e1. Floeter, M. K., B. J. Traynor, J. Farren, L. E. Braun, M. Tierney, E. A. Wiggs, and T. Wu. Disease progression in C9orf72 mutation carriers. *Neurology.* 2017;89: 234-41; doi: 10.1212/WNL.0000000000004115
e2.Chio, A., G. Borghero, G. Restagno, G. Mora, C. Drepper, B. J. Traynor, et al. Clinical characteristics of patients with familial amyotrophic lateral sclerosis carrying the pathogenic GGGGCC hexanucleotide repeat expansion of C9ORF72. Brain. 2012;135: 784-93; doi: 10.1093/brain/awr366
e3.Byrne, S., M. Elamin, P. Bede, A. Shatunov, C. Walsh, B. Corr, et al. Cognitive and clinical characteristics of patients with amyotrophic lateral sclerosis carrying a C9orf72 repeat expansion: a population-based cohort study. Lancet Neurol. 2012;11: 232-40; doi: 10.1016/S1474-4422(12)70014-5
e4.Umoh, M. E., C. Fournier, Y. Li, M. Polak, L. Shaw, J. E. Landers, et al: Comparative analysis of C9orf72 and sporadic disease in an ALS clinic population. Neurology. 2016;87: 1024-30; doi: 10.1212/WNL.0000000000003067
e5.Williams, K. L., J. A. Fifita, S. Vucic, J. C. Durnall, M. C. Kiernan, I. P. Blair, et al. Pathophysiological insights into ALS with C9ORF72 expansions. J Neurol Neurosurg Psychiatry. 2013;84: 931-5; doi: 10.1136/jnnp-2012-304529
e6.Strong, M. J., G. M. Grace, M. Freedman, C. Lomen-Hoerth, S. Woolley, L. H. Goldstein, et al. Consensus criteria for the diagnosis of frontotemporal cognitive and behavioural syndromes in amyotrophic lateral sclerosis. Amyotroph Lateral Scler. 2009;10: 131-46; doi: 10.1080/17482960802654364
e7.Rascovsky, K., J. R. Hodges, D. Knopman, M. F. Mendez, J. H. Kramer, J. Neuhaus, et al. Sensitivity of revised diagnostic criteria for the behavioural variant of frontotemporal dementia. Brain. 2011;134: 2456-77; doi: 10.1093/brain/awr179
e8.Strong, M. J., S. Abrahams, L. H. Goldstein, S. Woolley, P. McLaughlin, J. Snowden, et al. Amyotrophic lateral sclerosis - frontotemporal spectrum disorder (ALS-FTSD): Revised diagnostic criteria. Amyotroph Lateral Scler Frontotemporal Degener. 2017;18: 153-74; doi: 10.1080/21678421.2016.1267768
e9.McHutchison, C. A., D. J. Leighton, A. McIntosh, E. Cleary, J. Warner, M. Porteous, et al. Relationship between neuropsychiatric disorders and cognitive and behavioural change in MND. J Neurol Neurosurg Psychiatry. 2020;91: 245-53; doi: 10.1136/jnnp-2019-321737
e10.Woolley, S. C., M. K. York, D. H. Moore, A. M. Strutt, J. Murphy, P. E. Schulz, et al. Detecting frontotemporal dysfunction in ALS: utility of the ALS Cognitive Behavioral Screen (ALS-CBS). Amyotroph Lateral Scler. 2010;11: 303-11; doi: 10.3109/17482961003727954
e11.Abrahams, S., J. Newton, E. Niven, J. Foley, and T. H. Bak. Screening for cognition and behaviour changes in ALS. Amyotroph Lateral Scler Frontotemporal Degener. 2014;15: 9-14; doi: 10.3109/21678421.2013.805784
e12.Lomen-Hoerth, C., T. Anderson, and B. Miller: The overlap of amyotrophic lateral sclerosis and frontotemporal dementia. Neurology. 2002;59: 1077-9; doi: 10.1212/wnl.59.7.1077
e13.Murphy, J., P. Factor-Litvak, R. Goetz, C. Lomen-Hoerth, P. L. Nagy, J. Hupf, et al. Cognitive-behavioral screening reveals prevalent impairment in a large multicenter ALS cohort. Neurology. 2016;86: 813-20; doi: 10.1212/WNL.0000000000002305
e14.Silverman, H. E., J. S. Goldman, and E. D. Huey. Links Between the C9orf72 Repeat Expansion and Psychiatric Symptoms. Curr Neurol Neurosci Rep. 2019;19: 93; doi: 10.1007/s11910-019-1017-9
e15.Turner, M. R., R. Goldacre, K. Talbot, and M. J. Goldacre. Psychiatric disorders prior to amyotrophic lateral sclerosis. Ann Neurol. 2016;80: 935-38; doi: 10.1002/ana.24801
e16.O'Brien, M., T. Burke, M. Heverin, A. Vajda, R. McLaughlin, J. Gibbons, et al. Clustering of Neuropsychiatric Disease in First-Degree and Second-Degree Relatives of Patients With Amyotrophic Lateral Sclerosis. JAMA Neurol. 2017;74: 1425-30; doi: 10.1001/jamaneurol.2017.2699
e17.Marogianni, C., D. Rikos, A. Provatas, K. Dadouli, P. Ntellas, P. Tsitsi, et al. The role of C9orf72 in neurodegenerative disorders: a systematic review, an updated meta-analysis, and the creation of an online database. Neurobiol Aging. 2019;84: 238 e25-38 e34; doi: 10.1016/j.neurobiolaging.2019.04.012
e18.Snowden, J. S., J. Harris, A. Richardson, S. Rollinson, J. C. Thompson, D. Neary, et al. Frontotemporal dementia with amyotrophic lateral sclerosis: a clinical comparison of patients with and without repeat expansions in C9orf72. Amyotroph Lateral Scler Frontotemporal Degener. 2013;14: 172-6; doi: 10.3109/21678421.2013.765485

e19.Devenney, E. M., R. M. Ahmed, G. Halliday, O. Piguet, M. C. Kiernan, and J. R. Hodges. Psychiatric disorders in C9orf72 kindreds: Study of 1,414 family members. Neurology. 2018;91: e1498-e507; doi: 10.1212/WNL.0000000000006344
e20.Boeve, B. F., K. B. Boylan, N. R. Graff-Radford, M. DeJesus-Hernandez, D. S. Knopman, O. Pedraza, et al. Characterization of frontotemporal dementia and/or amyotrophic lateral sclerosis associated with the GGGGCC repeat expansion in C9ORF72. Brain. 2012;135: 765-83; doi: 10.1093/brain/aws004
e21.Floris, G., G. Borghero, A. Cannas, F. Di Stefano, E. Costantino, M. R. Murru, et al. Frontotemporal dementia with psychosis, parkinsonism, visuo-spatial dysfunction, upper motor neuron involvement associated to expansion of C9ORF72: a peculiar phenotype?. J Neurol. 2012;259: 1749-51; doi: 10.1007/s00415-012-6444-3
e22.O'Dowd, S., D. Curtin, A. J. Waite, K. Roberts, N. Pender, V. Reid, et al. C9ORF72 expansion in amyotrophic lateral sclerosis/frontotemporal dementia also causes parkinsonism. Mov Disord. 2012;27: 1072-4; doi: 10.1002/mds.25022
e23.Estevez-Fraga, C., F. Magrinelli, D. Hensman Moss, E. Mulroy, G. Di Lazzaro, A. Latorre, et al. Expanding the Spectrum of Movement Disorders Associated With C9orf72 Hexanucleotide Expansions. Neurol Genet. 2021;7: e575; doi: 10.1212/NXG.0000000000000575
e24.Nuytemans, K., G. Bademci, M. M. Kohli, G. W. Beecham, L. Wang, J. I. Young, et al. C9ORF72 intermediate repeat copies are a significant risk factor for Parkinson disease. Ann Hum Genet. 77: 351-63; doi: 10.1111/ahg.12033
e25.Theuns, J., A. Verstraeten, K. Sleegers, E. Wauters, I. Gijselinck, S. Smolders, et al. Global investigation and meta-analysis of the C9orf72 (G4C2)n repeat in Parkinson disease. Neurology. 2014;83: 1906-13; doi: 10.1212/WNL.0000000000001012
e26.Nuytemans, K., V. Inchausti, G. W. Beecham, L. Wang, D. W. Dickson, J. Q. Trojanowski, et al. Absence of C9ORF72 expanded or intermediate repeats in autopsy-confirmed Parkinson's disease. Mov Disord. 2014;29: 827-30; doi: 10.1002/mds.25838
e27.Carneiro, F., D. Saracino, V. Huin, F. Clot, C. Delorme, A. Meneret, et al. Isolated parkinsonism is an atypical presentation of GRN and C9orf72 gene mutations. Parkinsonism Relat Disord. 2020;80: 73-81. doi: 10.1016/j.parkreldis.2020.09.019
e28.Wilke, C., J. K. Pomper, S. Biskup, C. Puskas, D. Berg, and M. Synofzik. Atypical parkinsonism in C9orf72 expansions: a case report and systematic review of 45 cases from the literature. J Neurol. 2016;263: 558-74; doi: 10.1007/s00415-016-8021-7
e29.Cooper-Knock, J., A. Frolov, J. R. Highley, G. Charlesworth, J. Kirby, A. Milano, et al. C9ORF72 expansions, parkinsonism, and Parkinson disease: a clinicopathologic study. Neurology. 81: 808-11. doi: 10.1212/WNL.0b013e3182a2cc38
e30.Shinagawa, S., G. Naasan, A. M. Karydas, G. Coppola, M. Pribadi, W. W. Seeley, et al. Clinicopathological Study of Patients With C9ORF72-Associated Frontotemporal Dementia Presenting With Delusions. J Geriatr Psychiatry Neurol. 2015;28: 99-107; doi: 10.1177/0891988714554710
e31.Lesage, S., I. Le Ber, C. Condroyer, E. Broussolle, A. Gabelle, S. Thobois, et al. C9orf72 repeat expansions are a rare genetic cause of parkinsonism. Brain. 2013;136: 385-91;doi: 10.1093/brain/aws357
e32.Hensman Moss, D. J., M. Poulter, J. Beck, J. Hehir, J. M. Polke, T. Campbell, et al. C9orf72 expansions are the most common genetic cause of Huntington disease phenocopies. Neurology. 2014;82: 292-9; doi: 10.1212/WNL.0000000000000061
e33.Martins, J., J. Damasio, A. Mendes, N. Vila-Cha, J. E. Alves, C. Ramos, et al. Clinical spectrum of C9orf72 expansion in a cohort of Huntington's disease phenocopies. Neurol Sci. 2018;39: 741-44; doi: 10.1007/s10072-018-3268-7
e34.Kostic, V. S., V. Dobricic, I. Stankovic, V. Ralic, and E. Stefanova: C9orf72 expansion as a possible genetic cause of Huntington disease phenocopy syndrome. J Neurol. 2014;261: 1917-21; doi: 10.1007/s00415-014-7430-8
e35.van Blitterswijk, M., B. Mullen, M. G. Heckman, M. C. Baker, M. DeJesus-Hernandez, P. H. Brown, et al. Ataxin-2 as potential disease modifier in C9ORF72 expansion carriers. Neurobiol Aging. 2014;35: 2421 e13-7; doi: 10.1016/j.neurobiolaging.2014.04.016
e36.Figueroa, K. P., S. R. Gan, S. Perlman, G. Wilmot, C. M. Gomez, J. Schmahmann, et al. C9orf72 repeat expansions as genetic modifiers for depression in spinocerebellar ataxias. Mov Disord. 2018;33: 497-98; doi: 10.1002/mds.27258

e37.Snowden, J. S., S. Rollinson, J. C. Thompson, J. M. Harris, C. L. Stopford, A. M. Richardson, et al. Distinct clinical and pathological characteristics of frontotemporal dementia associated with C9ORF72 mutations. Brain. 2012;135: 693-708; doi: 10.1093/brain/awr355
e38.O'Rourke, J. G., L. Bogdanik, Akmg Muhammad, T. F. Gendron, K. J. Kim, A. Austin, et al. C9orf72 BAC Transgenic Mice Display Typical Pathologic Features of ALS/FTD. Neuron. 2015;88: 892-901. doi: 10.1016/j.neuron.2015.10.027
e39.Peters, O. M., G. T. Cabrera, H. Tran, T. F. Gendron, J. E. McKeon, J. Metterville, et al. Human C9ORF72 Hexanucleotide Expansion Reproduces RNA Foci and Dipeptide Repeat Proteins but Not Neurodegeneration in BAC Transgenic Mice. Neuron. 2015;88: 902-09; doi: 10.1016/j.neuron.2015.11.018
e40.Liu, Y., A. Pattamatta, T. Zu, T. Reid, O. Bardhi, D. R. Borchelt, et al. C9orf72 BAC Mouse Model with Motor Deficits and Neurodegenerative Features of ALS/FTD. Neuron. 2016;90: 521-34; doi: 10.1016/j.neuron.2016.04.005
e41.Mordes, D. A., B. M. Morrison, X. H. Ament, C. Cantrell, J. Mok, P. Eggan, et al. Absence of Survival and Motor Deficits in 500 Repeat C9ORF72 BAC Mice. Neuron. 2020;108: 775-83 e4; doi: 10.1016/j.neuron.2020.08.009
e42Nguyen, L., L. A. Laboissonniere, S. Guo, F. Pilotto, O. Scheidegger, A. Oestmann, et al. Survival and Motor Phenotypes in FVB C9-500 ALS/FTD BAC Transgenic Mice Reproduced by Multiple Labs. Neuron. 2020;108: 784-96 e3; doi: 10.1016/j.neuron.2020.09.009
e43Nguyen, L., F. Montrasio, A. Pattamatta, S. K. Tusi, O. Bardhi, K. D. Meyer, et al. Antibody Therapy Targeting RAN Proteins Rescues C9 ALS/FTD Phenotypes in C9orf72 Mouse Model. Neuron. 2020;105: 645-62 e11; doi: 10.1016/j.neuron.2019.11.007
e44.Pattamatta, A., L. Nguyen, H. R. Olafson, M. M. Scotti, L. A. Laboissonniere, J. Richardson, et al. Repeat length increases disease penetrance and severity in C9orf72 ALS/FTD BAC transgenic mice. Hum Mol Genet. 2021;29: 3900-18; doi: 10.1093/hmg/ddaa279
e45.Ciura, S., S. Lattante, I. Le Ber, M. Latouche, H. Tostivint, A. Brice, and E. Kabashi. Loss of function of C9orf72 causes motor deficits in a zebrafish model of amyotrophic lateral sclerosis. Ann Neurol. 2013;74: 180-7; doi: 10.1002/ana.23946
e46eTherrien, M., G. A. Rouleau, P. A. Dion, and J. A. Parker. Deletion of C9ORF72 results in motor neuron degeneration and stress sensitivity in C. elegans. PLoS One. 2013;8: e83450; doi: 10.1371/journal.pone.0083450
e47.Koppers, M., Blokhuis, A. M., Westeneng, H. J., Terpstra, M. L., Zundel, C. A., Vieira de Sá, R., et al. C9orf72 ablation in mice does not cause motor neuron degeneration or motor deficits. Annals of neurology. 2015;78(3), 426–438; doi: 10.1002/ana.24453
e48.Zhu, Q., J. Jiang, T. F. Gendron, M. McAlonis-Downes, L. Jiang, A. Taylor, et al. Reduced C9ORF72 function exacerbates gain of toxicity from ALS/FTD-causing repeat expansion in C9orf72. Nat Neurosci. 2020;23: 615-24; doi: 10.1038/s41593-020-0619-5