

- 1 Glanzmann B, Lombard D, Carr J, Bardien S. Screening of two indel polymorphisms in the 5' UTR of the DJ-1 gene in South African Parkinson's disease patients. *J Neural Transm* (Vienna). 2014;121(2):135-8.
- 2 Brey N, Henning F. Relapsing Neuromyelitis Optica temporally related to recurrent pulmonary tuberculosis. *Int J Tuberc Lung Dis*. 2014; 18 (5): 632-633.
- 3 Henning F, Bouic P. Increased Frequency of Guillain-Barré Syndrome in HIV Infection: A Prospective Cohort Study. *J AIDS Clin Res*. 2014; 5 (8): 332-336.
- 4 Blanckenberg J, Ntsapi C, Carr JA, Bardien S. EIF4G1 R1205H and VPS35 D620N mutations are rare in Parkinson's disease from South Africa. *Neurobiology of Ageing* 2014 Feb;35(2):445.e1-3.
- 5 Van der Merwe C, Loos B, Swart C, Kinnear C, Henning F; van der Merwe L, Pillay K, Muller N, Zaharie D, Engelbrecht L, Carr J, Bardien S. Mitochondrial impairment observed in fibroblasts from South African Parkinson's disease patients with parkin mutations. *Biochemical and Biophysical Research Communications*. 2014 May 2;447(2):334-40.
- 6 Geldenhuys G, Glanzmann B, Lombard D, Boolay S, Carr J, Bardien S. Identification of a common founder couple for 40 South African Afrikaner families with Parkinson's disease. *S Afr Med J*. 2014;104(6):413-9.
- 7 Heckman MG, Elbaz A, Soto-ortolaza AI, et al. Protective effect of LRRK2 p.R1398H on risk of Parkinson's disease is independent of MAPT and SNCA variants. *Neurobiol Aging*. 2014; 35(1):266.e5-14.
- 8 Theuns J, Verstraeten A, Sleegers K, et al. Global investigation and meta-analysis of the C9orf72 (G4C2) n repeat in Parkinson disease. *Neurology*. 2014; 83(21):1906-13
- 9 Carr J, Van Coller R. A putative founder effect for Parkinson's disease in South African Afrikaners. *S Afr Med J*. 2014;104(6):411-2.
- 10 Wang L, Aasly J, Annesi G, Bardien B, Bozi M, Brice A, Carr J, et al. Large Scale Assessment of Polyglutamine Repeat Expansions in Parkinson Disease . *Neurology*. 2015; 85(15):1283-92
- 11 Emsley R, Asmal L, Chiliza B, du Plessis S, Carr J, Kidd M, Malhotra AK, Vink M, Kahn RS et al. Changes in brain regions associated with food-intake regulation, body mass and metabolic profiles during acute antipsychotic treatment in first-episode schizophrenia. *Psychiatry Res*. 2015.
- 12 Fredericks P, Britz M, Eastman R, Carr JA, Bateman KJ. Listerial brainstem encephalitis--treatable, but easily missed. *S Afr Med J*. 2015;105(1):17-20.
- 13 Emsley R, Asmal L, Du plessis S, et al. Dorsal striatal volumes in never-treated patients with first-episode schizophrenia before and during acute treatment. *Schizophr Res*. 2015; 169(1-3):89-94..
- 14 Haylett W, Swart C, Van der westhuizen F, et al. Altered Mitochondrial Respiration and Other Features of Mitochondrial Function in Parkin-Mutant Fibroblasts from Parkinson's Disease Patients. *Parkinsons Dis*. 2016;2016:1819209.
- 15 Carr J, Guella I, Szu-tu C, et al. Double homozygous mutations (R275W and M432V) in the Parkin Gene associated with late-onset Parkinson's disease. *Mov Disord*. 2016;31(3):423-5.
- 16 Van der merwe C, Carr J, Glanzmann B, Bardien S. Exonic rearrangements in the known Parkinson's disease-causing genes are a rare cause of the disease in South African patients. *Neurosci Lett*. 2016;619:168-171.

- 17 Mahne AC, Carr JA, Bardien S, Schutte CM. Clinical findings and genetic screening for copy number variation mutations in a cohort of South African patients with Parkinson's disease. *S Afr Med J*. 2016;106(6):623-5.
- 18 Suliman S, Anthonissen L, Carr J, et al. Posttraumatic Stress Disorder, Overweight, and Obesity: A Systematic Review and Meta-analysis. *Harv Rev Psychiatry*. 2016;24(4):271-93
- 19 Carr J. In memoriam Raul De la Fuente-Fernández 1959-2016. *Parkinsonism Relat Disord*. 2016; 31:1-2.
- 20 Mahne AC, Carr JA, Bardien S, Schutte CM. Clinical findings and genetic screening for copy number variation mutations in a cohort of South African patients with Parkinson's disease. *S Afr Med J*. 2016;106(6):623-5.
- 21 Haylett W, Swart C, van der Westhuizen F, van Dyk H, van der Merwe L, van der Merwe C, Loos B, Carr J, Kinnear C, Bardien S. Altered Mitochondrial Respiration and Other Features of Mitochondrial Function in Parkin-Mutant Fibroblasts from Parkinson's Disease Patients. *Parkinsons Dis*. 2016;2016:1819209. doi: 10.1155/2016/1819209. Epub 2016 Mar 8. PMID: 27034887; PMCID: PMC4807059.
- 22 Carr J. Neurological letter from Cape Town. *Pract Neurol* 2017;17:74–76.
- 23 Henning F, Cunninghame CA, Martin MA, Rubio JA, Arenas J, Lucia A, Hernández-Lain A, Kohn TA. Muscle fiber type proportion and size is not altered in McArdle disease. *Muscle Nerve*. 2017 Jun;55(6):916-918.
- 24 Van Hillegondsberg L, Carr J, Brey N, Henning F. Using Eulerian video magnification to enhance detection of fasciculations in people with amyotrophic lateral sclerosis. *Muscle Nerve*. 2017 Dec;56(6):1063-1067.
- 25 Emsley R, Chiliza B, Asmal L, Kilian S, Olivier MR, Phahladira L, Ojagbemi A, Scheffler F, Carr J, Kidd M, Dazzan P. Neurological soft signs in first-episode schizophrenia: State and trait related relationships to psychopathology, cognition and antipsychotic medication effects. *Schizophrenia Res* 2017 Jan 24. pii: S0920-9964(17)30047-6.
- 26 Van Hillegondsberg L, Carr J, Brey N, Henning F. Using Eulerian video magnification to enhance detection of fasciculations in people with amyotrophic lateral sclerosis. *Muscle Nerve* 56: 1063-1067, 2017.
- 27 Mokaya J, Gray WK, Carr J. Beliefs, knowledge and attitudes towards Parkinson's disease among a Xhosa speaking black population in South Africa: A cross-sectional study. *Parkinsonism Relat Disord*. 2017;41:51-57
- 28 Anderson DG, Walker RH, Connor M, Carr J, Margolis RL, Krause A. A Systematic Review of the Huntington Disease-Like 2 Phenotype. *J Huntingtons Dis*. 2017;6(1):37-46.
- 29 Anderson D, van Coller R, Carr J. South African guidelines on Deep Brain Stimulation in Parkinson's disease. *S Afr Med J*. 2017.
- 30 Du Plessis S, Bossert M, Vink M, et al. Reward processing dysfunction in ventral striatum and orbitofrontal cortex in Parkinson's disease. *Parkinsonism Relat Disord*. 2017.
- 31 Anderson DG, Carmona S, Naidoo K, et al. Absence of Acanthocytosis in Huntington's Disease-like 2: A Prospective Comparison with Huntington's Disease. *Tremor Other Hyperkinet Mov (N Y)*. 2017; 7:512.

- 32 Kuipers DJS, Carr J, Bardien S, et al. PTRHD1 Loss-of-function mutation in an african family with juvenile-onset Parkinsonism and intellectual disability. *Mov Disord*. 2018;33(11):1814-1819.
- 33 Baboon (*Papio ursinus*) single fibre contractile properties are similar to that of trained humans. Dada S, Henning F, Feldmann DC, Kohn TA. *J Muscle Res Cell Motil*. 2018 Dec;39(5-6):189-199.
- 34 Oluwole OG, Kuivaniemi H, Carr JA, Ross OA, Olaogun MOB, Bardien S, Komolafe MA. Parkinson's disease in Nigeria: A review of published studies and recommendations for future research. *Parkinsonism Relat Disord*. 2018; Dec 8. pii: S1353-8020(18)30530-3.
- 35 Anderson DG, Haagensen M, Ferreira-correia A, Pierson P, Carr JA, Krause A, Margolis R. Emerging differences between Huntington's disease-like 2 and Huntington's disease: A comparison using MRI brain volumetry. *Neuroimage Clin*. 2019; 21:101666.
- 36 Abrahams S, Haylett WL, Johnson G, Carr JA, Bardien S. Antioxidant effects of curcumin in models of neurodegeneration, aging, oxidative and nitrosative stress: A review. *Neuroscience*. 2019; 406:1-21.
- 37 Van Coller R, Bardien S, Neethling A, Carr J, Schutte C. A South African family with myoclonus-dystonia syndrome with a novel mutation in the SGCE gene responding to deep brain stimulation. *Parkinsonism Relat Disord*. 2019; 68:63-64.
- 38 Clinical features and prognosis of amyotrophic lateral sclerosis in Africa: the TROPALS study. Luna J, Diagana M, Ait Aissa L, Tazir M, Ali Pacha L, Kacem I, Gouider R, Henning F, Basse A, Cisse O, Balogou AAK, Kombate D, Agbetou M, Houinato D, Millogo A, Agba T, Belo M, Penoty M, Raymondeau-Moustafa M, Hamidou B, Couratier P, Preux PM, Marin B; TROPALS Collaboration. *J Neurol Neurosurg Psychiatry*. 2019 Jan;90(1):20-29.
- 39 Adams B, Nunes JM, Page MJ, et al. Parkinson's disease: A Systemic Inflammatory Disease Accompanied by Bacterial Inflammagens. *Front Aging Neurosci*. 2019; 11:210.
- 40 C9orf72 repeat expansions in South Africans with amyotrophic lateral sclerosis. Nel M, Agenbag GM, Henning F, Cross HM, Esterhuizen A, Heckmann JM. *J Neurol Sci*. 2019 Apr 17;401:51-54.
- 41 Oluwole OG, Kuivaniemi H, Abrahams S, et al. Targeted next-generation sequencing identifies novel variants in candidate genes for Parkinson's disease in Black South African and Nigerian patients. *BMC Med Genet*. 2020;21(1):23.
- 42 Anderson DG, Ferreira-Correia A, Rodrigues FB, et al. Comparison of the Huntington's Disease like 2 and Huntington's Disease Clinical Phenotypes. *Mov Disord Clin Pract*. 2019;6(4):302-311. Published 2019 Mar 12. doi:10.1002/mdc3.12742
- 43 Vollstedt EJ, Kasten M, Klein C; MJFF Global Genetic Parkinson's Disease Study Group. Using global team science to identify genetic parkinson's disease worldwide. *Ann Neurol*. 2019 Aug;86(2):153-157. doi: 10.1002/ana.25514. Epub 2019 Jun 26. PMID: 31155756; PMCID: PMC7410260.
- 44 du Toit N, van Coller R, Anderson DG, Carr J, Bardien S. Frequency of the LRRK2 G2019S mutation in South African patients with Parkinson's disease. *Neurogenetics*. 2019 Oct;20(4):215-218. doi: 10.1007/s10048-019-00588-z. Epub 2019 Sep 6. PMID: 31493133.
- 45 Dekker MCJ, Coulibaly T, Bardien S, Ross OA, Carr J, Komolafe M. Parkinson's Disease Research on the African Continent: Obstacles and Opportunities. *Front Neurol*. 2020;11:512. Published 2020 Jun 19. doi:10.3389/fneur.2020.00512

- 46 van den Heuvel LL, du Plessis S, Stalder T, et al. Hair glucocorticoid levels in Parkinson's disease. *Psychoneuroendocrinology*. 2020;117:104704. doi:10.1016/j.psyneuen.2020.104704
- 47 Bardien S, Carr J. A model PD registry for countries with limited resources [published online ahead of print, 2020 Aug 3]. *Nat Rev Neurol*. 2020;10.1038/s41582-020-0396-5. doi:10.1038/s41582-020-0396-5
- 48 A comparative study of South African and Portuguese amyotrophic lateral sclerosis cohorts. Braga AC, Gromicho M, Pinto S, de Carvalho M, Henning F. *J Neurol Sci*. 2020 Jul 15;414:116857.
- 49 Authors' reply: Differences between South African and Portuguese ALS cohorts from an environmental perspective. Henning F, Braga AC, Gromicho M, Pinto S, de Carvalho M. *J Neurol Sci*. 2020 Jul 15;414:116932
- 50 An exploratory study of contractile force production in muscle fibers from patients with inflammatory myopathies. Henning F, Kohn TA. *Muscle Nerve*. 2020 Aug;62(2):284-288.
- 51 Incidence of motor neuron disease/amyotrophic lateral sclerosis in South Africa: a 4-year prospective study. Henning F, Heckmann JM, Naidu K, Vlok L, Cross HM, Marin B. *Eur J Neurol*. 2020 Sep 5.
- 52 Young carers and ALS/MND: exploratory data from families in South Africa. Kavanaugh MS, Henning F, Mochan A. *Vulnerable Children and Youth Studies*. 2020 DOI: 10.1080/17450128.2020.1837409
- 53 Mahungu AC, Anderson DG, Rossouw AC, et al. Screening of the glucocerebrosidase (GBA) gene in South Africans of African ancestry with Parkinson's disease. *Neurobiol Aging*. 2020;88:156.e11-156.e14. doi:10.1016/j.neurobiolaging.2019.12.011
- 54 du Plessis S, Bekker M, Buckle C, Vink M, Seedat S, Bardien S, Carr J, Abrahams S. Association Between a Variable Number Tandem Repeat Polymorphism Within the DAT1 Gene and the Mesolimbic Pathway in Parkinson's Disease. *Front Neurol*. 2020 Sep 2;11:982. doi: 10.3389/fneur.2020.00982. PMID: 32982958; PMCID: PMC7493621.
- 55 Hassan A, Mari Z, Gatto EM, Cardozo A, Youn J, Okubadejo N, Bajwa JA, Shalash A, Fujioka S, Aldaajani Z, Cubo E; International Telemedicine Study Group. Global survey on telemedicine utilization for movement disorders during the COVID-19 pandemic. *Mov Disord*. 2020 Aug 24;10.1002/mds.28284. doi: 10.1002/mds.28284. Epub ahead of print. PMID: 32833273; PMCID: PMC7461376.
- 56 Spies G, Mokaya J, Steadman J, Schuitmaker N, Kidd M, Hemmings SMJ, Carr JA, Kuivaniemi H, Seedat S; SHARED ROOTS Group. Attitudes among South African university staff and students towards disclosing secondary genetic findings. *J Community Genet*. 2020 Nov 20. doi: 10.1007/s12687-020-00494-0. Online ahead of print. PMID: 33219499
- 57 Vuuren MJV, Nell TA, Carr JA, Kell DB, Pretorius E. Iron Dysregulation and Inflammagens Related to Oral and Gut Health Are Central to the Development of Parkinson's Disease. *Biomolecules*. 2020 Dec 29;11(1):30. doi: 10.3390/biom11010030. PMID: 33383805; PMCID: PMC7823713.
- 58 Sebate B, Cuttler K, Cloete R, Britz M, Christoffels A, Williams M, Carr J, Bardien S. Prioritization of candidate genes for a South African family with Parkinson's disease using in-silico tools. *PLoS One*. 2021 Mar 26;16(3):e0249324. doi: 10.1371/journal.pone.0249324. PMID: 33770142; PMCID: PMC7997022.

- 59 Chetty D, Abrahams S, van Coller R, Carr J, Kenyon C, Bardien S. Movement of prion-like α -synuclein along the gut-brain axis in Parkinson's disease: A potential target of curcumin treatment. *Eur J Neurosci*. 2021 May 27. doi: 10.1111/ejn.15324. Epub ahead of print. PMID: 34043864.
- 60 Hamid E, Ayele BA, Massi DG, Ben Sassi S, Tibar H, Djonga EE, El-Sadig SM, Amer El Khedoud W, Razafimahefa J, Kouame-Assouan AE, Ben-Adji D, Lengané YTM, Musubire AK, Mohamed MH, Phiri TE, Nestor N, Alwahchi WA, Neshuku SN, Ocampo C, Sakadi F, Moidal MA, Ngwende GW, Hooker J, Okeng'o K, Charway-Felli A, Atadzhanov M, Carr J, Okubadejo NU, Shalash A. Availability of Therapies and Services for Parkinson's Disease in Africa: A Continent-Wide Survey. *Mov Disord*. 2021 Jun 3. doi: 10.1002/mds.28669. Epub ahead of print. PMID: 34080713.
- 61 Clinical management and disease-modifying treatment for amyotrophic lateral sclerosis in African hospital centres: the TROPALS study. Luna J, Jost J, Diagana M, Aissa LA, Tazir M, Pacha LA, Kacem I, Gouider R, Henning F, Basse A, Cisse O, Balogou AAK, Kombate D, Agbetou M, Houinatoum D, Gnonlonfoun DD, Millogo A, Agba T, Belo M, Sengxeu N, Hamidou B, Preux P-M, Marin B, Couratier P, TROPALS Collaboration. *Amyotroph Lateral Scler Frontotemporal Degener*. 2021 Aug 28:1-5
- 62 Müller-Nedebock AC, Komolafe MA, Fawale MB, Carr JA, van der Westhuizen FH, Ross OA, Bardien S. Copy Number Variation in Parkinson's Disease: An Update from Sub-Saharan Africa. *Mov Disord*. 2021 Jul 6. doi: 10.1002/mds.28710. Epub ahead of print. PMID: 34228376.
- 63 Welton T, Cardoso F, Carr JA, *et al*. Essential tremor. *Nat Rev Dis Prim* 2021; **7**: 20894.
- 64 Cuttler K, Hassan M, Carr J, Cloete R, Bardien S. Emerging evidence implicating a role for neurexins in neurodegenerative and neuropsychiatric disorders. *Open Biol*. 2021 Oct;11(10):210091. doi: 10.1098/rsob.210091. Epub 2021 Oct 6. PMID: 34610269.
- 65 van Rensburg ZJ, Abrahams S, Chetty D, Step K, Acker D, Lombard CJ, Elbaz A, Carr J, Bardien S. The South African Parkinson's Disease Study Collection. *Mov Disord*. 2021 Oct 22. doi: 10.1002/mds.28828. Epub ahead of print. PMID: 34676912.
- 66 Doruyter AGG, Parkes J, Carr J, Warwick JM. PET-CT in brain disorders: The South African context. *SA J Radiol*. 2021 Nov 10;25(1):2201. doi: 10.4102/sajr.v25i1.2201. PMID: 34858659; PMCID: PMC8603194.
- 67 Hendrikse CJ, Malan T, Du Plessis S, Carr JA, Kidd M, Emsley RA, Seedat S. Incidental neuroimaging findings in South African adult research participants with and without neuropsychiatric disorders. *Neuroimage: Reports* 2022; **2**(4):100130.
- 68 Modification of the ALSFRS-R for Utilization in Individuals Not Using Noninvasive Ventilation. Vlok L, Rossouw L, Henning F. *Respir Care*. 2022 May;67(5):553-561.
- 69 Preservation of shortening velocity and power output in single muscle fibres from patients with idiopathic inflammatory myopathies. Henning F, Kohn TA. *J Muscle Res Cell Motil*. 2022 Dec 15. doi: 10.1007/s10974-022-09638
- 70 Fevga C, Tesson C, Mascaro AC, Courtin T, van Coller R, Sakka S, Ferraro F, Farhat N, Bardien S, Damak M, Carr J, Ferrien M, Boumeester V, Hundscheid J, Grillenzoni N, Kessissoglou IA, Kuipers DJS, Quadri M; French and Mediterranean Parkinson disease Genetics Study Group; International Parkinsonism Genetics Network, Corvol JC, Mhiri C, Hassan BA, Breedveld GJ, Lesage S, Mandemakers W, Brice A, Bonifati V. PTPA variants and impaired PP2A activity in early-

onset parkinsonism with intellectual disability. *Brain*. 2022 Sep 8;awac326. doi: 10.1093/brain/awac326. Epub ahead of print. PMID: 36073231.

- 71 Hamid E, Okengo K, Ayele BA, Gams Massi D, Ben Sassi S, Tibar H, El-Sadig SM, Mahoui S, Razafimahefa J, Kouame-Assouan AE, Ben-Adji D, Modeste LYT, Mohamed MH, Nsengiyumva N, Alwahchi WA, Neshuku SN, Ocampo C, Sakadi F, Caiano JB, Rodrigues Fortes A, Fall M, Ngwende GW, Hooker J, Charway-Felli A, Atadzhanov M, Carr J, Okubadejo NU, Shalash A. The Gaps and Prospects of Movement Disorders Education and Research in Africa: A Continental Survey. *Mov Disord*. 2023 Jan 26. doi: 10.1002/mds.29313. PMID: 36703239.
- 72 Neuromuscular disease genetics in underrepresented populations: increasing data diversity. Wilson LA, Macken WL, Perry LD, Record CJ, Schon KR, Frezatti RSS, Raga S, Naidu K, Köken ÖY, Polat I, Kapapa MM, Dominik N, Efthymiou S, Morsy H, Nel M, Fassad MR, Gao F, Patel K, Schoonen M, Bisschoff M, Vorster A, Jonvik H, Human R, Lubbe E, Nonyane M, Vengalil S, Nashi S, Srivastava K, Lemmers RJLF, Reyaz A, Mishra R, Töpf A, Trainor CI, Steyn EC, Mahungu AC, van der Vliet PJ, Ceylan AC, Hiz AS, Çavdarlı B, Semerci Gündüz CN, Ceylan GG, Nagappa M, Tallapaka KB, Govindaraj P, van der Maarel SM, Narayanappa G, Nandeesh BN, Wa Somwe S, Bearden DR, Kvalsund MP, Ramdharry GM, Oktay Y, Yiş U, Topaloğlu H, Sarkozy A, Bugiardini E, Henning F, Wilmshurst JM, Heckmann JM, McFarland R, Taylor RW, Smuts I, van der Westhuizen FH, Sobreira CFDR, Tomaselli PJ, Marques W, Bhatia R, Dalal A, Srivastava MVP, Yareeda S, Nalini A, Vishnu VY, Thangaraj K, Straub V, Horvath R, Chinnery PF, Pitceathly RDS, Muntoni F, Houlden H, Vandrovcova J, Reilly MM, Hanna MG. *Brain*. 2023 Jul 30;awad254. doi: 10.1093/brain/awad254
- 73 Myelin oligodendrocyte glycoprotein antibody-associated disease (MOGAD) and Human Immunodeficiency virus infection: dilemmas in diagnosis and management: a case series. Gadama Y, Du Preez M, Carr J, Theron S, Albertyn C, Ssebambulidde K, Saylor D, Brey N, Henning F. *J Med Case Rep*. 2023 Oct 17;17(1):457. doi: 10.1186/s13256-023-04191-7
- 74 Clinical, biochemical, and genetic spectrum of MADD in a South African cohort: an ICGNMD study. Bisschoff M, Smuts I, Dercksen M, Schoonen M, Vorster BC, van der Watt G, Spencer C, Naidu K, Henning F, Meldau S, McFarland R, Taylor RW, Patel K, Fassad MR, Vandrovcova J; ICGNMD Consortium; Wanders RJA, van der Westhuizen FH. *Orphanet J Rare Dis*. 2024 Jan 14;19(1):15. doi: 10.1186/s13023-023-03014-8.
- 75 Best practice guidelines on genetic diagnostics of facioscapulohumeral muscular dystrophy: Update of the 2012 guidelines. Giardina E, Camaño P, Burton-Jones S, Ravenscroft G, Henning F, Magdinier F, van der Stoep N, van der Vliet PJ, Bernard R, Tomaselli PJ, Davis MR, Nishino I, Oflazer P, Race V, Vishnu VY, Williams V, Sobreira CFR, van der Maarel SM, Moore SA, Voermans NC, Lemmers RJLF. *Clin Genet*. 2024 Apr 29. doi: 10.1111/cge.14533.
- 76 Narotam-Jeena H, Guttman M, van Hilleghondsberg L, van Coller R, Krause A, Carr J. Atypical Presentations of Huntington Disease-like 2 in South African Individuals. *Mov Disord Clin Pract*. 2024 Jul;11(7):850-854. doi: 10.1002/mdc3.14052. Epub 2024 May 9. PMID: 38725192; PMCID: PMC11233840.