REGINA H REYNOLDS

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KEY PUBLICATIONS

2023

Local genetic correlations exist among neurodegenerative and neuropsychiatric diseases[1]

NPJ Parkinson's disease

- Reynolds, RH, Wagen, AZ, Lona-Durazo, F, Scholz, SW, Shoai, M, Hardy, J, Gagliano Taliun, SA, Ryten, M
- Role: Co-first author, lead analyst and corresponding author.
- Analysis[2] of local genetic correlations between neurodegenerative and neuropsychiatric disorders, with the aim of identifying genomic regions and genes that may drive pleiotropy.

2021

Cross-platform transcriptional profiling identifies common and distinct molecular pathologies in Lewy body disorders[3]

Acta Neuropathologica

- Feleke, R, Reynolds, RH, Smith, A, Tilley, B, Gagliano Taliun, SA, Hardy, J, Matthews, PM, Gentleman, S, Owen , D, Johnson, MR, Srivastava, P, Ryten, M
- Role: Co-first author and analyst.
- Transcriptomic analysis[4] of cell-type-specific changes in the Lewy body diseases.

2020

Dystonia genes functionally converge in specific neurons and share neurobiology with psychiatric disorders[5]

Brain

- Mencacci, NE, Reynolds, RH, Garcia Ruiz, S, Vandrovcova, J, Forabosco, P, Sánchez-Ferrer, A, Volpato, V, UK Brain Expression Consortium, International Parkinson's Disease Genomics Consortium, Weale, ME, Bhatia, KP, Webber, C, Hardy, J, Botía, JA, Ryten, M
- Role: Co-first author and analyst.
- Integrative omics analysis of monogenic dystonias, with the aim of improving our understanding of the pathways driving this clinically heterogeneous group of movement disorders.

2019

Informing disease modelling with brain-relevant functional genomic annotations[6]

Brain

- Reynolds, RH, Hardy, J, Ryten, M, Gagliano Taliun, SA
- Role: First author.
- Review of conceptual advances in the generation of brain-relevant functional genomic annotations and among tools that allow integration of these annotations with genome-wide association summary statistics.

CONTACT

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GitHub

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ResearchGate

The long-form version of my CV is available here.

The source code is available GitHub.

Last updated on 2025-06-22.

2019

Moving beyond neurons: the Role of cell type-specific gene regulation in Parkinson's disease heritability[7]

NPJ Parkinson's disease

- Reynolds, RH, Botía, JA, Nalls, MA, International Parkinson's Disease Genomic Consortium (IPDGC), System Genomics of Parkinson's Disease (SGPD), Hardy, J, Gagliano Taliun, SA, Ryten, M
- Role: First author and lead analyst.
- Analysis of Parkinson's disease common variation, with the aim of identifying cell types and pathways of importance to disease risk.

2018

Perturbations in the p53/miR-34a/SIRT1 pathway in the R6/2 Huntington's disease model[8]

Molecular and Cellular Neuroscience

- Reynolds, RH, Petersen, MH, Willert, CW, Heinrich, M, Nymann, N, Dall, M, Treebak, JT, Björkqvist, M, Silahtaroglu, A, Hasholt, L and Nørremølle, A
- Role: First author, lead experimentalist and analyst.

PUBLICATIONS

2025

Splicing accuracy varies across human introns, tissues, age and disease[9]

Nature Communications

- Garcia-Ruiz, S, Zhang, D, Gustavsson, EK, Rocamora-Perez, G, Grant-Peters, M, Fairbrother-Browne, A, Reynolds, RH et al.
- Role: Adviser. Reviewed and critiqued statistical analyses and manuscript.

2025

Astrocytic RNA editing regulates the host immune response to alpha-synuclein[10]

Science Advances

- D'Sa, K, Choi, ML, Wagen, AZ, Seto-Savlia, N, Kopach, O, Evans, JR, Rodrigues, M, Lopez-Garcia, P, Lachica, J, Clarke, BE, Singh, J, Ghareeb, A, Bayne, J, Grant-Peters, M, Garcia-Ruiz, S, Chen, Z, Rodriques, S, Athauda, A, Gustavsson, EK, Gagliano-Taliun, SA, Toomey, C, Reynolds RH et al.
- Role: Analyst and adviser. Contributed to tooling around alignment, advised on deconvolution and RNA-editing. Critiqued manuscript.

2024

The annotation of GBA1 has been concealed by its protein-coding pseudogene GBAP1[11]

Science Advances

- Gustavsson, EK, Sethi, S, Gao, Y, Brenton, J, Ruiz, SG, Zhang, D, Garza, R, Reynolds, RH et al.
- Role: Adviser. Critiqued manuscript.

2024

Genome sequence analyses identify novel risk loci for multiple system atrophy[12]

Neuron

- Chia, R, Ray, A, Shah, Z, Dong, J, Ruffo, P, Fujita, M, Menon, V, Saez-Atienzar, S, Reho, P, Kaivola, K, Walton, R, Reynolds RH et al.
- Role: Adviser. Reviewed and critiqued colocalisation analyses.

2023

The non-specific lethal complex regulates genes and pathways genetically linked to Parkinson's disease[13]

Brain

- Hicks, AR, Reynolds, RH et al.
- Role: Adviser. Reviewed and critiqued statistical analyses and manuscript.

2023

ensemblQueryR: fast, flexible and high-throughput querying of Ensembl LD API endpoints in R[14]

GigaByte

- Fairbrother-Browne, A, Garcia-Ruiz, S, Reynolds, RH et al.
- Role: Adviser. Contributed to conceptualisation and critiqued manuscript.

2023

Analysis of subcellular RNA fractions demonstrates significant genetic regulation of gene expression in human brain post-transcriptionally[15]

Scientific Reports

- D'Sa, K, Guelfi, S, Vandrovcova, J, Reynolds RH et al.
- Role: Adviser. Contributed to troubleshooting of analyses.

2023

aws-s3-integrity-check: an open-source bash tool to verify the integrity of a dataset stored on Amazon S3[16]

GigaByte

- Garcia-Ruiz, S, Reynolds, RH et al.
- Role: Adviser. Contributed to idea development and critiqued manuscript.

2023

Genome-wide Analysis of Motor Progression in Parkinson Disease[17]

Neurology Genetics

- Martinez-Carrasco, A, Real, R, Lawton, M, Reynolds, RH et al.
- Role: Adviser. Reviewed and critiqued statistical analyses and manuscript.

2023

Regional genetic correlations highlight relationships between neurodegenerative disease loci and the immune system[18]

Communications Biology

- Lona-Durazo, F, Reynolds, RH et al.
- Role: Analyst and advisor. Contributed to code base and critiqued manuscript.

2023

Association between the LRP1B and APOE loci and the development of Parkinson's disease dementia[19]

Brain

- Real, R, Martinez-Carrasco, A, Reynolds, RH et al.
- Role: Adviser. Critiqued manuscript.

2023

The contribution of Neanderthal introgression and natural selection to neurodegenerative diseases[20]

Neurobiology of Disease

- Chen, Z, Reynolds RH et al.
- Role: Author and analyst. Performed partitioned SNP-heritability analyses and contributed to writing of results.

2023

The IPDGC/GP2 Hackathon – an open science event for training in data science, genomics, and collaboration using Parkinson's disease data[21]

NPJ Parkinson's disease

- Leonard, HL, Reynolds, RH et al. on behalf of The International Parkinson Disease Genomics Consortium (IPDGC) and The Global Parkinson's Genetics Program (GP2)
- Role: Author and analyst. Contributed to creation of tools.

2022

IntroVerse: a comprehensive database of introns across human tissues[22]

Nucleic Acid Research

- García-Ruíz, S, Gustavsson, EK, Zhang, D, Reynolds, RH et al.
- Role: Adviser. Critiqued manuscript.

2022

Genome-wide association study of REM sleep behavior disorder identifies polygenic risk and brain expression effects[23]

Nature Communications

- Krohn, L, Heilbron, K, Blauwendraat, C, Reynolds, RH, et al.
- Role: Analyst. Performed colocalisation analyses and gene expression analyses.

2022

ggtranscript: an R package for the visualization and interpretation of transcript isoforms using ggplot2[24]

Bioinformatics

- Gustavsson, EK, Zhang, D, Reynolds, RH, Ruiz, SG, Ryten, MR
- Role: Adviser. Performed beta testing and critiqued manuscript.

2021

A genetic link between risk for Alzheimer's disease and severe COVID-19 outcomes via the OAS1 gene[25]

Brain

- Magusali, N, Graham, AC, Piers, TM, Panichnantakul, P, Yaman, U, Shoai, M, Reynolds, RH et al.
- Role: Adviser. Reviewed and critiqued statistical analyses and manuscript.

2021

Mitochondrial-nuclear cross-talk in the human brain is modulated by cell type and perturbed in neurodegenerative disease[26]

Communications Biology

- Fairbrother-Browne, A, Ali, A, Reynolds, RH et al.
- Role: Adviser. Advised on tools available for cell-type specificity and heritability analyses.

2021

A systems-level analysis highlights microglial activation as a modifying factor in common epilepsies[27]

Neuropathology and Applied Neurobiology

- Altmann, A, Ryten, M, Di Nunzio, M, Ravizza, T, Tolomeo, D, Reynolds, RH et al.
- Role: Analyst. Collated gene lists designed to capture microglial states.

2021

An integrated genomic approach to dissect the genetic landscape regulating the cell-to-cell transfer of a-synuclein[28]

Cell Reports

- Kara, E, Crimi, A, Wiedmer, A, Emmenegger, M, Manzoni, C, Bandres-Ciga, S, D'Sa, K, Reynolds, RH et al.
- Role: Analyst. Performed expression-weighted cell-type enrichment analyses.

2021

Heritability enrichment implicates microglia in Parkinson's disease pathogenesis[29]

Annals of Neurology

- Andersen, MS, Bandres-Ciga, S, Reynolds, RH et al.
- Role: Adviser. Reviewed and critiqued statistical analyses and manuscript.

2021

Human-lineage-specific genomic elements: relevance to neurodegenerative disease and APOE transcript usage[30]

Nature Communications

- Chen, Z, Zhang, D, Reynolds, RH et al.
- Role: Analyst. Performed partitioned SNP-heritability analyses.

2021

Identification of Candidate Parkinson Disease Genes by Integrating Genome-Wide Association Study, Expression, and Epigenetic Data Sets[31]

JAMA Neurology

- Kia, DA, Zhang, D, Guelfi, S, Manzoni, C, Hubbard, L, Reynolds, RH et al.
- Role: Analyst. Involved in cell-type annotation of prioritised genes.

2021

Modelling multifunctionality of genes with secondary gene co-expression networks in human brain provides novel disease insights[32]

Bioinformatics

- Sánchez, JA, Gil-Martínez, AL, Cisterna, A, García-Ruíz, S, Gómez-Pascual, A, Reynolds, RH et al.
- Role: Adviser. Reviewed and critiqued statistical analyses and manuscript.

2021

Genome sequencing analysis identifies new loci associated with Lewy body dementia and provides insights into the complex genetic architecture[33]

Nature Genetics

- Chia, R, Sabir, MS, Bandres-Ciga, S, Saez-Atienzar, S, Reynolds, RH et al.
- Role: Analyst. Performed colocalisation analyses and gene expression analyses.

2021

CoExp Web, a web tool for the exploitation of co-expression networks[34]

Frontiers in Genetics

- García-Ruíz, S, Gil-Martínez, AL, Cisterna, A, Jurado, F, Reynolds, RH et al.
- Role: Analyst. Involved in cell-type annotation of gene co-expression networks.

2021

Genetic determinants of survival in progressive supranuclear palsy: a genome-wide association study[35]

Lancet Neurology

- Jabbari, E, Koga, S, Valentino, R, Reynolds, RH et al.
- Role: Analyst. Performed colocalisation analyses.

2021

Genome-wide association studies of cognitive and motor progression in Parkinson's disease[36]

Movement Disorders

- Tan, MMX, Lawton, MA, Jabbari, E, Reynolds, RH et al.
- Role: Adviser. Reviewed and critiqued statistical analyses and manuscript.

2021

Genetic analysis of amyotrophic lateral sclerosis identifies contributing pathways and cell types[37]

Science Advances

- Saez-Atienzar, S, Bandres-Ciga, S, Langston, RG, Kim, JJ, Choi, SW, Reynolds, RH et al.
- Role: Adviser. Reviewed and critiqued statistical analyses and manuscript.

2020

Differences in network controllability and regional gene expression underlie hallucinations in Parkinson's disease[38]

Brain

- Zarkali, A, McColgan, P, Ryten, M, Reynolds, RH et al.
- Role: Adviser. Reviewed and critiqued statistical analyses and manuscript.

2020

Dementia risk in Parkinson's disease is associated with interhemispheric connectivity loss and determined by regional gene expression[39]

NeuroImage: Clinical

- Zarkali, A, McColgan, P, Ryten, M, Reynolds, RH et al.
- Role: Adviser. Reviewed and critiqued statistical analyses and manuscript.

2020

Analysis of brain atrophy and local gene expression in genetic frontotemporal dementia[40]

Brain Communications

- Altmann, A, Cash, DM, Bocchetta, M, Heller, C, Reynolds, RH et al.
- Role: Analyst. Performed expression-weighted cell-type enrichment analyses.

2020

Incomplete annotation of disease-associated genes is limiting our understanding of Mendelian and complex neurogenetic disorders[41]

Science Advances

- Zhang, D, Guelfi, S, Ruiz, SG, Costa, B, Reynolds, RH et al.
- Role: Analyst. Performed expression-weighted cell-type enrichment analyses.

2020

Regulatory sites for known and novel splicing in human basal ganglia are enriched for disease-relevant information[42]

Nature Communications

- Guelfi, S*, D'Sa, K*, Botía, JA*, Vandrovcova, J, Reynolds, RH et al.
- Role: Experimentalist and analyst. Performed PCR validation of transcription and partitioned SNP-heritability analyses.

2019

Genetic variability in response to amyloid beta deposition influences Alzheimer's risk[43]

Brain Communications

- Salih, DA, Bayram, S, Guelfi, S, Reynolds, RH et al.
- Role: Analyst. Performed colocalisation analyses.

2019

Mitochondria function associated genes contribute to Parkinson's Disease risk and later age at onset[44]

NPJ Parkinson's disease

- Billingsley, KJ, Barbosa, IA, Bandrés-Ciga S, Quinn, JP, Bubb, VJ, Deshpande, C, Botía, JA, Reynolds, RH et al.
- Role: Analyst. Performed expression-weighted cell-type enrichment analyses.

2018

Variation at the TRIM11 locus modifies Progressive Supranuclear Palsy phenotype[45]

Annals of Neurology

- Jabbari, E, John, W, Tan, MMX, Maryam, S, Pittman, A, Ferrari, R, Mok, KY, Zhang, D, Reynolds, RH et al.
- Role: Analyst. Performed gene-based association testing and analysis of gene expression data.

PRE-PRINTS

2025

Molecular and cellular signatures differentiate Parkinson's disease from Parkinson's disease with dementia[46]

bioRxiv

- Fairbrother-Browne, A, Grant-Peters, M, Brenton, JW, Nelvagal, H, Reynolds, RH et al.
- Role: Experimentalist and analyst. Involved in nuclei isolation and 10X library preparation. Aligned and QC-ed single-nucleus RNA-sequencing. Critiqued manuscript.

2025

17q21.31 locus regulates Parkinson's disease relevant pathways through KANSL1 activity[47]

bioRxiv

- Hicks, AR, O'Callaghan, B, Brenton, JW, Grant-Peters, M, Fairbrother-Browne, A, Perez, GR, Loh, CA, Reynolds, RH et al.
- Role: Experimentalist and analyst. Involved in nuclei isolation and 10X library preparation. Aligned and QC-ed single-nucleus RNA-sequencing.

2024

Ancestry-specific gene expression in peripheral monocytes mediates risk of neurodegenerative disease[48]

bioRxiv

- Wagen, AZ, Reynolds RH et al.
- Role: Adviser. Reviewed and critiqued statistical analyses and manuscript.

2021

Detection of pathogenic splicing events from RNA-sequencing data using dasper[49]

bioRxiv

- Zhang, D, Reynolds, RH, et al.
- Role: Adviser. Helped guide and troubleshoot feature selection for outlier detection of splicing events.

2019

ERASE: Extended Randomization for assessment of annotation enrichment in ASE datasets[50]

bioRxiv

- D'Sa, K, Reynolds RH et al.
- Role: Analyst. Performed partitioned SNP-heritability analyses.

2018

G2P: Using machine learning to understand and predict genes causing rare neurological disorders[51]

bioRxiv

- Botía, JA, Guelfi, S, Zhang, D, D'Sa, K, Reynolds, RH et al.
- Role: Adviser. Helped interpret the output of machine-learning-based models to distinguish disease genes from non-disease genes.