NIA Promotion and Tenure Committee  
 2013 Panel member, NINDS Repository Scientific Liaison meeting  
 2016 Chair, Earl Stadtman Tenure Track Search Committee for Bioinformatics, NIH  
 2016 – 2020 Member, NIA Promotion and Tenure Committee  
 2017 – present Member, Medical Research Scholars Program Advisors Committee  
 2020 – present Member, NIH Gene Therapy Taskforce

### Editorial Activities

#### Editorial Board appointments

2013 – present Editorial board, *Journal of Neurology, Neurosurgery & Psychiatry*  
 2013 – 2014 Guest editor, special issue of *Experimental Neurology* on ALS genetics and pathogenesis  
 2015 – present Editorial board, *Neurobiology of Aging*  
 2017 – 2021 Editorial board, *JAMA Neurology*  
 2021 – Editorial board, *eClinical Medicine*  
 2021 – Associate editor, *Brain*

#### Journal Peer review activities

Acta Neuropathologica  
 American Journal of Human Genetics  
 American Journal of Psychiatry  
 Annals of Neurology  
 Annals of Human Genetics  
 BMC Neurology  
 Brain  
 Brain Research  
 Cell  
 Chest  
 Cochrane Library  
 eLife  
 European Journal of Epidemiology  
 European Journal of Neurology  
 European Journal of Neuroscience  
 Expert opinion on Investigational Drugs  
 Experimental Neurology  
 Genome Research  
 Glia  
 Human Genetics  
 Human Molecular Genetics  
 Human Mutation  
 JAMA  
 JAMA Neurology  
 Journal of Clinical Investigation  
 Journal of Neurochemistry

Journal of Neurology  
 Journal of Neurology, Neurosurgery and Psychiatry  
 Lancet  
 Lancet Neurology  
 Medical Journal of Australia  
 Molecular Basis of Disease  
 Muscle and Nerve  
 Nature Communications  
 Nature Genetics  
 Nature Medicine  
 Nature Reviews Genetics  
 Nature Neuroscience  
 Nature Scientific Reports  
 Neurobiology of Aging  
 Neuroepidemiology  
 NeuroMolecular Medicine  
 Neurology  
 Neuron  
 New England Journal of Medicine  
 PLoS Genetics  
 PLoS ONE  
 PNAS  
 Science

#### Advisory Committees, Review Groups / Study Sections

2004 – 2010 Steering committee member, the European ALS Epidemiological Consortium (EURALS)  
 2009 – 2010 Panel member, Annual Meeting on the National ALS Registry, Agency for Toxic Substances and Disease Registry, Center for Disease Control and Prevention  
 2010 – 2020 Member, Integration Panel for the Congressionally Mandated Department of Defense Amyotrophic Lateral Sclerosis Research Program  
 2015 – 2019 Chair, Integration Panel for the Congressionally Mandated Department of Defense Amyotrophic Lateral Sclerosis Research Program  
 2012 – 2015 Member, Integration Panel for the Congressionally Mandated Department of Defense Alzheimer's Disease Research Program  
 2015 – 2021 Member, Genetics of Health and Disease NIH Study Section  
 2020 – 2023 Member, Scientific Program Advisory Committee, American Neurological Association

## RECOGNITION

#### Awards, Honors, and Accomplishments

1993 Graduated from medical school ranked second in class, University College Dublin  
 1993 Sean Malone Medal and Francis McLaughlin Prize in Psychiatry, presented by the University College Dublin  
 2001 Medal for best published journal article in Neurology, presented by the Royal Academy of Medicine of Ireland  
 2002 – 2004 Full scholarship for the Clinical Investigator Training Program, Harvard University and Massachusetts Institute of Technology  
 2004 – present Member, Harvard Alumni Association  
 2004 – present Member, Massachusetts Institute of Technology Alumni Association  
 2007 Donald M. Palatucci Advocacy Leadership Forum, presented by the American Academy of Neurology  
 2011 NIA Director's Award, presented by the National Institute on Aging  
 2012 John W. Griffin Innovator Award, presented by the Packard Center for ALS at Hopkins  
 2012 Elected Fellow of the American Neurological Association  
 2012 Derek Denny-Brown Neurological Scholar Award, presented by the American



	Neurological Association
2012	<u>NIH Director's Award</u> , presented by the National Institutes of Health
2013	<u>Sheila Essey Award for ALS Research</u> , presented by the American Academy of Neurology
2013	Diamond Award for ALS Research, presented by the Muscular Dystrophy Association
2016	<u>Potamkin Prize for Dementia</u> , presented by the American Academy of Neurology
2016 – 2020	Highly Cited Researcher, presented by Thomson Reuters/Clarivates
2018	Healthcare and Life Sciences 50 Honoree, Presented by <i>Irish America</i>
2018	Profiled by <i>The Lancet Neurology</i>
2019	Nominated for membership of Sigma Xi
2020	Certification, Leadership principles for Scientists, Engineers, and Researchers, MIT xPRO
2020	Elected Fellow of the Royal College of Physicians of Ireland ( <u>FRCPi</u> )
2021	Elected Fellow of the Royal College of Physicians ( <u>FRCP</u> )

## Invited Talks

### JHMI/Regional

1999	Invited lecture, National Center for Medical Genetics of Ireland, Crumlin Hospital, Dublin, "Role of mutations in the Cu/Zn superoxide dismutase gene in the pathogenesis of amyotrophic lateral sclerosis in Ireland"
2003	Grand Rounds, Neurology Department, Boston University Medical Center, Boston, MA, "Epidemiology and clinical features of amyotrophic lateral sclerosis"
2004	Invited lecture, Harvard School of Public Health, Boston, MA, "Epidemiology of amyotrophic lateral sclerosis"
2005	Invited lecture, ALS Association National Advocacy Day, Washington DC, "Genetics of amyotrophic lateral sclerosis"
2005	Invited lecture, National Institutes of Health, Bethesda, MD, "Genetics and epidemiology of amyotrophic lateral sclerosis"
2005	Invited lecture, Robert Packard Center for ALS Research at Johns Hopkins, Baltimore, MD, "Genome wide association study of sporadic amyotrophic lateral sclerosis"
2006	Invited lecture, ALS Association Regional meeting, Fairfax, VA, "Advances in the genetics of amyotrophic lateral sclerosis"
2006	Invited lecture, ALS Association Regional meeting, Virginia Beach, VA, "Genetics of amyotrophic lateral sclerosis"
2006	Invited lecture, National Naval Medical Center, Bethesda, MD, "Genetics of amyotrophic lateral sclerosis"
2006	Invited lecture, Robert Packard Center for ALS Research at Johns Hopkins, Baltimore, MD, "Genome wide association of sporadic amyotrophic lateral sclerosis: an update"
2007	Invited lecture, ALS Association National Advocacy Day, Washington DC, "Advances in the genetics of amyotrophic lateral sclerosis"
2009	Grand Rounds, George Washington University Medical Center, Washington DC, "Genetics of amyotrophic lateral sclerosis"
2009	Invited lecture, Packard Center Annual Meeting, Baltimore, MD, "Genetic control of DNA methylation and expression in human brain"
2010	Invited lecture, National Institute on Neurological Disorders and Stroke, Bethesda, MD, "Advances in our understanding of the genetics of amyotrophic lateral sclerosis"
2010	Invited lecture, Packard Center Annual Meeting, Baltimore, MD, "Exome sequencing of familial ALS cases"
2010	Invited lecture, Thomas Jefferson University, Philadelphia, PA, "Genomics of amyotrophic lateral sclerosis"
2011	Invited lecture, Genetics of Neurodegenerative Disease – Neurochip and Beyond Workshop, "Genome wide association studies of ALS"
2011	Grand Rounds, George Washington University Medical Center, Washington DC, "Genomics of amyotrophic lateral sclerosis"
2012	Grand Rounds, National Institute on Neurological Disorders and Stroke, Bethesda, MD, "Genomics of ALS"
2012	Invited lecture, The Fifth Donald L Price Symposium, Baltimore, MD, "Hexanucleotide expansion repeat of C9ORF72 in ALS/FTD"

- 2013 Invited lecture, Alzheimer Disease Research Center (ADRC) Annual Conference on Aging and Dementia, Johns Hopkins, Baltimore, MD, "Unraveling neuronal selectivity (or lack thereof) in ALS & FTD using genomics"
- 2013 Invited lecture, Alzheimer Disease-Related Dementias Workshop: Research Challenges and Opportunities, Bethesda, MD, "Genetics of TDP43 and FUS biology"
- 2013 Invited lecture, Annual meeting of the Packard Center for ALS Research, Baltimore, MD, "Genetics of ALS. Quo vadis?"
- 2014 Invited lecture, American Neurological Association Annual Conference, Baltimore, MD, "C9ORF72: A tale of two neurodegenerative diseases"
- 2014 Invited lecture, Clinical Center Bethesda, MD, "ALS in the genomics age: Facts, uncertainties and the way forward"
- 2014 Invited lecture, Johns Hopkins Neurology, Baltimore, MD, "Genomics of ALS"
- 2014 Invited lecture, Research In Progress, NIA, Baltimore, MD, "Genomics of ALS"
- 2014 Invited lecture, Society for Neuroscience Annual Conference, Washington DC, "Genetics of C9orf72-related ALS and FTD"
- 2015 Invited lecture, Muscular Dystrophy Association Annual Conference, Washington DC, "Genetics of amyotrophic lateral sclerosis"
- 2016 Invited lecture, Uniformed Services University of the Health Sciences, Bethesda, MD, "Genomics of ALS"
- 2017 Grand Rounds, Georgetown University, Washington DC, "Expanding the genetics of amyotrophic lateral sclerosis"
- 2017 Invited lecture, NIH Research Festival, Bethesda, MD, "What genomics is telling us about the clinical phenotype of neurodegenerative diseases"
- 2018 Invited lecture, Demystifying medicine lecture series, NIH, Bethesda, MD, "Genomics of ALS"
- 2019 Invited lecture, Demystifying medicine lecture series, NIH, Bethesda, MD, "Dementia in the Genomic Era: Current Understanding of Frontotemporal Dementia"
- 2019 Invited lecture, Neurosurgery NIH, Bethesda, MD, "Genomics of ALS"
- 2019 Invited lecture, Packard Center for ALS Research, Baltimore, MD, "Using genomic data to define lifestyle factors for ALS"
- 2021 NIH Clinical Center Grand Rounds, "Using genetics to unravel the mysteries of ALS"
- National**
- 2005 Invited lecture, Drug company workgroup, ALS Association, Miami, FL, "Drug therapies in amyotrophic lateral sclerosis"
- 2005 Invited lecture, North East ALS Consortium, Miami, FL, "Systematic evaluation of potential therapeutic agents for clinical trials in amyotrophic lateral sclerosis"
- 2006 Invited lecture, 5th International conference on Frontotemporal Dementia, San Francisco, CA, "Genetics of frontotemporal dementia and amyotrophic lateral sclerosis"
- 2006 Invited lecture, ALS Association Regional meeting, NJ, "Advances in the genetics of amyotrophic lateral sclerosis"
- 2007 Grand Rounds, Baylor College of Medicine, Houston, TX, "Genetics of frontotemporal dementia and amyotrophic lateral sclerosis"
- 2007 Invited lecture, Workshop in Genetics, Microsoft Research, Bellevue, WA, "Whole genome association studies: methodology and pitfalls"
- 2008 Invited lecture, Neurodegenerative Diseases: Biology and Therapeutics Meeting, Cold Spring Harbor, NY, "Genetics of sporadic amyotrophic lateral sclerosis"
- 2008 Invited lecture, Microsoft eScience Workshop, Indianapolis, IN, "What do scientists really need to facilitate time to discovery?"
- 2009 Invited lecture, Microsoft eScience Workshop, Redmond, WA, "Advances in Genomics"
- 2010 Invited lecture, Neurodegenerative Diseases: Biology and Therapeutics Meeting, Cold Spring Harbor, NY, "Genome-wide association studies and beyond"
- 2010 Invited lecture, American Neurological Association 135th Annual Meeting, Neuromuscular Special Interest Group, San Francisco, CA, "Exome sequencing identifies VCP as a cause of familial ALS"
- 2010 Invited lecture, International ALS Symposium, Orlando, FL, "Exome sequencing reveals

- VCP as a cause of familial ALS"
- 2010 Invited lecture, Microsoft External Research Symposium, Redmond, WA, "Genome-wide association study of ALS in Finland"
- 2010 Invited lecture, Microsoft Research eScience Workshop, Berkeley, CA, "Genetics of amyotrophic lateral sclerosis"
- 2012 The Derek Denny-Brown Lecture, Annual meeting of the American Neurological Association, Boston, MA, "A Large Repeat Expansion in the C9ORF72 Gene is a Common Cause of ALS and FTD"
- 2012 Invited lecture, Center for Disease Control and Prevention, Atlanta, GA, "Large scale genome-wide association study of ALS"
- 2012 Invited lecture, The 2012 California ALS pac10 and research network meeting, Los Angeles, CA, "Expanding the genetics of ALS and FTD"
- 2013 The Sheila Essey Lecture, Annual meeting of the American Academy of Neurology, San Diego, CA, "Escapades in ALS and FTD genetics"
- 2014 The Edward H. Lambert Plenary Lecture, 39th Annual Edward H Lambert Plenary Lecture, AANEM Annual Conference, Savannah, GA, "Personalized genomics in neuromuscular medicine"
- 2016 Invited lecture, Cold Spring Harbor meeting on Neurodegenerative Diseases: Biology & Therapeutics, Cold Spring Harbor, NY, "Genomics of ALS"
- 2016 Grand Rounds, University of Tennessee, Memphis, TN, "Genomics of ALS"
- 2017 Grand Rounds, Mount Sinai New York, New York, NY, "Expanding the genetics of ALS and FTD"
- 2017 Grand Rounds, University of North Carolina, Durham, NC, "Expanding the genomics of ALS"
- 2019 Invited lecture, University at Albany, State University of New York, NY, "Genomics of ALS and FTD"

#### International

- 2000 Grand Rounds, National Neuroscience Center of Ireland, Beaumont Hospital, Dublin, Ireland, "Epidemiology of amyotrophic lateral sclerosis"
- 2004 Invited lecture, 127th ENMC International Workshop, Amsterdam, Netherlands, "Twin studies in amyotrophic lateral sclerosis"
- 2004 Invited lecture, Irish Diaspora Scientific Meeting, Dublin, Ireland, "Amyotrophic lateral sclerosis"
- 2006 Invited lecture, 17th International ALS/MND Symposium, Yokohama, Japan, "A genome wide association study of sporadic ALS"
- 2007 Invited lecture, 2nd International conference on ALS-FTD, London, Ontario, Canada, "Genetics of frontotemporal dementia and amyotrophic lateral sclerosis"
- 2007 Plenary lecture, Annual National Workshop, The Italian ALS Association, Turin, Italy, "Genome wide association study of amyotrophic lateral sclerosis"
- 2007 Invited lecture, Institute of Neurology, Queen Square Hospital, London, UK, "Genome wide association study of amyotrophic lateral sclerosis"
- 2007 Invited lecture, International collaboration on ALS Genetics, World Federation of Neurology, Toronto, ON, Canada, "Genetics of frontotemporal dementia and amyotrophic lateral sclerosis"
- 2008 Grand Rounds, University of Helsinki, Finland, "Genetics of sporadic amyotrophic lateral sclerosis"
- 2009 Plenary lecture, Annual National Workshop, The Italian ALS Association, Genoa, Italy, "Genetics of amyotrophic lateral sclerosis"
- 2010 Invited lecture, 62nd AAN Annual Meeting, Toronto, ON, Canada, "Emerging Concepts in Genetics Integrated Neuroscience Session"
- 2011 Invited lecture, 22nd International ALS/MND Symposium, Sydney, Australia, "A hexanucleotide repeat expansion in C9ORF72 is the cause of chromosome 9-linked ALS and FTD"
- 2011 Invited lecture, American Society of Human Genetics Annual Meeting, Montreal, Canada, "A hexanucleotide repeat expansion in C9ORF72 is the cause of chromosome 9-linked ALS and FTD"

- 2011 Invited lecture, World Federation of Neurology - ALS, Sydney, Australia, "A hexanucleotide repeat expansion in C9ORF72 is the cause of chromosome 9-linked ALS and FTD"
- 2012 Invited lecture, 16th International Congress of Parkinson's Disease and Movement Disorders, Dublin, Ireland, "Genetics of fronto-temporal dementia"
- 2012 Plenary lecture, International Conference on Frontotemporal Dementias, Manchester, UK, "C9ORF72 and ALS/FTD"
- 2012 Invited lecture, The 2012 Finnish Neuromuscular Society Annual Meeting, Helsinki, Finland, "Genetics of amyotrophic lateral sclerosis in Finland"
- 2012 Invited lecture, Workshop on amyotrophic lateral sclerosis organized by the Italian ALS Association, Naples, Italy, "Genetics of ALS What's in and what's out?"
- 2013 Invited lecture, ALS Association of Canada Annual Meeting, Toronto, Canada, "Genetics of ALS and FTD"
- 2016 The Potamkin Lecture, American Academy of Neurology, Vancouver, Canada, "Discovery of the C9orf72 repeat expansion"
- 2016 Invited lecture, International Congress of Human Genetics, Kyoto, Japan, "Genomics of amyotrophic lateral sclerosis"
- 2016 Invited lecture, Italian Society of Hospital Neurologists, Neurosurgeons, and Neuroradiologists Congress, Catania, Sicily, "What have we learned about ALS from genetics"
- 2016 Invited lecture, Nature Conference on Precision Medicine, Chengdu, China, "How genomics is driving drug discovery in ALS"
- 2016 Grand Rounds, University of Toronto, ON, Canada, "Genomics of ALS: past, present and future"
- 2017 Grand Rounds, University of Calgary, AB, Canada, "Genomics of ALS"
- 2017 Invited lecture, University of Turin, Italy, "From GWAS to genome sequence: opportunities and challenges"
- 2018 Invited lecture, ALS Symposium, University of Helsinki, Finland, "Genomics of ALS"
- 2020 Invited lecture, International Congress on Neuromuscular Diseases, "Advances in Genetics and Epigenetics of ALS"