



Project Title: Improving clinical genomic diagnosis using Machine Learning, multi-omics and other algorithmic optimisation techniques		Code: NCS2
Host School / Institute: Northern Clinical School/ Kolling Institute		Address: Kinghorn Centre for Clinical Genomics, Garvan Institute of Medical Research, Darlinghurst NSW
Certificates & Clearances required: No		
Primary Supervisor: Prof Leslie Burnett		
Phone: 02 9355 5842	Email: leslie.burnett@sydney.edu.au	
Co-Supervisor/team: The student will be directly supervised by Prof. Leslie Burnett. The project will involve interaction with clinicians, scientists and bioinformaticians who are colleagues and collaborators of Prof. Burnett within the Garvan Institute of Medical Research.		
Project Type: Data Analysis; Laboratory based; Clinical		
Project Category: Genetics; Bioinformatics		
Skills / Attributes of a successful student: Interest in genetics/genomics. Basic understanding of Cell Biology/Genetics. Basic skills in working with spreadsheets, simple databases, etc. Understanding or experience of bioinformatics or computer science concepts would be an advantage.		
Project Keywords: Genomics; Rare diseases; Pathology; Bioinformatics		
<p>Project Description: The introduction of high-throughput genomics ('next generation sequencing') has revolutionised clinical diagnostic practice. Performing whole exome or whole genome analysis is increasingly becoming an early, or even a first-line, investigation in many rare diseases diagnoses. However, analysis and interpretation of large data-sets such as human genomes are major challenges requiring bioinformatics and professional resources in limited supply and there is need for bioinformatic tools that simplify the diagnostic process.</p> <p>We have recently developed an algorithm (named "Orrery") for whole genome analysis, whose performance equals state-of-the-art clinical genomic diagnosis by human experts. It has just been presented (June 2019) to the international genetics community, and has also just been accredited for human clinical diagnostic use.</p> <p>This Summer Scholarship project will involve joining the team who developed this Orrery algorithm. Although the Orrery algorithm already equals the diagnostic performance of leading diagnostic clinicians, there are opportunities to improve it further. The Scholarship project will involve: learning how human whole genome analysis is performed; applying the Orrery algorithm to curated sets of human genomes with known diagnoses; extending the Orrery algorithm by adding new capabilities (such as Copy Number Variants, Structural Variants, Splice Site predictors, and intronic splice site regions); and evaluating the diagnostic performance of these algorithms with the performance of human genetic experts.</p> <p>This Summer Scholarship project represents an opportunity to become exposed to the absolute "cutting edge" of diagnostic clinical genomics, and to make a valuable contribution to the development of new diagnostic tools and techniques.</p>		