RHETT MARCHANT

Researcher & Data Analyst

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PROFESSIONAL EXPERIENCE

GENETICS RESEARCHER (DOCTORAL STUDENT) **Westmead, NSW**
The University of Sydney, Faculty of Medicine and Health **March 2023 - October 2024**

- Designed and built end-to-end analysis pipelines using bash, R, and Python, transforming raw genetic data into actionable reports, cutting turn-around times from months to weeks for over 160 patients/families.
- Synthesised complex genetic and clinical data to resolve uncertain findings, directly contributing to accurate diagnoses in 3 of 7 families with previously unknown neuromuscular disorders.
- Built a web tool to visualise and interpret genetic mutations, enabling non-technical users to explore patterns and assess mutation impact through an interactive, intuitive interface.
- Built relationships across disciplines—software, data, clinical, academic—to translate complex results into meaningful insights, supporting decision-making and cross-functional understanding.

RESEARCH ASSISTANT **Westmead, NSW**
Children's Medical Research Institute, Functional Neuromics **July 2021 - February 2023**

- Led an analysis of a decade's worth of genetic testing data, integrating multiple data sources and clinical records to better understand diagnostic outcomes and inform future clinical practice.
- Developed and applied clear inclusion criteria, cleaned and transformed fragmented datasets (Excel, reports), and identified patterns in diagnostic trajectories, methods, and mutation types.
- Designed clear visualisations and summary figures to communicate complex trends to a broad clinical audience; managed correspondence and data clarification with over 50 contributors.
- Wrote and revised a report manuscript, resulting in publication in *Annals of Clinical and Translational Neurology*, contributing to the broader understanding of genetic disease diagnostics.

RESEARCH STUDENT **Westmead, NSW**
The University of Sydney, Faculty of Medicine and Health **July 2020 - June 2021**

- Developed and implemented a reproducible analysis pipeline using R and proprietary software for processing complex genetic data, diagnosing 2 of a cohort of 5 undiagnosed families and discovering a novel disease gene, initialising ongoing clinical research.

EDUCATION

Bachelor of Advanced Studies (Applied Medical Science Honours) **The University of Sydney, Sydney, NSW**
Honours Class I and The University Medal **July 2020 - June 2021**

Bachelor of Science in Biomedical Science **University of Technology Sydney, Sydney, NSW**
Grade: 3.85/4.00 GPA **March 2017 - June 2020**

PROFESSIONAL COURSES

Operational Analysis of Suspicious Transaction Reports (eLearning), Basel Institute on Governance **April 2025**
Open-Source Intelligence (eLearning), Basel Institute on Governance **April 2025**
Combating Terrorism Financing (eLearning), Basel Institute on Governance **April 2025**

SKILLS & LANGUAGES

- Analysis Skills: Hypothesis Generation, Pattern Recognition, Problem Solving, Critical Thinking
- Communication Skills: Public Speaking, Community Engagement, Academic Writing, Report Preparation
- Technical Skills: Excel, Python, R, SQL, HTML/CSS, bash
- Languages: English (Native), Spanish (Intermediate Proficiency)

PUBLICATIONS

Marchant, R.G., Bryen, S.J., Bahlo, M., Cairns, A., Chao, K.R., Corbett, A., et al. Genome and RNA sequencing boost neuromuscular diagnoses to 62% from 34% with exome sequencing alone. Ann Clin Transl Neurol 11, 1250-1266 (2024). https://doi.org/10.1002/acn3.52041

Zhang, K.Y., Joshi, H., **Marchant, R.G.**, Bryen, S.J., Dawes, R., Yuen, M., Cooper, S.T., Evesson, F.J. Refining clinically relevant parameters for mis-splicing risk in shortened introns with donor-to-branchpoint space constraint. Eur J Hum Genet 32, 972–979 (2024). https://doi.org/10.1038/s41431-024-01632-9

Dawes, R., Bournazos, A.M., Bryen, S.J., Bommireddipalli, S., **Marchant, R.G.**, Joshi, H., and Cooper, S.T. SpliceVault predicts the precise nature of variant-associated mis-splicing. Nat Genet 55, 324–332 (2023). https://doi.org/10.1038/s41588-022-01293-8