

Sam Buckberry, PhD

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About

I am a biologist who works at the intersection of epigenetics, genomics, bioinformatics, medical research and public health.

I currently lead the Epigenetics program in Indigenous Genomics at the Telethon Kids Institute. Here, my research is focused on using genomics technologies and cutting-edge data analytics to advance chronic disease prediction, prevention, and treatment. I also work on innovating ways to implement best practices in indigenous data sovereignty, privacy and security into state-of-the-art data systems for public and precision health.

From 2015-2021, I was a post-doctoral researcher in the Lister Lab where my research focused on DNA methylation and its role in epigenetic gene regulation. This included characterising the epigenome in stem cell reprogramming and human brain development and ageing. Here I worked extensively with whole genome DNA methylation, ATAC-seq, ChIP-seq and RNA-seq data.

I am impact-focused and enjoy developing bespoke analysis frameworks for complex biological questions. I have broad experience in molecular and cell biology, scientific programming, high-performance computing, statistics and data visualisation.

Current roles

Head, Epigenetics | Indigenous Genomics

Telethon Kids Institute

Senior Lecturer | Australian National University

ANU College of Health and Medicine

Education

Doctor of Philosophy (2012-2015)

The University of Adelaide

Thesis: An integrative analysis of the human placental transcriptome.

Awarded the Dean's commendation for doctoral thesis excellence.

Bachelor of Health Science - Honours (2011)

The University of Adelaide

Awarded first class honours

Bachelor of Science (2008-2010)

Flinders University

Extended Major in Biological Sciences

Awarded the Chancellor's Commendation

Postdoctoral work history

Telethon Kids Institute | Australian National University (2022-present) Head, Epigenetics | Indigenous Genomics
Black Ochre Data Labs

- **Project lead:** Whole genome DNA methylation profiling of Type-2 diabetes in the prospective longitudinal PROPHECY cohort (n=1,400) for precision medicine
- **Project lead:** MRFF Genomics Health Futures Mission Grant (2022-2024) Establishing epigenetic biomarkers in Indigenous Australians for precision health
- **Architect and developer:** Computational systems for Indigenous data sovereignty
- **Contributor:** Australian Alliance for Indigenous Genomics (ALIGN)

The University of Western Australia (2015-2021) Postdoctoral Research Fellow
Lister Lab | Epigenetics and Genomics

- **Project lead for stem cell epigenetics program**
 - Project lead for correcting epigenetic memory in human iPS cells. See **Buckberry et al. (2023)** *Nature*
 - Developed a patent for improving human iPS cell reprogramming. See *Methods of Reprogramming a Cell* (2019)
 - Project lead for multi-omics time-course profiling of iPS cell reprogramming. See Knaupp & Buckberry et al. (2017) *Cell Stem Cell* - Contributor to epigenetic profiling of iPSC reprogramming published in *Nature* and *Cell Reports*
- **Contributor to epigenetics of brain development and function program**
 - Key contributions to work on Alzheimer's disease, epigenetics of brain development, evolution of neuron-specific DNA methylation published in *Cell*, *Nature Neuroscience*, *Nature Communications*, *Nature Ecology & Evolution*, *Cell Reports*
- **Developer:** Custom analysis pipelines for MethylC-seq, ATAC-seq, ChIP-seq, RNA-seq and data integration.
- **Lecturer:** Master of Biotechnology, Masters of Bioinformatics
- **Supervision:** 1 Honours, 2 Masters, 1 PhD

Service

Australasian Genomics Technology Association (AGTA) - Executive Committee (2017-)

WA Department of Health Human Research Ethics Committee (2020-)

Grant reviewer: NHMRC, New Zealand Heart Foundation, WA Department of Health

Journal reviewer: BMC Genomics, Genome Research, Clinical Epigenetics, Molecular Therapy, Oncogenesis, Placenta, F1000 Research, Scientific Reports, among others.

STEM mentor: School of Indigenous Studies, The University of Western Australia (2018)

Research funding

- MRFF Early to Mid-Career Researchers: The missing heritability of human disease: discovery to implementation (2022-2027) **\$4,877,532**
- MRFF Genomics Health Futures Mission Grant (2022-2024) Establishing epigenetic biomarkers in Indigenous Australians for precision health **\$991,506**

- WA Department of Health Merit Award (2020) **BUCKBERRY S (CIA) \$95,000**
- NHMRC-ARC Dementia Research Development Fellowship Grant (2016-2019): The Role of the neuronal methylome in natural brain ageing and the progression of Alzheimer’s Disease (APP1111206). **BUCKBERRY S (CIA) \$584,644**
- Robinson Research Institute Seed Funding (2015-2016): A multi-faceted approach to understanding the consequences of diabetes in early pregnancy. **\$25,000**
- Robinson Research Institute Seed Funding (2014-2015): Investigating new mechanisms regulating transcription in the pre-implantation embryo. **\$25,000**

Awards and recognition

- Finalist (1 of 4): Woodside Early Career Scientist of the Year (2020), WA Premier’s Science Awards
- Raine Research Prize (2019) - Raine Medical Research Foundation
- People’s Choice Award - Epigenetics Consortium of South Australia annual research meeting (2019)
- Ana Africh Stem Cell Research Travel Scholarship (2019)
- Best Oral Presentation by Early Career Researcher at Australasian Genomic Technologies Association (AGTA) conference (2016)
- NHMRC-ARC Dementia Research Fellowship (2016-2019)
- NHMRC Peter Doherty Biomedical Early Career Fellowship (2016-2019) *relinquished*
- Y.W Loke Young Investigator Award, IFPA Conference (2015)
- Dean’s Commendation for Doctoral Thesis Excellence (2015)
- Promega PhD Student Award, Lorne Genome Conference (2014)
- Healthy Development Adelaide & Channel 7 Children’s Research Foundation PhD Scholarship (2012-2015)
- The Robinson Research Institute Petrucco Honours Scholarship (2011)
- AR Riddle Honours Grant, The University of Adelaide (2011)
- Flinders University Chancellors Commendation (2011)
- Flinders University Chancellors commendation (2010)
- Flinders University Summer Research Scholarship (2010)
- Commonwealth of Australia Education Scholarship (2008-2010)

Patents

Buckberry S, Liu X, Polo JM & Lister R. Methods of Reprogramming a Cell. International patent PCT/AU2019/051296 World Intellectual Property Organization (WIPO)

Media publications and public engagement

Buckberry S A ‘memory wipe’ for stem cells may be the key to better therapies. *The Conversation* 17 Aug 2023 (re-published >25 outlets)

Using Big Data to Understand the Human Brain Pint of Science Festival (2018).

Buckberry S & Roberts CT. *Why are males more at risk in the womb?* **Australasian Science**, November 2014 (cover story)

Academic publications

Buckberry S, Liu X, Poppe D, Tan JP, Sun G, Chen J, Nguyen TV, de Mendoza A, Pflueger J, Frazer T, Vargas-Landín DB, Paynter JM, Smits N, Liu N, Ouyang JF, Rossello FJ, Chy HS, Rackham OJL, Laslett

AL, Breen J, Faulkner GJ, Nefzger CM, Polo JM & Lister R (2023). Transient naive reprogramming corrects hiPS cells functionally and epigenetically. *Nature* 620, 863–872

Herring CA, Simmons RK, Freytag S, Poppe D, Moffet JJD, Pflueger J, **Buckberry S**, Vargas-Landin DB, Clément O, Echeverría EG, Sutton GJ, Alvarez-Franco A, Hou R, Pflueger C, McDonald K, Polo JM, Forrest ARR, Nowak AK, Voineagu I, Martelotto L & Lister R (2022). Human prefrontal cortex gene regulatory dynamics from gestation to adulthood at single-cell resolution. *Cell* 185, 4428–4447.e28.

de Mendoza A, Nguyen TV, Ford E, Poppe D, **Buckberry S**, Pflueger J, Grimmer MR, Stolzenburg S, Bogdanovic O, Oshlack A, Farnham PJ, Blancafort P & Lister R (2022). Large-scale manipulation of promoter DNA methylation reveals context-specific transcriptional responses and stability. *Genome Biology* 23, 163

Grubman A, Choo XY, Chew G, Ouyang JF, Sun G, Croft NP, Rossello FJ, Simmons R, **Buckberry S**, Landin DV, Pflueger J, Vandekolk TH, Abay Z, Zhou Y, Liu X, Chen J, Larcombe M, Haynes JM, McLean C, Williams S, Chai SY, Wilson T, Lister R, Pouton CW, Purcell AW, Rackham OJL, Petretto E & Polo JM (2021). Transcriptional signature in microglia associated with AB plaque phagocytosis. *Nature Communications*. 12, 3015

de Mendoza A, Poppe D, **Buckberry S**, Pflueger J, Albertin CB, Daish T, Bertrand S, de la Calle-Mustienes E, Gómez-Skarmeta JL, Nery JR, Ecker JR, Baer B, Ragsdale CW, Grützner F, Escriva H, Venkatesh B, Bogdanovic O & Lister R (2021). The emergence of the brain non-CpG methylation system in vertebrates. *Nature Ecology & Evolution*

Liu X, Ouyang JF, Rossello FJ, Tan JP, Davidson KC, Valdes DS, Schröder J, Sun YBY, Chen J, Knaupp AS, Sun G, Chy HS, Huang Z, Pflueger J, Firas J, Tano V, **Buckberry S**, Paynter J.M, Larcombe MR, Poppe D, Choo XY, O’Brien CM, Pastor WA, Chen D, Leichter AL, Naeem H, Tripathi P, Das PP, Grubman A, Powell DR, Laslett AL, David L, Nilsson SK, Clark AT, Lister R, Nefzger CM, Martelotto LG, Rackham OJL & Polo JM (2020). Reprogramming roadmap reveals route to human induced trophoblast stem cells. *Nature* 586, 101–107

Grubman A, Chew G, Ouyang JF, Sun G, Choo XY, McLean C, Simmons R, **Buckberry S**, Landin DV, Poppe D, Pflueger J, Lister R, Rackham OJ, Petretto E and Polo JM (2019). A single-cell atlas of entorhinal cortex from individuals with Alzheimer’s disease reveals cell-type-specific gene expression regulation. *Nature Neuroscience* 22:2087–2097

de Mendoza A, Hatleberg WL, Pang K, Leininger S, Bogdanovic O, Pflueger J, **Buckberry S**, Technau U, Hejnlol A, Adamska M, Degnan BM, Degnan SM and Lister R (2019). Convergent evolution of a vertebrate-like methylome in a marine sponge. *Nature Ecology and Evolution* 3;10:1464-1473

de Mendoza A, Bonnet A, Vargas-Landin DB, Ji N, Hong F, Yang F, Li L, Hori K, Pflueger J, **Buckberry S**, Ohta H, Rosic N, Lesage P, Lin S and Lister R (2018). Recurrent acquisition of cytosine methyltransferases into eukaryotic retrotransposons. *Nature Communications* 9;1:1341

Stuart T, **Buckberry S**, Lister R (2018). Approaches for the Analysis and Interpretation of Whole Genome Bisulfite Sequencing Data. *Methods in Molecular Biology* 1767:299-310 - **Book chapter**

Mayne BT, Leemaqz SY, **Buckberry S**, Rodriguez Lopez CM, Roberts CT, Bianco-Miotto T and Breen J (2018). msgbsR: An R package for analysing methylation-sensitive restriction enzyme sequencing data. *Scientific Reports* 8;1:2190

Knaupp AS1, **Buckberry S**1, Pflueger J, Lim SM, Ford E, Larcombe MR, Rossello FJ, de Mendoza A, Alaei S, Firas J, Holmes ML, Nair SS, Clark SJ, Nefzger CM, Lister R and Polo JM (2017). Transient and permanent reconfiguration of chromatin and transcription factor occupancy drive reprogramming. *Cell Stem Cell* 21, 1-12 **Co-first author**

Nefzger CM, Rossello FJ, Chen J, Liu X, Knaupp AS, Firas J, Paynter J, Pflueger J, **Buckberry S**, Lim SM, Williams B, Alaei S, Keshav Faye-Chauhan K, Nilsson SK, Lister R, Ramialison M, Powel DR, Rackham OJL and Polo JM (2017). Cell type of origin dictates the route to pluripotency. *Cell Reports* 21

- Wilson RL, Leemaqz SY, Goh Z, McAninch D, Jankovic-Karasoulos T, Leghi GE, Phillips JA, Colafella KM, Tran C, O’Leary S, **Buckberry S**, Pederson S, Robertson SA, Bianco-Miotto T, Roberts CT (2017). Zinc is a critical regulator of placental morphogenesis and maternal hemodynamics during pregnancy in mice. *Scientific Reports* 9;7(1):15137
- Valensisi C, Andrus C, **Buckberry S**, Doni Jayavelu N, Lund RJ, Lister R and Hawkins RD (2017). Epigenomic Landscapes of hESC-Derived Neural Rosettes: Modeling Neural Tube Formation and Diseases. *Cell Reports* 8:20
- Lumb R, **Buckberry S**, Secker G, Lawrence D and Schwarz Q (2017). Transcriptome profiling reveals expression signatures of cranial neural crest cells arising from different axial levels. *BMC Developmental Biology* 17:5
- Buckberry S**, Bianco-Miotto T, Bent SJ, Clifton V, Shoubridge C, Shankar K and Roberts CT (2017). Placental transcriptome co-expression analysis reveals conserved regulatory programs across gestation. *BMC Genomics* 18:10
- Mayne B, Bianco-Miotto T, **Buckberry S**, Breen J, Clifton V, Shoubridge C, Roberts CT (2016). Large scale gene expression meta-analysis reveals tissue-specific, sex-biased gene expression in humans. *Frontiers in Genetics*
- Hight AR, **Buckberry S**, Mayne BT, Khoda SM, Bianco-Miotto T, Roberts CT (2016). First trimester trophoblasts forming endothelial-like tubes in vitro emulate a ‘blood vessel development’ gene expression profile. *Gene Expression Patterns*
- Bianco-Miotto T, Mayne B, **Buckberry S**, Breen J, Rodriguez Lopez C, Roberts CT (2016). Recent progress towards understanding the role of DNA methylation in human placental development. *Reproduction*
- Hight HR, Khoda SM, **Buckberry S**, Leemaqz S, Bianco-Miotto T, Harrington E, Ricciardelli C & Roberts CT (2015). Hypoxia induced HIF-1/HIF-2 activity alters trophoblast transcriptional regulation and promotes invasion. *European Journal of Cell Biology* 94(12):589-602.
- Buckberry S**, Spronk F, Wilson RL, Laurence JA, Bianco-Miotto T, Leemaqz S, O’Leary S, Anderson PH & Roberts CT (2015). The effect of Vdr gene ablation on global gene expression in the mouse placenta. *Genomics Data* 6:72-73
- Wilson RL1, **Buckberry S**1, Spronk F1, Laurence JA, Leemaqz S, O’Leary S, Bianco-Miotto T, Du J, Anderson PH & Roberts CT (2015). Vitamin D Receptor Gene Ablation in the Conceptus Has Limited Effects on Placental Morphology, Function and Pregnancy Outcome. *PLoS ONE* 10(6):e0131287. 1 *These authors contributed equally to this work.*
- Buckberry S**, Bianco-Miotto T, Bent SJ, Dekker GA & Roberts CT (2014). Integrative transcriptome meta-analysis reveals widespread sex-biased gene expression at the human fetal-maternal interface. *Molecular Human Reproduction* 20(8):810-9
- Buckberry S**, Bent SJ, Bianco-Miotto T & Roberts CT (2014). MassiR: a method for predicting the sex of samples in gene expression microarray datasets. *Bioinformatics* 30(14):2084-5
- Buckberry S**, Bianco-Miotto T & Roberts CT (2014). Imprinted and X-linked non-coding RNAs as potential regulators of human placental function, *Epigenetics* 9(1):81-9
- Gatford KL, Heinemann GK, Thompson SD, Zhang JV, **Buckberry S**, Owens J, Dekker GA & Roberts CT (2014). Circulating IGF1 and IGF2 and SNP genotypes in pregnant and non-pregnant women and men. *Endocrine Connections* 3(3):138–149
- Buckberry S**, Bianco-Miotto T, Hiendler S & Roberts CT (2012). Quantitative Allele-Specific Expression and DNA Methylation Analysis of H19, IGF2 and IGF2R in the Human Placenta across Gestation Reveals H19 Imprinting Plasticity. *PLoS ONE* 7(12):e51210
- Buckberry S** & Burke da Silva K (2012). Evolution: improving the understanding of undergraduate students with an active pedagogical approach. *Evolution, Education & Outreach* 5(2):266-273

Select presentations

- **Invited pannelist - AusBioTech 2022** Extract maximum value from your data with high-performance computing.
- Overcoming the epigenetic barriers to cellular reprogramming. *South Australia Health and Medical Research Institute 2019. Adelaide, Australia. Invited presentation.*
- Profiling the shifting epigenetic landscape during human reprogramming. **People's Choice Award, Epigenetics Consortium of South Australia Annual Research Meeting 2019. Adelaide Australia. Invited presentation.**
- Characterising epigenome dynamics during the reprogramming of somatic cells to iPS cells. *Lorne Genome Conference 2017. Lorne, Australia.*
- Characterising epigenome dynamics during the reprogramming of somatic cells to iPS cells. **Best ECR oral presentation, Australasian Genomic Technologies Association (AGTA) Conference 2016. Auckland, New Zealand.**
- Moving towards integrative epigenomic analyses. *International Federation of Placenta Associations (IFPA) Conference 2015. Brisbane, Australia. Invited presentation.*
- Integrative analysis of placental transcriptome organization reveals highly conserved regulatory programs and points toward a preeclampsia gene cluster. **New investigator award session. International Federation of Placenta Associations (IFPA) Conference 2015. Brisbane, Australia.**
- Integrative transcriptome meta-analysis reveals widespread sex-biased gene expression patterns in human extra-embryonic tissue. **Promega PhD Student Award, Lorne Genome Conference 2014. Lorne Victoria, Australia.**
- What can big data tell us about big tummies? *Robinson Research Institute Symposium 2014. Adelaide, Australia. Invited presentation.*
- Using RNA sequencing to comprehensively profile the transcriptomes of reproductive tissues. *Society for Reproductive Biology Conference 2014. Melbourne, Australia. Invited presentation.*
- Sexual inequality begins *in utero* with genome-wide differences in the human placental transcriptome. **ANZPRA new investigator award session, Society for Reproductive Biology Conference 2013. Sydney, Australia.**
- Using RNA sequencing to quantify placental transcriptomes. *Methods in Reproductive Biology Symposium 2013. Mercy Hospital for Women, Melbourne, Australia. Invited presentation.*
- Sex-biased gene expression at the human fetal-maternal interface. *Australian Society for Medical Research Annual Meeting 2013. Adelaide, Australia.*
- Quantitative allele-specific expression analysis reveals imprinting plasticity of the H19 lincRNA. **ANZPRA new investigator award session, Society for Reproductive Biology Conference 2012. Gold Coast, Australia.**
- Allele-specific expression of H19 and IGF2 in the human placenta across gestation. **Honours award session, Australian Society for Medical Research Annual Meeting 2012. Adelaide, Australia.**

Teaching

- Lecturer, Masters of Biotechnology, School of Chemistry and Biochemistry, The University of Western Australia (2019, 2020, 2021).

- Lecturer, *The Objectives and Applications of Genomics 2017* (SCIE4001). School of Chemistry and Biochemistry, The University of Western Australia (2017).
- Teaching Assistant for RNA-Seq analysis workshops at BioInfoSummer Conference 2013, Australian Mathematical Sciences Institute and the University of Adelaide.
- Teaching Assistant for the Bioinformatics Workshops 2013, the University of Adelaide.
- Tutor and Coordinator for undergraduate biology Peer Assisted Study Sessions (PASS), Flinders University (2010–2011).