Available Projects for Intercalated BSc (iBSc) in Medical Science

QUEEN'S UNIVERSITY BELFAST

2021-2022

Contents

Projects Hosted by the Wellcome-Wolfson Institute For Experimental Medicine

Project Title	**Exploration of Receptor Collaboration During Aspergillus Infections**	
Supervisor(s)	1. Selinda Orr 2. Rebecca Coll	
School / Centre	WWIEM	
Supervisor's Contact Details	Email: S.Orr@qub.ac.uk	Tel: +44(0)2890976341
Project is suitable for the following scholarship applications	*General awards* Wolfson Foundation	
Background information:	Fungal infections including invasive aspergillosis (IA) kill more people (~1.5M) per year than TB or malaria. IA-related mortality rates are unacceptably high (30-95%) in immunosuppressed patients such as acute myeloid leukaemia patients, patients undergoing chemotherapy or stem cell transplant patients. The development of novel immunotherapies is urgently required. C-type lectin-like receptors (CLRs) are important for anti-fungal immunity and represent potential targets for immunotherapy. This project will determine the collective role of two CLRs, Dectin-1 and Dectin-2, during anti-*Aspergillus* immunity. Dectin-1 recognizes ß-glucans in fungal cell walls while Dectin-2 recognizes mannans in fungal cell walls. Our preliminary data indicates that Dectin-1 and Dectin-2 induce synergistic responses to *Aspergillus*, however these synergistic responses have not been explored in depth and the mechanism mediating this synergism is currently unknown. Importantly, exploitation of collaborative/synergistic responses by therapeutic receptor co-stimulation has led to a cure of chronic chromoblastomycosis in mice. Here, we propose to define collaborative/synergistic CLR anti-*Aspergillus* responses and signalling pathways in order to determine whether CLR modulation will improve clearance of various life-threatening fungal pathogens.	
Aims / objectives	1. Determine which *Aspergillus*-induced cytokine responses are mediated by synergism between Dectin-1 and Dectin-2 using cells from novel multi-CLR-deficient mice. 2. Determine which signalling pathways mediate these responses, using a newly generated RNAseq dataset. 3. Confirm the role of Dectin-1 and Dectin-2 in the signalling pathways identified from the RNAseq dataset using western blotting analysis.	

Techniques employed:	Cell culture ELISAs RNAseq Analysis Western blotting

Project Title	**Markers of myofibroblast conversion and associated micoRNAs in a new *in vitro* model of fibrotic interstitial lung disease.**	
Supervisor(s)	1. Bettina Schock (*) 2. Fionnuala Lundy (*) 3. Fiona Furlong (#)	
School / Centre	(*) SMDBS, Wellcome-Wolfson Institute for Experimental Medicine (#) School of Pharmacy	
Principal Supervisor's Contact Details	Email: b.schock@qub.ac.uk	Tel: 02890972558
Project is suitable for the following scholarship applications	*General awards* Wolfson Foundation	
Background information:	Fibrosing interstitial lung diseases (ILDs) describe a large group of >200 parenchymal pulmonary disorders (Cottin *et al.,* 2018). Although rare, a proportion of patients develop a progressive-fibrosing phenotype characterised by progressive lung scarring, lung function decline, limited response to immunomodulatory therapies, decreased quality of life and premature death. Transforming growth factor beta 1 (TGF-β1) is a major factor leading to the fibrotic conversion to myofibroblasts. However, in cell culture (*in vitro*) this is reversible when TGF-β1 is withdrawn (Sarrazy *et al.*, 2011), suggesting that additional factors are required *in vivo* to develop the persistent fibrotic myofibroblast. In systemic sclerosis, ILD (SSc-ILD) is a common complication and a leading cause of morbidity and mortality. In SSc-ILD, microvascular injury (hypoxia), is the initiating factor leading to an over-exuberant immune response with high TGF-β1 secretion. Using lung fibroblasts, we have used subsequent exposures of TGF-β1 and hypoxia and have observed increased/faster proliferation of the lung fibroblasts *in vitro.* In this project we will characterise the new model by analysing (mRNA and protein) of myofibroblast specific markers and will investigate epigenetic factors associated with this fibrotic phenotype (microRNAs). Better in vitro models of fibrotic lung diseases will increase our knowledge about the pathological process, and this is mandatory to develop more efficient pharmacological therapies.	
Aims / objectives	Characterisation of a modified *in vitro* model of lung fibrosis using subsequent exposures of TGF-β1 and hypoxia. This will be achieved by: 1. Analysing myofibroblast specific markers (mRNA and protein) 2. Investigating epigenetic factors associated with the fibrotic phenotype (microRNAs)	
Techniques employed:	- Cell culture of human lung fibroblasts - TGFβ1-primed fibroblasts will be exposed to normoxia/hypoxia (1%O_2) in the presence and absence of TGFβ1 - Characterisation of the fibroblast phenotype by analysing (mRNA and protein) for α-SMA, Focal Adhesion Kinase, TGFβ-1 and IFN-ɣ. - Investigating microRNAs associated with this fibrotic phenotype (microRNA gene array and confirmation of candidate microRNA expression by RT-PCR)	

Project Title	**Investigating the influence of oxidative stress on endothelial progenitor cell function**	
Supervisor(s)	1. Professor David Grieve 2. Dr Karla O'Neill	
School / Centre	SMDBS / Wellcome-Wolfson Institute for Experimental Medicine	
Principal Supervisor's Contact Details	Email: d.grieve@qub.ac.uk	Tel: 028 9097 6468
Project is suitable for the following scholarship applications	*General awards* Wolfson Foundation	
Background information:	Impaired angiogenesis is known to influence the progression of ischaemic cardiovascular disease. Recent attention has focused on the therapeutic potential of endothelial progenitor cells (EPCs), which are mobilised by ischaemia and are important in vascular homeostasis. Our group has characterised a distinct EPC subtype, termed endothelial colony-forming cells (ECFCs), with well-defined endothelial progenitor properties which promote new blood vessel formation in both health and disease. Oxidative stress, and specifically NADPH oxidases, is known to play a key role in cardiovascular disease and emerging evidence indicates that it is also a key regulator of EPC function. Specifically, we have shown that ECFCs are influenced by oxidative stress, display differential gene expression compared to mature endothelial cells, and are modulated by hypoxia which is a characteristic feature of the ischaemic microenvironment (O'Neill et al, Cardiovasc Res 2020).	
Aims / objectives	This project therefore aims to investigate the specific influence of oxidative stress and NADPH oxidases on in vitro ECFC function. It is hoped that the results will identify key pathways which may become dysregulated in disease and could represent potential targets to enhance the reparative capacity of these cells and their clear potential for the treatment of ischaemic cardiovascular disease.	
Techniques employed:	In order to characterise the effects of oxidative stress and NADPH oxidases on ECFC function, studies will be undertaken in cultured cells treated with pro-oxidant compounds in the presence or absence of specific inhibitors of candidate pathways or after genetic manipulation. Expression of key signalling genes will be quantified by real-time RT-PCR and/or western blot and in vitro ECFC tube formation assays will be performed to assess functional effects. The student can realistically expect to make an important contribution to ongoing ECFC research which will be acknowledged through manuscript co-authorship.	

Projects Hosted by the Patrick G. Johnston Centre for Cancer Research

Project Title	**Data-driven precision medicine in Bone Health management for patients with Metastatic Prostate Cancer**	
Supervisor(s)	1. Prof Suneil Jain 2. Prof Joe O'Sullivan 3. Ethna McFerran, Post-Doctoral Fellow – Cancer Health Economics	
School / Centre	PJGCCR	
Principal Supervisor's Contact Details	Email: s.jain@qub.ac.uk	Tel: ext; 2180
Project is suitable for the following scholarship applications	*General awards* Wolfson Foundation	
Background information:	Technological advancements in the availability of data play an important role in health economics and outcomes research. Where precision medicine applications allow focus on the current use of resources, economic evaluations can help discover and align treatment pathways with the highest likelihood of treatment success and quality of life for specific patient clusters. Using existing patient-level data on the management of metastatic prostate cancer this project will examine the current practice approaches to patient bone health management. The student will work alongside clinicians and researchers to collate data on the utilization of supportive care therapies in metastatic prostate cancer bone health management from HSC records, they will work in a team helping to analyze the data to understand the different effects of existing prescribing practices on service use, and outline potential routes to service optimization. Technological advancements in the availability of data play an important role in health economics and outcomes research and where precision medicine applications allow focus on the current use of resources, economic evaluations can help discover and align treatment pathways with the highest likelihood of treatment success and quality of life for specific patient clusters. Potential for industry funding will be explored if required to meet the needs of this project.	
Aims / objectives	- Provide a literature review of the evidence on the cost-effectiveness of metastatic prostate cancer bone health management - Collect data on prescribing practices and accompanying service use for the management of bone health in metastatic prostate cancer - Analysis of data on prescribing practices and accompanying service use for the management of bone health in metastatic prostate cancer ○ examining effects by age, drug regimen, SES or other factors potentially affecting treatment decisions, including potential differences by baseline diagnostics such as PET/ MRI	
Techniques employed:	Literature Review Descriptive statistics Data mining Causal inference/regression analyses	

Projects Hosted by the Centre for Medical Education

Project Title	**A phenomenological study into 'gut instinct' experiences in simulation based education.**
Supervisor(s)	Prof Gerry Gormley[1], Dr Martina Kelly[2], Dr Paul Hamilton[1]
School / Centre	[1]Centre for Medical Education, SMDBS, QUB. [2]Calgary University, Canada
Supervisor's Contact Details	Email: g.gormley@qub.ac.uk Tel: MS teams
Project is suitable for the following scholarship applications	*General awards* Wolfson Foundation
Background information:	Simulation is a widely used modality in health profession education. In this experiential form of learning, students are provided with opportunities to develop their clinical skills and behaviours. Often in simulation, learners are extended beyond their level of competency in a guided and scaffolded approach. Beyond intellectual learning, simulation based education aims to develop behaviours and habits for real clinical practice. A psychological discourse dominates the literature in learner's experiences in simulation. Whilst this can offer important insights, it can fall short of providing a more holistic view of learner's experiences in simulation. There is increasing interest in the literature on bodily experiences in learning. Rather than the body being a *passive* medium, we actively engage our bodies in experiencing and making sense of the world we inhabit. Often such experiences can be tacit, pre-linguistic and embodied. By exploring such experiences of simulation - has potential to unearth insights that may help us remodel aspects of this constructed form of teaching in order to optimise learning.
Aims / objectives	The aim of this project is to explore learners embodied experiences in simulation based education.
Techniques employed:	In this project we will aim to explore medical students embodied experiences of simulation through a phenomenological approach. Interpretive phenomenology is a well-established approach to analysing first-person experiences and producing valid knowledge. Through qualitative analysis of first-person accounts, we will use phenomenology to examine medical students reported experiences as it presents to their consciousness. This will provide an appropriate conceptual framework for examining and interpreting the richness of their experiences. Moreover, our analysis will be informed by Merleau-Ponty's work of embodied experiences. This focuses not just on objective bodily experiences, but also on perceptual, intentional and embodied dimensions of experiences, embedded in their contexts. In terms of delivering on this project the following steps will be required • Perform a literature review • Develop an immersive simulation experience in collaboration with experience academics in the new QUB InterSim Centre. • Evaluate medical students experience of this learning experience using a phenomenological approach • Develop a protocol and have approved for ethics • Recruit 10 medical students to experience the learning modality and then conduct interviews. • Using video glasses – we will capture 'in the moment' experiences of medical students in a simulation based exercise.

Skinner J, Gormley GJ. Point of view filming and the elicitation interview. Perspect Med Educ. 2016 Aug;5(4):235-9. doi: 10.1007/s40037-016-0278-0.

- Thematically analysis these interviews using the template analysis approach (and PoV footage to elicit interviews)

In this project – the successful student will have the chance to work with experience academics and work towards presenting and publishing their work in academic journals.

Project Title	**Conversational adaptations when talking with visually impaired others. (Conversational Analysis)**	
Supervisor(s)	1. Michael Williams, Senior Lecturer, Queen's University of Belfast 2. Natalie Flint, Lecturer, University of Ulster	
School / Centre	Centre for Medical Education	
Principal Supervisor's Contact Details	Email: m.williams@qub.ac.uk	Tel: 0044 2890245133
Project is suitable for the following scholarship applications	*General awards* Wolfson Foundation	
Background information:	Conversation is fascinating, part of everyday life and taken for granted but with its own substructure, patterns and variations. A variety of resources are used to convey meaning and facilitate understanding in addition to words. Actions such as gestures, facial expressions, position and movement all play a role. However these cues are less easily available to people with visual impairment. Anecdotally, visually impaired people relate how sighted people become awkwardly *unnatural* in their talk when they realise their co-conversant is visually impaired! Unusual word choices, self-repairs, overlapping turns and other conversational 'trouble sources' may occur. Features such as departures from the norm, what is treated as problematic by participants and consequent adaptations as interactants try to save face *in the setting of visual impairment* have not however been formally studied. Conversation Analysis (CA) provides a forensic method to study conversation to examine what gets said, how it gets said and the impact that it has. In CA, social interaction is recorded and analysed with a detailed focus on what "gets done" by the talk. The key of the method is to understand what an utterance 'is doing'. A recent review of CA and communicative impairments (Wilkinson R, Research on Language and Social Interaction 2019;52(3):281-99) considers several conditions and clinical features (such as dysarthria, aphasia, hearing impairment and dementia), but not visual impairment. Studying natural interaction between visually impaired and sighted participants will help to catalogue some examples of interesting conversational features of talk in this setting, as well as engendering reflection and discussion on the topic when results are disseminated.	
Aims / objectives	To study interaction between sighted and visually impaired participants by analysing audio and video recordings of conversation.	
Techniques employed:	Conversation Analysis: transcription and analysis of video and audio recordings of conversation, collaborative work with other linguistic groups, consideration of the impact of the cause of visual impairment on interaction.	

Project Title	**Conversational adaptations when talking with visually impaired others. (Randomised Control Cross Over Trial)**	
Supervisor(s)	1. Michael Williams, Senior Lecturer, Queen's University of Belfast	
School / Centre	Centre for Medical Education	
Principal Supervisor's Contact Details	Email: m.williams@qub.ac.uk	Tel: 0044 2890245133
Project is suitable for the following scholarship applications	*General awards* Wolfson Foundation	
Background information:	Clinical skills are a core part of training of healthcare professionals. Learning by exposure to real patients forms the highest level of learning, replicating as it does real practice. However patients, particularly those with signs, are a finite resource. Thus many approaches are taken to allow rehearsal of skills and recognition of what is normal and what is abnormal, including practising on peers, on simulated patients and on mannequins. Virtual reality (VR) offers a new way of learning clinical skills. An app has been developed for the Oculus Quest, a VR Headset, in which the user can examine a range of pupillary abnormalities. This allows the user to be immersed in a clinical environment and perform the exact sequences of movements required to elicit pupil responses, and then interpret the signs they have seen. No evidence exists yet on the acceptability to learners and the educational effectiveness of this method.	
Aims / objectives	To determine the acceptability to learners and the educational effectiveness of the ophthalmology Oculus Quest VR app (the 'Virtual Clinical Classroom').	
Techniques employed:	A randomised controlled cross over trial, with outcomes of knowledge and skills in ophthalmic examination.	

Project Title	**Medical student learning during early clinical contact**
Supervisor(s)	1. Dr Grainne Kearney 2. Dr Richard Conn 3. Dr Helen Reid
School / Centre	Centre for Medical Education
Supervisor's Contact Details	Email: g.kearney@qub.ac.uk Tel: 02890975840
Project is suitable for the following scholarship applications	*General awards* Wolfson Foundation
Background information:	"*He who studies medicine without books sails an uncharted sea, but he who studies medicine without patients does not go to sea at all*." William Osler Medical education increasingly focuses on "off the job" or desk based learning, reducing the available time for learners to be actively involved in the care of patients. One pedagogy which specifically looks at how medical students learn in the clinical workplace is Experience-based learning or ExBL. In 2019, an AMEE guide and related article were published on ExBL, representing the culmination of nearly two decades of research (1, 2). This large-scale literature search synthesised best evidence from around of the world on learning from real patients. ExBL focused on medical students in clinical years and so specifically excluded any early clinical contact that students may have. This project aims initially to look at the literature on how medical students learn in the clinical workplace, in the specific context of early clinical contact. Following this, the work will gauge medical students' experience of learning from patients during early clinical contact (facilitated through the Family Medicine module in Queen's University, Belfast). **References** 1. Dornan T, Conn R, Monaghan H, Kearney G, Gillespie H, Bennett D. Experience based learning (ExBL): Clinical teaching for the twenty-first century. AMEE Guide 129. Dundee: Association for Medical Education in Europe, 2019. 2. Dornan T, Conn R, Monaghan H, Kearney G, Gillespie H. Bennett D. Experience based learning (ExBL): Clinical teaching for the twenty-first century. *Med Teach* 2019;41(10):1098-1105.
Aims / objectives	Aim – to understand more about student learning in the clinical workplace during early clinical contact (Family Medicine). This aim will be achieved through the following objectives 1) Performing a literature review relating to this topic 2) Using qualitative data collection methods and data analytic techniques to explore medical students' learning in early clinical contact 3) Inform guidance around learning during early clinical contact 4) Prepare a paper for publication based on this research project
Techniques employed:	The student will perform a relevant literature review. The student will learn about a variety of qualitative data collection methods such as semi-structured interviews and focus groups, in order to select an appropriate method for this project. They will recruit and consent medicals students to take part in the study. The student will learn about the processes of thematic analysis and phenomenology when considering analysis of the data. The results of this study will be presented at conferences and prepared as a paper for academic publication. The results of this study will help bring about knowledge around learning in the clinical workplace during early clinical contact.

Project Title	**Improving the detection of pseudohyperkalaemia in the clinical biochemistry laboratory**	
Supervisor(s)	1. Dr Paul Hamilton 2. Dr Vikki O'Neill	
School / Centre	Centre for Medical Education	
Principal Supervisor's Contact Details	Email: paul.hamilton@qub.ac.uk	Tel: QUB Extension 8983 or via MS Teams
Project is suitable for the following scholarship applications	*General awards* Wolfson Foundation	
Background information:	The measurement of potassium is one of the commonest blood tests undertaken in healthcare settings. Hyperkalaemia (an elevated blood potassium concentration) is a medical emergency that can cause death due to alterations in cardiac conduction. Patients found to have hyperkalaemia may require life-saving emergency treatment in a hospital setting. Pseudohyperkalaemia is the term used to describe the false finding of a high potassium concentration in a blood sample when the true potassium level in the patient's bloodstream is normal. It can result from a variety of medical conditions (e.g. leukaemia) as well as problems that can arise during blood collection (e.g. repeated fist clenching during blood taking or a prolonged application of a tourniquet). It is not uncommon for patients to have blood taken in the community, only to be contacted later that day with a message to attend an emergency department for an urgent potassium re-check. This results in patient anxiety and inconvenience, and puts additional burden on emergency departments. Most often, the re-check of potassium reveals a completely normal result and the patient is discharged without intervention. There is therefore the need for a method of reliably differentiating between true hyperkalaemia and pseudohyperkalaemia.	
Aims / objectives	This project will involve the analysis of a large volume of information from the clinical biochemistry laboratory information system in the Belfast Health and Social Care Trust. Anonymised data will be analysed to identify patients who have been found to have true hyperkalaemia and pseudohyperkalaemia and a statistical exploration will be undertaken to investigate whether there are any factors that might differentiate between the two conditions. The laboratory are planning to introduce a new type of blood collection tube in 2021 which may reduce the incidence of pseudohyperkalaemia. The usefulness of this intervention will also be explored.	
Techniques employed:	Manipulation and organisation of large volumes of numerical biochemical data, introducing the student to the concept of 'Big Data' and its utility in healthcare settings. Hypothesis testing and statistical analysis to include the use of summary statistics, comparisons of groups, sensitivity/specificity analyses, and the construction of receiver operating characteristic curves. The student will have the opportunity of learning about the running of a hospital laboratory and will develop expertise in potassium disorders. The student will work closely with the two project supervisors. Dr Hamilton is a chemical pathologist and Dr O'Neill is a statistician. Both work within the Centre for Medical Education.	

Projects Hosted by the Centre for Public Health

Project Title	**Mediterranean diet and cognitive performance in older Northern Irish men: The PRIME study.**	
Supervisor(s)	1. Dr Bernadette McGuinness 2. Dr Claire McEvoy	
School / Centre	CPH	
Principal Supervisor's Contact Details	Email:b.mcguinness@qub.ac.uk	Tel:02890978959
Project is suitable for the following scholarship applications	*General awards* Wolfson Foundation	
Background information:	Diet and nutrient intake may influence cognitive health. The Mediterranean diet (MD), characterised by a high intake of fruits, vegetables, wholegrain, nuts, legumes and olive oil, a moderate intake of fish, poultry and alcohol and a low intake of red meat, is shown to have anti-inflammatory, metabolic and vascular benefits and hence, may be neuroprotective for dementia. Greater adherence to a MD is associated with a significant reduction in cognitive decline and dementia risk. However, research is based on observational evidence and findings are limited, firstly by the small number of studies conducted to date, and secondly by limitations and heterogeneity, both in the methods to determine MD adherence and in respect to cognitive outcome(s) assessed. Furthermore, little is known about relations between MD and cognitive function in non-Mediterranean populations. More robust evidence is required to comprehensively investigate relationships between MD and cognitive health, from the earliest through to the later stages of cognitive decline and dementia and in different population groups. In the PRIME study of 1400 Northern Irish men, baseline adherence to MD has already been determined using a Mediterranean Diet Score (MDS) derived from dietary data collected at baseline (1991-1994), and already measured biomarkers of nutritional status. Baseline cognition was assessed using the Mini Mental State Examination (MMSE). The PRIME cohort have been followed up for 25 years (2017-2020) for both repeated measures of diet and cognition. In addition, men have undergone health measurements by a trained research nurse to include: anthropometrics (weight, height, waist circumference and mid-upper arm circumference); physical function using the Bristol Activities of Daily Living Scale and muscle strength using grip strength dynamometer. Mood/depression was also assessed using the Geriatric Depression Scale short form. Stored serum is available from baseline and fresh blood samples were taken 2017-2020 to enable analysis of circulating levels of pro-and anti-inflammatory markers (including CRP, IL-1b, TNF-a, IL-6, IL-8, INF-g, IL-10, IL12p70, TGF-β.	
Aims / objectives	To determine the longitudinal association between MD adherence and cognitive decline, incident mild cognitive impairment and dementia over 25 years in older Northern Irish men To explore whether patterns of peripheral pro- and anti-inflammatory cytokines mediate the relationship between MD adherence and cognitive decline.	
Techniques employed:	Three dementia/AD specialists have determined a consensus diagnosis of MCI and/or dementia, using accepted clinical criteria. The student will calculate the MD score at baseline and year 25 to determine MD adherence in 900 PRIME men Appropriate statistical (logistic regression) analysis will be used to investigate the relationships between MD adherence and cognitive outcomes. The analysis will take into account the effects of possible confounders, including APOE ε4 status, lifestyle factors and known cardiovascular and metabolic risk factors. Fresh and stored serum will be analysed for levels of pro- and anti- inflammatory cytokines. Statistical meditation models will be used to investigate whether MD-cognition relations are mediated by systemic inflammation.	

Project Title	**Improving diagnosis for rare diseases**
Supervisor(s)	1. Professor Amy Jayne McKnight 2. Dr Shane McKee
School / Centre	Centre for Public Health, SMDBS, QUB in collaboration with the Regional Genetics Centre, Belfast Health and Social Care Trust
Supervisor's Contact Details	Email: a.j.mcknight@qub.ac.uk Tel: 02890 976359
Project is suitable for the following scholarship applications	*General awards* Wolfson Foundation *Subject-specific awards* Digestive disorders foundation
Background information:	Rare diseases represent a major public health problem affecting ~ $^{1}/_{17}$ individuals; that's more than 105,000 people in Northern Ireland alone. More than half of rare diseases affect children, with sadly almost one-third of rare disease patients not living to reach their 5th birthday. Finding a diagnosis for each person's rare disease is a major challenge, with many individuals never actually getting a name or effective treatment for their disease. Many rare diseases have a molecular cause, but better tools are required to enable accurate, efficient diagnosis. Our Northern Ireland Genomic Medicine Centre was launched several years ago with a primary focus on improving diagnosis for patients with rare diseases. We are using state-of-the-art, 'multi-omic' approaches (including high-density arrays, next generation sequencing, and 3D digital PCR machines) that increase the diagnostic yield by 40%, alongside developing associated new information tools to convey results to healthcare professionals, patients and their families / carers. **This intercalated project will combine data from three rare disease projects to help improve the diagnosis of rare disease in NI patients:** 1) The deciphering developmental disorders (DDD) project aims to advance clinical genetic practice for children with developmental disorders 2) The 100,000 genomes project (100 KGP) was an NHS transformational project that led to the development of the current NHS Genomic Medicine Service. 3) The GenOCEANIC (Genomics Open Core Engine for Accelerating Northern Ireland Care) project is helping integrate genomic medicine to mainstream clinical care for NI patients. Some further rare disease information may be found here: https://www.qub.ac.uk/sites/RareDisease/ with a few recent publications from our rare disease students including: 1. Crowe A, McAneney H, Morrison P, Cupples M, **McKnight AJ** (2020). A quick reference guide for rare disease: supporting rare disease management in general practice. British Journal of General Practice 70 (694): 260-261. 2. Kerr K, McAneney H, Smyth LJ, Bailie C, McKee S, **McKnight AJ** (2020). A scoping review and proposed workflow for multi-omic rare disease research. Orphanet Journal of Rare Disease 15(1):107. 3. McMullan J,Crowe A, Downes K, McAneney H, McKnight AJ (2020). Carer reported experiences: supporting someone with a rare disease. *In press*. Preprint available at medRxiv 2020.07.10.20150581. 4. McMullan J, Crowe A, Bailie C, Moore K, McMullan L, Shamandi N, McAneney H, **McKnight AJ** (2020). Improvements needed to support people living and working with a rare disease in Northern Ireland: current rare disease support perceived as inadequate. Orphanet Journal of Rare Disease 15(1):315. 5. Steers NJ, Li Y, ...McKnight AJ..... Kiryluk K (2019). Genomic Mismatch at LIMS1 Locus and Kidney Allograft Rejection. New England Journal of Medicine 380(20:1918-1928.
Aims / objectives	1) To continue analysis of genomic data obtained from the DDD project, to identify new genetic diagnoses for children in NI. 2) To link genomic diagnoses from whole genome sequencing data within the 100 KGP with our developing GenOCEANIC platform to explore if enhanced phenotyping will improve the diagnostic yield. 3) To perform molecular – phenotypic analysis 4) To work with our team to help develop and prioritise a workflow to improve the diagnosis of rare diseases across clinical specialties by linking whole genome sequencing data with detailed medical records within Northern Ireland.

Techniques employed:	Pedigree-based analysis of rare diseases Genetic-based analysis of next generation sequencing data Appropriately handling DNA samples for lab-based analyses in a clinical diagnostic environment Phenotype-based analysis from electronic medical healthcare records 'Big data' epidemiology & bioinformatic skills interpreting results Literature review & scientific writing skills Communication, presentation, and interpersonal skills working within a busy research team. **There is scope for a dedicated student to** help generate new lab-based data to investigate multi-omics for diagnosis using state-of-the-art experimental tools.