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2012

RESEARCH

TEMPLE STREET

CHILDREN'S UNIVERSITY HOSPITAL

Temple Street
CHILDREN'S UNIVERSITY HOSPITAL
140 YEARS
Caring for Sick Children

Report on the research carried out, publications and academic activities of the staff of Temple Street Children's University Hospital, 2012

Report on the research carried out, publications and academic activities
of the staff of Temple Street Children's University Hospital, 2012

mission STATEMENT

Excellence in research - making a difference to the lives of children

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Letter of Welcome from the Department of Research

Dear Colleague,

It gives us great pleasure to write the introduction to this our second Research Report. The first report, published in December 2011, collated all the research undertaken in Temple Street Children's University Hospital in the preceding 3 years. This publication concentrates on activity for the calendar year 2012.

The Department of Research opened its doors in Temple Street in July 2011. Since then it has very much become part of the Hospital structure, embracing all aspects of research within the institution. The Research Committee in tandem with the Hospital's Ethics Committee oversee the appropriateness of all research being undertaken on the campus.

Establishing a research function from scratch invariably entails interaction with and the cooperation of many departments and individuals on a medical, financial, legal and ethical level. We are most grateful to these disparate groups and are happy to report that most of the ground-work has now been covered and systems established.

As the Research Report, hereafter, will be an annual publication, we have decided to highlight each year a number of individuals who have contributed significantly at a national or international level to the Hospital's research profile. These are structured around the three research themes of the hospital – the Well Child, Rare Disorders in Children and Specialist Surgery. Thus over the next few years we hope to cover and demonstrate the wide scope of successful activity across many areas.

This year has also seen close cooperation between our Paediatric Clinical Leads as well as the National Children's Research Centre in Crumlin. We have a joint vision for the future, namely, to embed a research culture within the new children's hospital. We have a unique opportunity to build a centre of excellence for the care of our children, reflecting the best of international practice. The incorporation of a Research Institute at its core would ensure that research is both protected and developed and its fruits reaped in the clinical domain. This Report ends with an excerpt from a combined document prepared in conjunction with our colleagues in the NCRC which reflects our vision for the future from a research perspective.

Finally we are grateful to all our contributors and for the on-going support of our associated charity, the Children's Fund for Health Limited (the fundraising arm of Temple Street) who actively support research within our hospital.

Mr Alec Blayney
Head Of Research

Dr Gayle Kenney
Research Manager

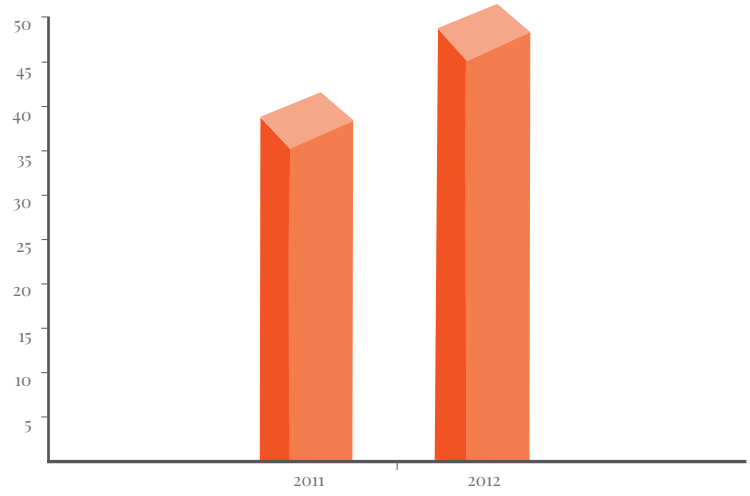
Introduction

The Research aim of the Hospital is to encourage and facilitate investigators to find new treatments and cures for childhood diseases, and to set new standards of paediatric care by improving clinical effectiveness, patient outcomes and children's healthcare experience.

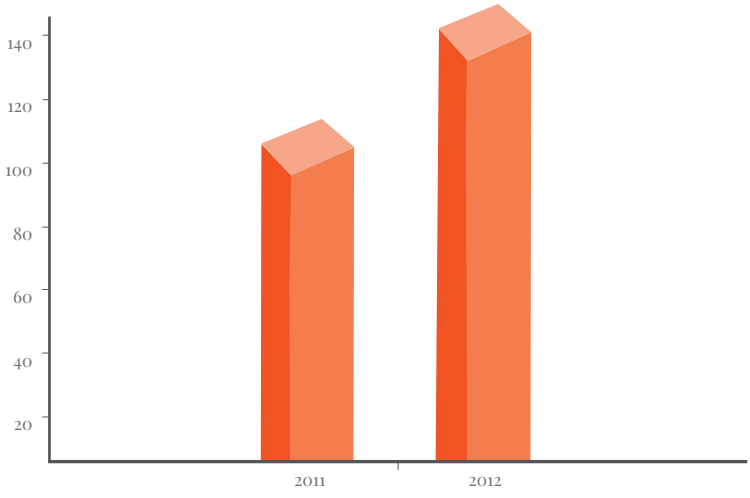
Research in 2012

Since the initiation of the Department of Research in 2011, research is increasingly viewed as essential for clinical excellence amongst front-line staff – we calculate that 56.5% of Temple Street consultants have been involved in a research project over the last 3 years (data compiled from TSCUH Research and Ethics Committees). We have seen an impressive increase in research proposals being submitted to, and approved by, the Research Committee and have also reported a large increase (from 2011 to 2012) in the number of publications authored by staff.

Proposals approved by TSCUH Research Committee



Publications Reported By TSCUH Staff



Research Themes

The Temple Street research strategy focuses on three broad themes that reflect the current and potential future strengths of the hospital, namely;

- The Well Child
- Rare Disorders in Children
- Specialist Surgery

In the following pages we highlight one or two areas of outstanding research under each of these three themes.

Overall

All the research captured in the rest of this report is organised into Departments. We have tried to capture as much information as we can in this document and, although there are some areas where information is incomplete, we trust that this Research Report provides an overview of the exceptional research carried out at Temple Street Children's University Hospital, Dublin in 2012.

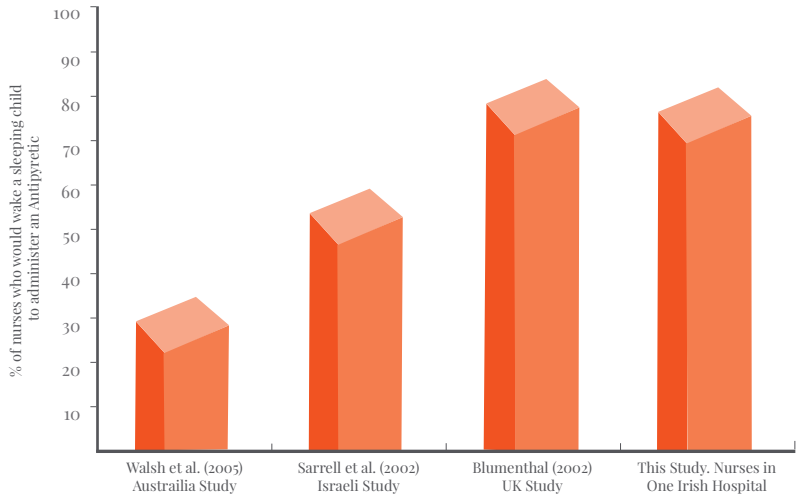
Research Highlights

The Well Child

Fever Management: Louise Greensmith

Fever has many immunological benefits; it inhibits bacterial growth and the replication of viruses. Many children tolerate mild to moderate fever with relative ease (Walsh et al. 2005). Despite this while working as a staff nurse here in Temple Street Children's University Hospital (TSCUH) I noted that rises in temperature were treated with antipyretic drugs such as paracetamol and ibuprofen regardless of the child's comfort. It seemed that nurses had a phobia of fever. Nurses administer antipyretics in an effort to reduce temperature and prevent febrile convulsions; however research has shown that the administration of antipyretic drugs are ineffective in preventing febrile convulsions (Carey 2010). There seemed to be a gap between the theory and practice of managing febrile children. This prompted me to research this area of nursing practice in further detail.

A descriptive quantitative study with the aim of describing nurses' knowledge of and attitudes towards fever and fever management in an Irish setting was undertaken (Greensmith 2013). All 400 staff nurses employed here in TSCUH were invited to complete anonymous questionnaires and 119 nurses responded. Fever is one of the most common symptoms managed by nurses. Despite this overall knowledge on the 20 knowledge items was low. Low knowledge scores were reported by international studies also. Findings revealed both appropriate and inappropriate attitudes amongst nurses to fever. Clinical guidelines do not support the administration of antipyretics to reduce fever alone or to prevent febrile convulsions. However 74% (n= 88) of nurses believe that sleeping children should be woken to receive antipyretic drugs. Again this is supported by international findings see table below.



Findings from this study highlighted the imperative need for nurses' to improve their knowledge of fever and to develop appropriate attitudes towards the management of fever.

In response to these findings the "pyrexia" nursing care plan was updated to reflect evidence based findings (NICE 2007). It now states that antipyretics should not be used with the sole aim of treating fever but should be administered as an analgesic agent if a febrile child becomes uncomfortable. Printed discharge advice given to parents or carers of febrile children was also updated to reflect evidence based findings. Findings from the study were also discussed with lecturers from the University which along with TSCUH trains children's nurses.

Other studies found peer education programmes successful in improving nurses' knowledge of and attitudes towards fever. Two focus groups were conducted with staff nurses to determine what they thought should be included in the education intervention and to find out how they thought this programme should be delivered. Findings from this focus group were very interesting. Nurses were aware of the immunological benefits of fever; however they felt that not treating fever "goes against their grain." They felt parental pressure to treat febrile children and felt that doctors wanted their

febrile patients to be treated with antipyretic drugs. All nurses involved in the focus groups agreed that education on the management of fever was required. Nurses believed that doctor's knowledge of and attitudes towards fever should also be examined. Nurses believed that multidisciplinary ward sessions would be most appropriate. In response to this I am currently working with my multidisciplinary colleagues to develop an educational intervention with the aim of improving TSCUH's staff knowledge of and attitudes towards fever management. I also am planning a second descriptive study of doctor's knowledge of and attitudes towards fever and fever management.

Dissemination of Findings

The full study is available for staff to access in the TSCUH library. Findings from the original study were presented to staff at the 2012 Nursing Research Conference and the study won the award for best presentation on that day. I travelled to Cambridge to present the findings at the 23rd International Networking for Healthcare Education Conference in 2012. I also presented the findings at the 13th Interdisciplinary Research Conference in Trinity College Dublin. I have been accepted to present the findings of the staff nurse focus groups at the 14th Interdisciplinary Conference in Trinity this year.

References

Carey J.V. (2010) Literature Review: should antipyretic therapies routinely be administered to patient fever? Journal of Clinical Nursing 19, 2377-2393.

Greensmith L. (2013) Nurses' knowledge of and attitudes toward fever and fever management in one Irish Children's Hospital. Journal of Child Health Care. Retrieved from http://chc.sagepub.com/content/early/2013/01/23/1367493512461457.full.pdf+html, DOI: 10.1177/1367493512461457.

National Institute for Health and Clinical Excellence (2007) Feverish illness in children; Assessment and initial management in children younger than 5 years. Retrieved from http://guidance.nice.org.uk/nicemedia/live/11010/30523/30523.pdf
Walsh A., Edwards H., Courtney M., Monaghan S. and Wilson, J. (2005) Fever management: paediatric nurses' knowledge, attitudes and influencing factors. Journal of Advanced Nursing 49(5), 453-464.

National Institute for Health and Clinical Excellence (2007) Feverish illness in children; Assessment and initial management in children younger than 5 years. Retrieved from http://guidance.nice.org.uk/nicemedia/live/11010/30523/30523.pdf
Walsh A., Edwards H., Courtney M., Monaghan S. and Wilson, J. (2005) Fever management: paediatric nurses' knowledge, attitudes and influencing factors. Journal of Advanced Nursing 49(5), 453-464.

Dr Veronica Lambert, Dublin City University, presenting the award for best presentation to Ms Louise Greensmith at the TSCUH 2012 Nursing Research Conference

Rare Disorders in Children

Professor Eileen Treacy and Ms Karen Coss with the Neil Buist Award

National Centre for Inheriteded Metabolic Disorders

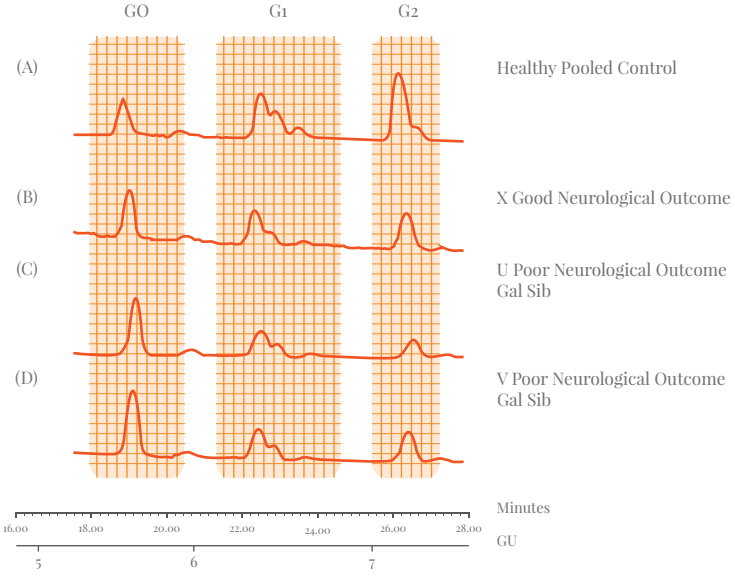
Karen Coss, a Translational Medicine PhD student at the UCD, Clinical Research Centre (CRC) and Childrens University Hospital was recognised with a prestigious research award (Neil Buist Award) by the US Society for Inherited Metabolic Disorders last March 2012. Her research was carried out with Prof Eileen Treacy, Clinical Professor of Inborn Errors at TCD, Metabolic Consultant at the National Centre for Inherited Metabolic Disorders, Childrens University and Mater Misericordiae University Hospital as the Principal Investigator and Co-Investigators, Dr Peter Doran, Scientific Director of the UCD CRC and Prof Pauline Rudd, Head of the Dublin-Oxford Glycobiology Laboratory, NIBRT.

Her research is focused on examining the biological origins of the poorly understood long-term complications associated with Classical Galactosaemia, a rare metabolic disorder of carbohydrate metabolism. This condition affects 1:16,476 births annually in Ireland (the highest incidence worldwide) (Coss et al., 2013) and is screened for by the National Newborn Screening Programme (NNSP). The only treatment is life-long dietary restriction of galactose. Despite early treatment the majority of affected patients go on to develop long-term complications such as intellectual impairment, neurological complications, speech difficulties and infertility in females.

The research group has shown in detailed biochemical and gene analysis studies that major abnormalities persist in treated individuals. These findings affect the production and function of complex molecules in the body, particularly glycoproteins which consist of sugar chains attached to proteins. Structural changes in these glycoproteins eventually lead to disturbances of the body's intrinsic cellular machinery. This helps to explain clinical findings such as delayed brain maturation, cognitive impairment and female infertility.

Over-restriction of dietary galactose in the long-term may exasperate the condition by starving the body's pathways of essential galactose required for correct production of these sugar chains.

In a presentation by Karen to the US Society for Inherited Metabolic Disorders, 'Classical Galactosaemia- a modifiable Glycosylation disorder?',(Coss KP, Doran PP, Byrne JC, Adamczyk B, Rudd PM and Treacy EP), Karen illustrated that Galactosaemia patients had abnormal sugar chain profiles and illustrated modification/improvements of these abnormalities with differing galactose increments (Coss et al, 2012,). The group have also shown dysregulation of numerous genes involved in sugar chain production in treated patients.



IgG N-glycan defects in adults. Coss *et. al.* 2012 MGM

This novel, innovative research suggests that long-term over-restriction of galactose in parallel with ongoing gene dysregulation has a significant role to play in long-term complications in Galactosaemia. This has provided new insights into the nature of Galactosaemia with detection of glycan and gene biomarkers with the potential to advance the field of research in the future and improve future treatment and patient outcomes.

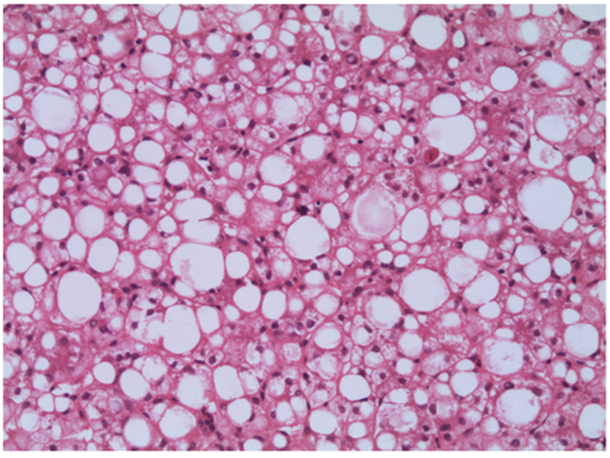
Medical Genetics

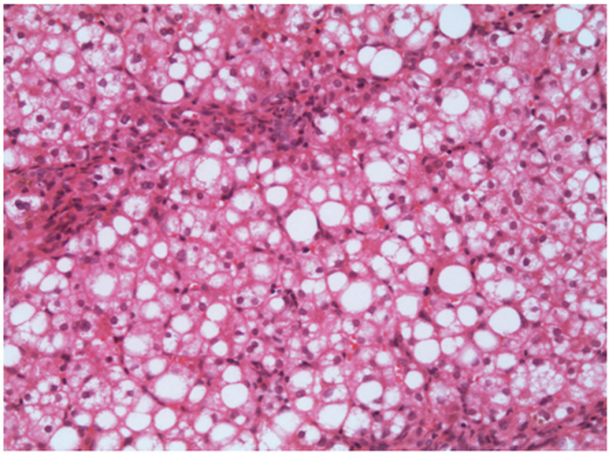
Dr Jillian Casey, an MRCG-funded Post-Doctoral researcher at the National Children's Research Centre, received the 'Best Platform Presentation Award' at the British Inherited Metabolic Disease Group Annual Meeting July 5th 2013 (London). http://www.bimdg.org.uk/BIMDG2013.asp

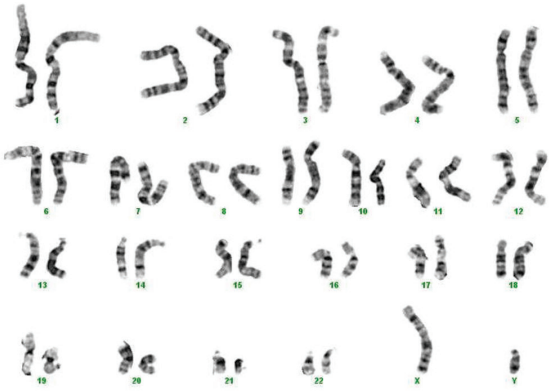
Dr Jillian Casey

The presentation showcased the research teams' recent identification of an alteration in the LARS gene that causes a rare form of liver failure affecting children, many of whom survive into adulthood. This is the first time that the LARS gene – an enzyme responsible for incorporating the essential amino acid leucine when making proteins - has been implicated in a human disorder. In addition to liver dysfunction, the children present with developmental delay, seizures and anaemia and, in some cases, the disorder can be life-threatening.

Despite the fact that there is no treatment for the children with this rare liver disorder, knowledge of the genetic mechanism has directly altered patient management with affected individuals now being given protein (that includes leucine) orally or by transfusion when ill, in the hope that this will improve outcomes. The clinical and research teams are monitoring the possible beneficial effects of this treatment strategy. The team also translated their research directly to a clinical setting through the development of a non-invasive diagnostic test for this disorder, which avoids the need to undergo painful liver and muscle biopsies and which has resulted in new diagnoses for additional patients.







Biopsy (above) & Karyotype

Further work investigating the underlying disease mechanism associated with LARS as a first step towards the development of a novel therapeutic strategy is ongoing.

The research study was co-led by Drs Jillian Casey, SallyAnn Lynch and Ellen Crushell (Our Lady's Children's Hospital Crumlin and Temple Street Children's University Hospital) and co-funded by the Children's Fund for Health Temple Street Children's University Hospital and a Medical Research Charities Group grant from the National Children's Research Centre and Health Research Board.

Specialist Surgery

Anaesthesia

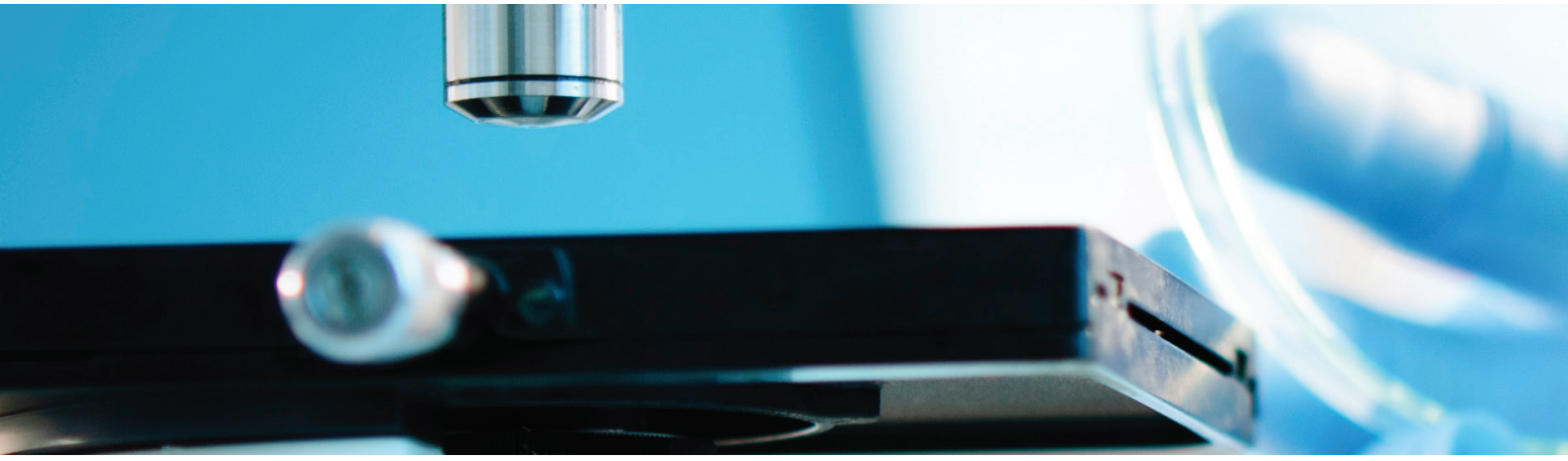
Dr Nandini Kandamany was awarded the "Winner of Best Presentation Award" at the Irish Paediatric Anaesthesia and Critical Care Society Annual Scientific Meeting in April 2012 for the abstract below.

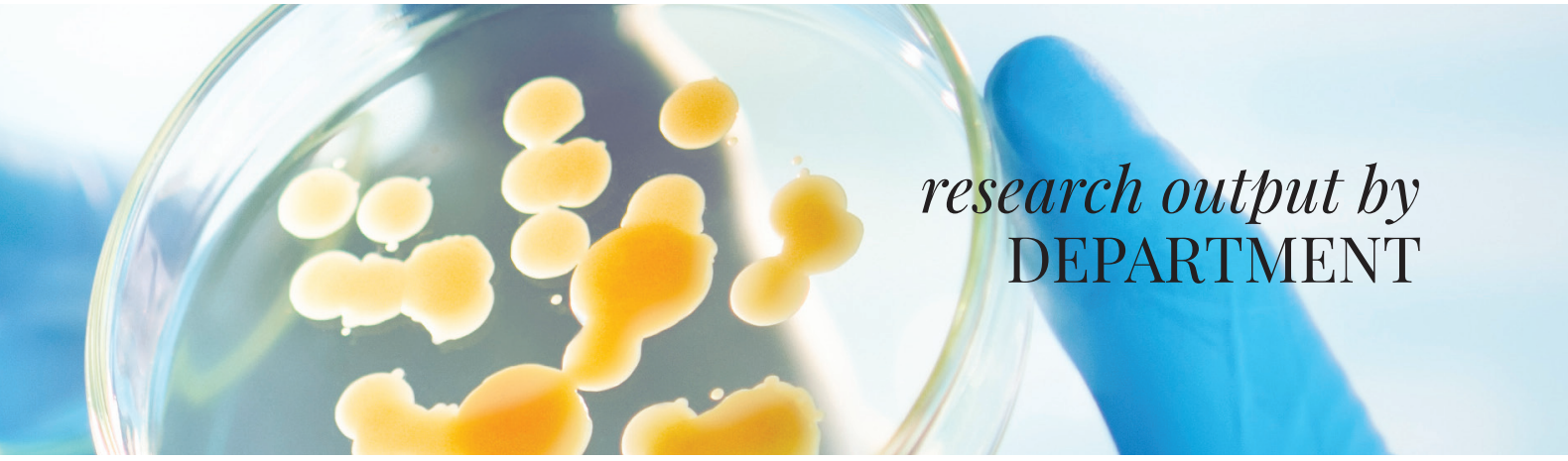
Treating bronchiolitis: could less be more?

Kandamany N, Sattar F, Conrick-Martin I, Carson K; Department of Paediatric Intensive Care, Children's University Hospital, Temple Street, Dublin 1.

Objectives: To audit the management of bronchiolitic infants admitted into the Paediatric Intensive Care Unit (PICU) with an aim to assessing the benefits of early extubation compared to conventional treatment.
Methods: All intubated patients admitted into ICU with a diagnosis of bronchiolitis over a 3 month period were included. Patient data was obtained retrospectively and incorporated time to extubation, use of non-invasive continuous positive airway pressure (nCPAP), duration of opioid, benzodiazepine and diuretic use, need for re-intubation, time to feeding and time to discharge from PICU and whether physiotherapy was performed amongst other parameters. Patients extubated within 24 hours of admission were analysed as a cohort group and data was compared to patients who were extubated more than 24 after admission.
Results: 27 patients were included in this audit. Patients who were extubated early showed improved outcomes in relation to need for re-intubation and time to discharge from PICU. Use of nCPAP, diuretics and physiotherapy were also positively associated with patient outcomes.
Conclusions: Our data would suggest that early extubation with diuretic use, physiotherapy, light sedation and early feeding in bronchiolitis in collaboration positively affect PICU patient management.

Dr Kevin Carson and Dr Nandini Kandamany





research output by DEPARTMENT

Anaesthesia

Presentations at National Conferences

Kandamany N, Sattar F and Carson K. **Managing Bronchiolitis: Could Less Be More.** Oral presentation at IPACS Meeting; 2012.

Sattar F, Carson K and Doherty D. **Nava – A true assertion of brain over medicine – A case report of successful use of NAVA in a 5 years old child with acute severe asthma.** Oral presentation at IPACS Meeting; 2012.

Sattar F, Kandamany N, Gendi S, O' Dwyer and Carson K. **A rare case of spontaneous esophageal perforation in a seven months old infant.** Oral presentation at IPACS Meeting; 2012.

Carson K. **Organ Donation: A Paediatric Perspective.** Invited lecture at ICSI Organ Donation. Autumn Meeting November 2012.

Local Presentations

Ward W, Kirby F. **Data from The Children's University Hospital, as part of the National Anaesthetic Survey in Ireland, conducted from 26th November to December 3rd.** Oral Presentation at the Audit / Research Day December 2012 at the Children's University Hospital, Dublin.

Awards / Prizes

Kandamany N. Awarded the prize for oral presentation at the IPACS Meeting; 2012.

Audiology

Initiated / Ongoing Research Projects

The establishment of normative data for Central Auditory Processing Disorders tests in Ireland.
Central Auditory Processing Disorder (CAPD) results in children not understanding the sounds that they hear, despite normal hearing. Diagnostic tests for this condition were developed in the United States. The purpose of our study is to establish norms for these CAPD tests on typically developing Irish children, which is an essential step towards the development of services to assess, diagnose and treat people with CAPD in Ireland.
Invesitgators: Pauline Ackermann (PI), Martin Cromb, Louise Keogh,Gillian McAlister, Clare Salley
Funded by the Children's Fund for Health RPAC-11-48

Central Decontamination Unit

Initiated / Ongoing Research Projects

Decontamination of Infant Formula Bottles and Accessories in the Hospital Setting- Can We Improve Our Practices?
In the healthcare setting appropriate decontamination of infant formula bottles, accessories and food preparation equipment used during treatment of illness, or episodes of care, is essential to minimise the risk of cross contamination of infant formula, formula bottles and equipment (Drudy et al., 2006; Faber & Forsythe 2008; Healy et al., 2010).
Investigator: Caroline Conneely

Validation of an endoscope drying cabinet for extended storage of non-channelled endoscopes - a clinical perspective.
Early data indicates that a change in practice will reduced endoscope reprocessing activity significantly, minimise the exposure of endoscopes to harsh chemicals and improve clinician access to essential equipment.
Investigator: Caroline Conneely

Clinical Biochemistry

Peer Reviewed Publications

Crushell E, O'Leary D, Irvine A, O'Shea A, Mayne P, Reardon W. **Methylenetetrahydrofolate reductase (MTHFR) deficiency presenting as a rash.** *Am J Med Genet* 2012; 158:2254-7.

Published Abstracts

Mayne PD, Roche G, Greenwood C, Cooney D, Rogers M, Roring S, McDevitt T, Barton DE. **Changing incidence of Cystic Fibrosis in Ireland?** Proceedings of the 35th European Cystic Fibrosis Association, 2012.

Presentations at International Conferences

Roche G, Greenwood C, Cooney D, Borovickova I, Rogers M, Barton DE, Mayne PD. **Cystic Fibrosis Newborn Screening in Ireland -The First Year experience.** International Society of Neonatal Screening (ISNS). 2012 Budapest, Hungary.

Presentations at National Conferences

Mayne PD, Brady J, Cooney D, Roche G. **Impact of CF newborn bloodspot screening on sweat testing.** Association of Clinical Biochemists of Ireland 2012, Dublin, Ireland.

Deverell D, Trench, C, Hughes, J, Mayne PD. **Investigation of Developmental Delay Reveals a Urea Cycle Defect in Two Siblings.** Association of Clinical Biochemists of Ireland 2012, Dublin, Ireland.

Borovickova I, Fitzsimons PE, Knerr I, Mayne PD. **Rhabdomyolysis in a young individual: When to think of inborn errors of metabolism.** Association of Clinical Biochemists of Ireland 2012, Dublin, Ireland.

Local Presentations

Scully D, Nicholson A, Mayne P. **Review of sodium levels of less that 125 mmol / L over a six month period in the Children's University Hospital, Temple Street.** Oral Presentation at the Research / Audit Day December 2012 at the Children's University Hospital, Dublin.

Borovickova I. **Measuring PAP in dried blood spots as another tier in newborn CF screening.** Oral Presentation at the Research / Audit Day December 2012 at the Children's University Hospital, Dublin.

Dermatology

Peer Reviewed Publications

Lynch M, O'Loughlin A, Devaney D, O'Donnell B. **BCGitis in a Patient with Transient Hypogammaglobulinemia of Infancy.** *Pediatr Dermatol*. 2012 Dec 18. doi: 10.1111 / pde.12036. [Epub ahead of print]

Published Abstracts

Lynch M, Lenane P, O'Donnell B. **Propranolol for the treatment of infantile haemangiomas: our experience with 33 patients (PA27).** In: (British Journal of Dermatology) 2012: 167 (Suppl. 1): 140.

Presentations at International Conferences

Lynch M, Lenane P, O'Donnell B. **Propranolol for the treatment of infantile haemangiomas – our experience with 33 patients.** Poster presentation at the British Association of Dermatologists Meeting (British Society for Paediatric

Lynch M, McAleer MA, Irvine AD, Watson R, Hughes R, O'Donnell B. **Three cases of RASA1-related capillary malformation-arteriovenous malformation syndrome.** *Br J Dermatol* 2012; 167: e35.

Cunningham, O'Loughlin AM, O'Donnell B. **A partially blinded, nonrandomized, controlled, 'half and half' study to compare the clinical and antimicrobial efficacy of topical steroid alone vs. an equivalent topical steroid combined with antibiotic, in the treatment of atopic dermatitis in a paediatric dermatology outpatient population, within a tertiary referral centre.** *Br J Dermatol* 2012: 166: e1.

Lynch M, Lenane P, O'Donnell B. **Propranolol for infantile haemangioms: our experience with 33 patients.** *Br J Dermatol* 2012: 167:e30.

Presentations at International Conferences

Lynch M, Lenane P, O'Donnell B. **Propranolol for the treatment of infantile haemangiomas – our experience with 33 patients.** Poster presentation at the British Association of Dermatologists Meeting (British Society for Paediatric Dermatology), July 2012: Birmingham, UK.

Presentations at National Conferences

Lynch M, McAleer MA, Irvine AD, Watson R, Hughes R, O'Donnell B. **Three cases of RASA1-related capillary malformation-arteriovenous malformation syndrome.** Oral presentation at the Irish Association of Dermatologists, Spring Meeting; April 2012: Fitzpatrick Castle Hotel, Dublin.

Lynch M, Lenane P, O'Donnell B. **Propranolol for infantile haemangiomas: our experience with 33 patients.** Oral presentation at the Irish Association of Dermatologists, Spring Meeting; April 2012: Fitzpatrick Castle Hotel, Dublin.

Matthews K, Murphy A, Watson R, Irvine A, O'Donnell B. **CLOVES Syndrome:** Oral presentation at the Royal Academy of Medicine in Ireland (Dermatology Section); OLHSC, Crumlin; Dec 2012: Dublin.

Murad A, O'Donnell B. **An unusual case of generalised oedema.** Oral presentation at the Royal Academy of Medicine in Ireland (Dermatology Section); OLHSC, Crumlin; Dec 2012: Dublin.

Emergency Medicine

Initiated / Ongoing Research Projects

Suspected abusive head trauma
Increase of knowledge and awareness. Dr. Ikechukwu Okafor.

Peer Reviewed Publications

Capra L, Levin AV, Howard A, Shouldice M. **Characteristics of femur fractures in ambulatory young children.** *(Emerg Med J).* 2012 October 4.

Local Presentations

Robert Marsh. **Audit to monitor the number of category 3 and 4 self referrals to CUH Temple Street Emergency Department.** Oral presentation at the Audit / Research Day June 2012 at the Children's University Hospital, Dublin.

Sheena Durnin. **Audit of Pre-Hospital Pain Management in Injuries.** Oral Presentation at the Audit / Research Day June 2012 at the Children's Universtiy Hospital, Dublin

Moore CM, Doyle J, Nicholson AJ. **An audit of accidental ingestion presentations to CUH 2010-2011.** Oral presentation at the Research / Audit Day December 2012 at the Children's University Hospital, Dublin.

Freyne B, Doyle J, Mc Namara R, Nicholson AJ. **Epidemiology of high falls from windows in Irish Children.** Oral presentation at the Research / Audit Day December 2012 at the Children's University Hospital, Dublin.

Awards / Prizes

Eva Forman, Mary Mc Kay, Sabine Maguire, AJ Nicholson. **Patterns of burns and scald injuries in young children - are Irish children any different?** Second Prize Awarded for oral presentation at the Audit / Research Day June 2012 at the Children's University Hospital, Dublin.

Endocrinology

Peer Reviewed Publications

Flanagan SE, Xie W, Caswell R, Damhuis A, Vianey-Saban C, Akcay T, Darendeliler F, Bas F, Guven A, Siklar Z, Ocal G, Berberoglu M, Murphy N, O'Sullivan M, Green A, Clayton PE, Banerjee I, Clayton PT, Hussain K, Weedon MN, Ellard S. **Next generation sequencing reveals deep intronic crypticABCC8 and HADH spicing founder mutations causing hyperinsulinism by psuedoexon activation.** *Am J Hum Genet*. 2013 Jan 10;92(1):131-6. Epub 2012 Dec 27.

McCarthy RA, McKenna MJ, Oyefeso O, Uduma O, Murray BF, Brady JJ, Kilbane MT, Murphy JF, Twomey A, Donnell CP, Murphy NP, Molloy EJ. **Vitamin D nutritional status in preterm infantsand response to supplementation.** *Br J Nutr*. 2012 Nov 27:1-8.

Onwuneme C, Carroll A, McCarthy R, Kilbane M, McKenna M, Murphy N, Molloy EJ. **Towards evidence based medicine for Paediatricians Question 2: What is the ideal dose of vitamin d supplementation for term neonates?** *Arch Dis Child*. 2012 Apr;97(4):387-9. doi: 10.1136/archdischild-2011-301764. Review.

O'Grady MJ, McGrath N, Quinn FM, Capra ML, McDermott MB, Murphy NP. **Spermatogenesis in a prepubertal boy.** *J Pediatr*. 2012 Aug;161(2):369-369.e1. doi: 10.1016/j.jpeds.2012.03.040. Epub 2012.

Presentations at International Conferences

Neville S, Hawkes CP, Allen NM, Moloney S, Laffan E, Murphy NP. **Characteristics of children with multiple pituitary hormone deficiency and isolated growth hormone deficiency presenting to an Irish tertiary hospital.** Poster presentation. Pediatric Academic Societies' Annual Meeting, May 4-7, in Washington, DC.

GM Murray, S Neville, N O'Brien, CP Hawkes, NP Murphy. **A neonate with severe failure to thrive, hyponatraemia and hyperkalaemia with a normal 17-hydroxyprogesterone.** Poster Presentation Irish American Paediatric Society, Belfast Sept 2012.

Niamh O'Brien, Grainne M Murray, Colin P. Hawkes, Nuala P. Murphy. **Are the NHS standards of care for children with type 1 diabetes practical in an Irish setting?** Oral Presentation; Irish American Paediatric Society, Belfast Sept 2012.

Local Presentations

R. Dhar, K. Nugent, C.M McDonnell, N.P Murphy. **Autoantibodies in type 1 diabetes: are we different?** Oral Presentation at the Audit / Research Day June 2012 at the Children's University Hospital, Dublin.

O'Brien N, Hawkes CP, Murphy NP. **Are the NHS standards of care for children with type 1 diabetes practical in an Irish setting?** Oral Presentation at the Audit / Research Day December 2012 at the Children's University Hospital, Dublin.

Presentations at National Conferences

Lynch M, McAleer MA, Irvine AD, Watson R, Hughes R, O'Donnell B. **Three cases of RASA1-related capillary malformation-arteriovenous malformation syndrome.** Oral presentation at the Irish Association of Dermatologists, Spring Meeting; April 2012: Fitzpatrick Castle Hotel, Dublin.

Lynch M, Lenane P, O'Donnell B. **Propranolol for infantile haemangiomas: our experience with 33 patients.** Oral presentation at the Irish Association of Dermatologists, Spring Meeting; April 2012: Fitzpatrick Castle Hotel, Dublin.

Matthews K, Murphy A, Watson R, Irvine A, O'Donnell B. **CLOVES Syndrome:** Oral presentation at the Royal Academy of Medicine in Ireland (Dermatology Section); OLHSC, Crumlin; Dec 2012: Dublin.

Murad A, O'Donnell B. **An unusual case of generalised oedema.** Oral presentation at the Royal Academy of Medicine in Ireland (Dermatology Section); OLHSC, Crumlin; Dec 2012: Dublin.

Ear, Nose and Throat

Peer Reviewed Publications

Lennon P, Khoo S G, Colreavy M P. **Complexities of revision mastoid surgery in a migratory population cohort.** *Ir Med J*. 2012 Mar;105(3):78-80.

Walsh R A, Gillivan-Murphy P, Murphy C, Colreavy M P, O'Rourke K, Lynch T. **Bilateral cerebellar stroke presenting with acute dysphonia and late palatal tremor.** *Mov Disord*. 2012 Mar; 27(3):346 – 8.

O'Duffy F, O'Dwyer T P. **The growing epidemic of HPV associated oropharyngeal malignancy.** *Ir Med J*. 2012 Apr;105(4): 101-102.

Royds J, Kieran S, Timon C. **Oxidised cellulose (Surgical) based reaction post thyroidectomy mimicking an abscess: A case report.** *Int J Surg Case Rep.* 2012 Apr 16; 3(7):338-9. Epub 2012 Apr 16.

Glynn, R. W., Colreavy, M., Rowley, H. & Gendy, S. **Division of tongue tie: review of practice through a tertiary paediatric otorhinolaryngology service.** *Int. J. Pediatr. Otorhinolaryngol*. 76, 1434–1436 (2012).

Books / Chapters

Kieran SM, Russell JD. **Chapter 24**. Choanal Atresia. In Newborn Surgery, 3rd Edition.

Kieran SM, Russell JD. **Chapter 38.** Bronchoscopy in the Newborn. In Newborn Surgery, 3rd Edition.

Presentations at International Conferences

SM Kieran, Kirby A, Oyemwense N, Roberson D. **Risk Factors for Desaturation after Tonsillectomy: Analysis of 4095 Consecutive Cases.** Triological Society meeting, Spring Meeting at COSM. April 2012: San Diego.

SM Kieran, K Watters, R Rahbar. **The lipid laden macrophage index in tracheal aspirates – Correlation with Alveolar Lavage.** American Society of Pediatric Otolaryngology Meeting. April 2012: San Diego.

Glynn, R. W., Colreavy, M., Rowley, H. & Gendy, S. **Division of tongue tie: Audit of practice through a paediatric otolaryngology service.** Poster Presentation by Mr. Ronan Glynn in 11th International Congress of the Europen Society of Paediatric Otorhinolaryngology, Amsterdam. May 2012.

Glynn RW, O'Duffy F, O'Dwyer T, Colreavy M, Rowley H. **Patterns of Internet Use by Parents of Children Attending a Paediatric Otolaryncology Service.** Oral Presentation by Mr. Ronan Glynn in 11th International Congress of the Europen Society of Paediatric Otorhinolaryngology, Amsterdam. May 2012.

National Presentations

Davies K, O'Dwyer T **Botox - A wrinkle free approach to managing sialorrhoea**
Irish Otolaryngology Society Sept 2012

Davies K, Rowley H **Management guidelines for cutaneous haemangioma of the head and neck**
Irish Otolaryngology Society Sept 2012

Local Presentations

Glynn RW, O'Duffy F, O'Dwyer T, Colreavy M, Rowley H. **Patterns of Internet Use by Parents of Children Attending a Paediatric Otolaryncology Service.** Oral Presentation at the Audit / Research Day June 2012 at the Children's University Hospital, Dublin.

General Surgery

Initiated / Ongoing Research Projects

The transition zone in Hirschsprungs disease.
This project will hopefully improve post-operative clinical outcome in children with Hirschsprungs disease and decrease long term morbidity.
Investigators: J.Gillick, D. Coyle, AM. O'Donnell, P. Puri

The origin of congenital foregut abnormalities in OA / TOF
Study to investigate the origins of congenital birth defects, specifically Oesophageal Atresia / Tracheo-Oesophageal Fistula.
Oesophageal atresia is an important clinical problem however the origins of this condition are not known. A better understanding of this condition in the context of its development could improve the potential for new therapies and diagnostic tests.
Investigators: Dr. Danielle McLaughlin, Prof Prem Puri (NCRC), Mr Alec Blayney, Dr Paula Murphy (TCD)
Co-funded from the Children's Fund for Health (PAC 11-57) and the National Children's Research Centre

Peer Reviewed Publications

Stephens LR, Donoghue V, Gillick J. **Radiological versus clinical evidence of malrotation, a tortuous tale-10-year review.** *Eur J Pediatr Surg.* 2012 Jun; 22(3):238-42.

McLaughlin D, Hajduk P, Murphy P, Puri P. **Adriamycin-Induced Models of VACTERL Association.** *Mol Synd. Mol Syndromol.* 2013 Feb;4(1-2):46-62

Presentations at International Conferences

McMahon S, Abdelhafeez A, McDowell DT, Gillick J. **Time taken to establish feeds after surgery for bowel atresia-when to worry?** Oral presentation at The Pacific Association of Pediatric Surgeons Annual meeting; 2012: Shanghai, China.

McDowell DT, Quinn F, Mortell A. **PSARP - Paediatric Surgery: Advice Regarding Publishing.** Oral Presentaion by Dr. Dermot McDowell at the EUPSA-BAPS Joint Congress in Italy.

Local Presentations

McDowell DT, Quinn F, Mortell. **PSARP - Paediatric Surgery: Advice Regarding Publishing.** Oral Presentaion at the Audit / Research Day June 2012 at the Children's University Hospital, Dublin.

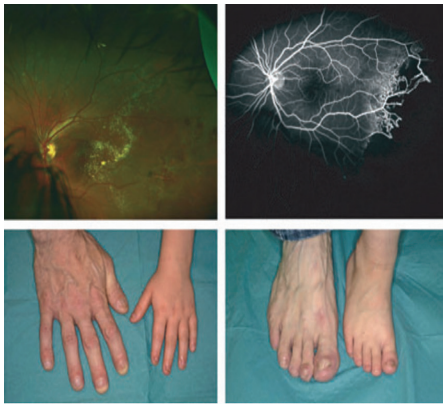
Nomsa Maphango, Dermot Thomas McDowell, Madhavi Kakade, Sami Awadalla. **Lily white and saved from the surgeon's knife: An audit of negative appendectomies in a tertiary referral paediatric unit.** Oral Presentaion at the Audit / Research Day June 2012 at the Children's University Hospital, Dublin.

Nochiparambil Mohanan, Sami Awadalla, John Gillick and Alan Mortell. **A comparative study of laparoscopy-assisted pull-through versus Soave and Trans anal pull through for Hirschsprung's disease.** Oral Presentaion at the Audit / Research Day June 2012 at the Children's University Hospital, Dublin.

Haematology

Peer Reviewed Publications

Gleeson M, O'Marcaigh A, Cotter M, Brosnahan D, Vulliamy T, Smith OP. **Retinal vasculopathy in autosomal dominant dyskeratosis congenita due to TINF2 mutation.** *Br J Haematol.* 2012 Dec; 159(5):498. doi: 10.1111 / bjh.12088. Epub 2012 Oct 24. PubMed PMID: 23094712



Quinlan C, Bates M, Cotter M, Riordan M, Waldron M, Awan A. **Tinzaparin is safe and effective in the management of hemodialysis catheter thrombosis.** *ASAIO J.* 2012 May-Jun; 58(3):288-90. PubMed PMID: 22456106.

History / Archives

Initiated / Ongoing Research Projects

Temple Street – Portrait of a Children's Hospital

The author is contracted to New Island to produce a new book about the history of Temple Street (publication date; Autumn 2014. It is aimed at a general audience.
In its UK-wide evaluation of the role of the History of Medicine has to play in contemporary healthcare, the Wellcome Trust (April 2000) noted that the discipline has a definite impact on:

- Promotion of funding activities outside the immediate discipline
- Increased public appreciation of the value of work undertaken by a hospital / other healthcare institution
- The direction taken by public health policy

Investigator / **Author:** Barry Kennerk

Peer Reviewed Publications

Kennerk B. **In Danger and Distress: Presentation of Gunshot Cases to Dublin Hospitals during the Height of Fenianism, 1866-1871.** *Social History of Medicine.* 2012 Jan 24: 3: 588-608.

Presentations at National Conferences

Kennerk B. **War on our Doorstep: Temple Street Hospital and its response to the 1916 Rising.** Oral presentation at Medicine, Health and Irish Experiences of War Conference (1914-45); 2012: University College Dublin (Chapter by same author in forthcoming UCD publication based on these proceedings).

Infectious Diseases and Immunology

Initiated / Ongoing Research Projects

Analysis of the anti-viral IFN-a JAK / STAT pathway in Hepatitis C (HCV) infected children. This project will determine the status of the JAK / STAT pathway in children with HCV and attempt to identify markers of therapeutic outcome for paediatric HCV patients
Investigators: Dr. Patrick Gavin, Dr. Julie Lucey and Dr Nigel Stevenson (TCD).
Funded by the Children's Fund for Health. RPAC-13-02

Peer Reviewed Publications

Rao U, Piccin A, Malone A, O'Hanlon K, Breatnach F, O'Meara A, McDermott M, Butler K, O'Sullivan N, Russell J, O'Marcaigh A, Smith OP. **Utility of bronchoalveolar lavage in the diagnosis of pulmonary infection in children with haematological malignancies.** *Ir J Med Sci.* 2012 Sep 16. [Epub ahead of print]

Drew RJ, Ó Maoldomhnaigh C, Gavin PJ, O' Sullivan N, Butler KM, Cafferkey M. **The impact of meningococcal polymerase chain reaction testing on laboratory confirmation of invasive meningococcal disease.** *Pediatr Infect Dis J.* 2012 Mar;31(3):316-8.

Bolton S, Keane E, Butler K, Gavin P. **Prospective national surveillance for Kawasaki disease in the Island of Ireland, 2008-2009.** *Pediatr Intern.* 2012; 54 (Suppl. 1): 68.

Drew RJ, O'Maoldomhnaigh C, Gavin PJ, Cunney R, Sullivan N, Butler KM, Cafferkey M. **The impact of meningococcal Polymerase Chain Reaction testing on laboratory confirmation of invasive meningococcal disease.** *Pediatr Infect Dis J.* 2012 Mar; 31(3):316-318.

Lucey JM, Gavin P, Cafferkey M, Butler KM. **Pneumococcal meningitis: clinical outcomes in a pre-vaccine era at a Dublin paediatric hospital, 1999-2007.** *Ir J Med Sci.* 2011 Mar;180(1):47-50. Epub 2010 Nov 12.

Gavin PJ. **Introduction of cognitive apprenticeships (sub-internships,trainee-intern year or student assistantships) and shadowing to undergraduate final-year curricula of Irish medical schools could significantly improve students' preparedness for internship and reduce stress of transition.** M.Sc Medical Education. Cardiff University. Jan 2012.

Published Abstracts

Goode M, Lynam P, Leahy R, Gavin P, Butler KM. **Immunogenicity and tolerability of AS03 adjuvanted H1N1 vaccine in HIV infected children.** Abstract. ESPID. 30th Annual meeting. Thessaloniki, Greece. May, 2012.

Leahy TR, Bacon L, Gavin PJ, Butler KM, O' Marcaigh A. **A two-dose regimen of the 2009 monovalent H1N1 influenza A vaccine increases seroconversion among children with acute lymphoblastic leukaemia.** Abstract 435. ESPID. 30th Annual meeting. Thessaloniki, Greece. May, 2012.

Cummins P, McCarthy B, R Cunney, A Oza, P Gavin. **A point prevalence study of antimicrobial use in two Irish tertiary paediatric hospitals. Abstract.** Irish Paediatric Society Annual Meeting. Belfast. April 2012.

Dhar R, Coughlin S, Cunney R, Cleary J, Reilly C, Gavin PJ. **2011 Measles Epidemic: The Children's University Hospital Temple Street story.** Abstract . Irish Paediatric Society Annual Meeting. Belfast. April 2012.

Purcel Cl, Dominguez M, Butler KM, Twomey E, Gavin PJ. **Caffey's disease: mimic of infection and trauma.** Abstract. Irish Paediatric Society Annual Meeting. Belfast. April 2012.

Presentations at International Conferences

Butler K and Starke J. **Case studies in pediatric infectious diseases.** Oral presentation at ICAAC (Interscioence conference on antimicrobial agents and chemotherapy); Sept 2012: San Francisco.

Butler K. Panelist. **Interactive case studies in paediatric infectious diseases.** Oral presentation at ESPID (European Society for Paediatric Infectious Diseases); May 2012: Thessaloniki, Greece.

Butler K. **Mother to child transmission of HIV infection – toward zero transmission.** Oral presentation at The Graham & Jenny Watson Study Day- Childhood Infection and Immunity, The Great North Children's Hospital, Newcastle- upon-Tyne. UK.

Local Presentations

Ibrahim L, Domingue M, Scanlan B, Sasaki E, Carter M, Rush R, O'Sullivan N, Gavin PG, Butler K. **Two year audit of Gram Negative Bacillus Bacteremia in Our Lady's Childrens Hospital Crumlin.** Oral presentation at the OLCHC Study Day May 2012.

R. Dhar, S. Coughlin, R. Cunney, J. Cleary, C. Reilly, P,J Gavin. **2011 Measles epidemic: the Children's University Hospital, Temple Street story.** Oral presentation at the Audit / Research Day June 2012 at the Children's University Hospital, Dublin.

Freyne B, Gavin P. **Routine Monitoring of children with Chronic Hepatitis C; yield and outcomes.** Oral presentation at the Research / Audit Day December 2012 at the Children's Universtiy Hospital, Dublin.

Awards

Drew R. The Faculty of Pathology (RCPI) George Green Medal for best research paper. **"The impact of Meningoccal PCR testing on laboratory confirmation of invasive meningococcal disease".**

Intensive Care

Initiated / Ongoing Research Projects

EPOCH. Multi center international study of evaluation of a bedside Pediatric Early Warning Score (PEWS).
Investigator: Dr. Dermot Doherty

Rate of Sedation Failure with Dexmedotomidine in a PICU: A retrospective cohort study.
Dexamedotomidine is a novel sedative agent in PICU. It has a failure-to sedate rate which has not been established.
Investigator: Dr. Dermot Doherty

Vitamin D and Immunomodulation In Paediatric Sepsis.
Infection is a serious problem for children in intensive care despite recent advances in treatment. Recent research has implicated vitamin D as having anti-inflammatory and immunomodulatory properties. Improving the understanding of the mechanisms of disease and the regulation of these inflammatory processes may lead to advances in diagnosis and new therapies to improve paediatric survival and neurodevelopmental outcome.
Investigators: Dr. Eleanor Molloy, Dr. Nuala Murphy, Dr. Dermot Doherty
Funded by the Children's Fund for Health. PAC-11-55

Peer Reviewed Publications

Neira VM, Bould MD, Nakajima A, Boet S, Barrowman N, Mossdorf P, Sydor D, Roeske A, Noseworthy S, Naik V, Doherty D, Writer H, Hamstra SJ. **"GIOSAT": a tool to assess CanMEDS competencies during simulated crises.** *Can J Anaesth.* 2013 Jan 19. [Epub ahead of print Dec 2012]

McNally JD, Menon K, Chakraborty P, Fisher L, Williams KA, Al-Dirbashi OY, Doherty DR; **The Association of Vitamin D Status With Pediatric Critical Illness.** *Pediatrics* 2012 Sep;130(3):429-36.

Clancy N, Onwuneme C, Carroll A, McCarthy R, McKenna M, Murphy M, Molloy EJ. **Vitamin D and Neonatal Immune Function.** *J Matern Fetal Neonatal Med.* Dec 2012 10. [Epub ahead of print]

Onwuneme C, Diya B, Uduma O, McCarthy R, McKenna M, Murphy N, Molloy EJ. **Hypovitaminosis D in Healthy Mothers and Infants: Resolution in Neonates on 200IU daily Vitamin D.**

Other Journal Publications

Doherty DR. **Pediatric Critical Care. 4th Edition** Bradley P. Fuhrman, Jerry J. Zimmerman. *Canadian Journal of Anesthesia* 2012(59):427-428.

Published Abstracts

Sattar F, Kandomany N, Gendi S, O'Dwyer T. Carson K. **A Rare Case of Spontaenous Esophageal perforation in a seven months-old infant.** Irish Paediatric Anaesthesia and Critical Care Society Annual Meeting; 2012 21 April.

Sattar F, Carson K, Doherty D. **NAVA – A true assertion of brain over machine: A case report of NAVA in a 5 years old with acute severe asthma.** Irish Paediatric Anaesthesia and Critical Care Society Annual Meeting; 2012 21 April.

Onwuneme C. **Vitamin D and Paediatric Intensive Care Patients"** Oral Paper, Irish-American Paediatric Society, Belfast, UK, Sept, 2012.

Onwuneme C, Doherty D, Healy M, Murphy N, McKenna M, Molloy EJ. **Vitamin D and Paediatric Intensive Care patients.** Irish & American Paediatric Society Meeting: Abstract book; 2012: Belfast, Northern Ireland, September 2012. P 32.

Onwuneme C, Martin F, McCarthy R, Murphy N, McKenna M, Molloy E. **Does Total Parenteral Nutrition raise serum Vitamin D level in Preterm Infants?** The 4th congress of the European Academy of Paediatric societies: 2012: Istanbul, Turkey: ADC 97: Suppl 2.

Onwuneme C, Eliwan HO, O'Hare FM, Watson RWG, O'Neill A, Molloy EJ. **Neonatal Endotoxin Tolerance: Enhanced Reactive Oxygen Intermediates but Diminished Monocyte and neutrophil Surface Antigen expression.** Proceedings of the Paediatric Academic Societies; 2012: Boston, Massachusetts; April 28, 2012.

Onwuneme C, Diya B, Uduma O, McCarthy R, McKenna M, Murphy N, Molloy EJ. **Increased Hypovitaminosis D in Preterm vs Term Mother Infant Pairs.** Proceedings of the Paediatric Academic Societies; 2012: Boston, Massachusetts; April 28, 2012.

Armstrong K, Onwunmeme C, Franklin O, Molloy E. **Vitamin D Levels and Myocardial Function in Preterm Infants.** The 4th Congress of the European Academy of Paediatric Societies; 2012: Istanbul, Turkey: *ADC* 97: Suppl 2.

Presentations at International Conferences

Doherty D. **Severe TBI; Paediatric Guideline versus Adult Guidelines.** Plenary Speaker. Association of Neuroanaesthesists of Great Britain and Ireland; May 2012 May: Belfast.

Doherty D. **How to stop brain cells dying; Plenary Lecture.** Paediatric Intensive Care Society Annual Scientific Meeting; Sept 2012: Dublin.

Onwuneme C, Doherty D, Healy M, Murphy N, McKenna M, Molloy EJ. **Vitamin D and Paediatric Intensive Care patients.** Poster presentation at the Paediatric Intensive Care Society Conference; 2012: Dublin, Ireland.

Presentations at National Conferences

Doherty D. **Paediatric Bed Bureau to PICU.ie:** Plenary Speaker. Intensive Care Society of Ireland; 2012 August: Dublin, Ireland.

Carson K. **Organ Donation: Paediatric Perspective.** Plenary Speaker Intensive Care Society of Ireland Autumn Meeting; 2012 November: Waterford, Ireland.

Doherty D. **Vitamin D in Critical Illness.** Irish Paediatric Anaesthesia and Critical Care Society Annual Meeting; 2012 April.

Onwuneme C, Doherty D, Healy M, Murphy N, McKenna M, Molloy EJ. **Decreased Vitamin D levels and Outcomes in Paediatric Intensive Care Patients.** Poster presentation at the North Dublin Hospital Group Inaugural Conference on Collaborative Lifetime Research; 2012: Dublin.

Onwuneme C, Diya B, Uduma O, McCarthy R, McKenna M, Murphy N, Molloy EJ. **Increased Hypovitaminosis D in Preterm vs Term Mother Infant Pairs.** Poster Presentation at the Irish Paediatric Association; 2012: Belfast, Northern Ireland.

Onwuneme C, Murphy N, Watson RWG, Molloy EJ. **Vitamin D and Immunomodulation in Paediatric Sepsis.** Oral Presentation at the Children's University Hospital Temple Street Research Day; 2012: Dublin.

Onwuneme C, Eliwan HO, O'Hare FM, Watson RWG, O'Neill A, Molloy EJ. **Altered Neonatal leukocyte phenotype: enhanced reactive oxygen intermediates but diminished monocyte and neutrophil surface antigen expression and endotoxin tolerance.** Poster Presentation at the Irish Paediatric Association; 2012: Belfast, Northern Ireland.

Onwuneme C, Diya B, Uduma O, McCarthy R, McKenna M, Murphy N, Molloy EJ. **Increased Hypovitaminosis D in Preterm vs Term Mother Infant Pairs.** Oral presentation at the 3rd Neonatal Research Symposium RCPI; 2012: Dublin.

Onwuneme C, Eliwan HO, O'Hare FM, Watson RWG, O'Neill A, Molloy EJ. **Neonatal Endotoxin Tolerance: Enhanced Reactive Oxygen Intermediates but Diminished Monocyte and neutrophil Surface Antigen expression.** Oral presentation at the 3rd Neonatal Research Symposium RCPI; 2012: Dublin.

Local Presentations

Neonatal Intensive Care Adverse Outcomes and Hypovitaminosis D; A future Paradigm?
Chike Onwuneme, Roberta McCarthy, Nuala Murphy, Mark Kilbane, Malachi McKenna, Eleanor J. Molloy. Oral presentation at the Audit / Research Day December 2012 at the children's University Hospital, Dublin.

Hanlon A. **CVC care bundles.** Oral presentation at the Audit / Research Day December 2012 at the children's University Hospital, Dublin.

Awards / Prizes

Onwuneme C. Awarded 1st Prize for best poster presentation at the the Irish Paediatric Association; 2012: Belfast, Northern Ireland.

Onwuneme C. Awarded the Gold Medal for best oral presentation at the Children's University Hospital Temple Street Research Day; 2012: Dublin.

Hanlon A. **CVC care bundles.** Awarded 2nd Prize for best oral presentation at the Audit / Research Day December 2012 at the children's University Hospital, Dublin.

Library

Peer Reviewed Publications

Burns J, Fallon H. **How to obtain Felowship status with the library Association of Ireland.** *An Leabharlam.* 2012 Oct (2) 14-24.

Awards / Prizes

Burns F. Awarded Fellowship Status of the Library Association of Ireland, March 19, 2012. Awarded for CPD Research and Commitment to the Library Profession in Ireland.

Medical Genetics

Initiated / Ongoing Research Projects

Identifying recessive genes for primary ciliary dyskinesia, microcephaly, & cardiomyopathy with retinopathy.
Our discoveries will lead to genetic tests that will allow earlier and more precise diagnoses. Better diagnoses will allow health care teams to reduce or prevent patient complications, to develop tailored treatments, and to provide more accurate reproductive counselling to families. In the long term, identification of disease genes is an essential step toward the development of drugs that will one day improve the lives of affected children.
Investigators: Dr Sally Ann Lynch [PI] and Dr Jillian Casey, Dr Amanda McCann

Identification of the genetic basis of a syndrome causing cleft palate, microcephaly & developmental delay in an Irish Traveller family attending Temple Street Hospital.
Our research aims to identify the underlying disease gene and develop a simple genetic test to enable faster diagnoses and avoid unnecessary and costly investigations.
The availability of our new genetic test impacts on the extended family as it will allow carriers of the disorder to be identified for the first time, improve genetic counselling with better estimates of recurrence risks, and facilitating primary prevention. Isolation of the causative gene is the first step towards development of new treatments and preventative strategies.
Investigators: Dr Sally Ann Lynch [PI] and Dr Jillian Casey
Part funded by the Children's Fund for Health Seed Funding SF-2012-04

Knowledge exchange & dissemination scheme for 'Identifying recessive genes for primary ciliary dyskinesia, microcephaly, & cardiomyopathy with retinopathy'
We have found it difficult to translate our work into the diagnostic setting. This grant will facilitate that by paying for essential training of laboratory staff. In addition, we are making first steps towards education of the health care professionals through articles in GP & Nurse journals.
Videos and SOPs will help GPs and healthcare professionals to counsel / advise families concerned about a genetic condition in their family....advise on genetic testing approach...provide info about risk of having affected child (ie a CF video is being planned and SOPs)
Investigators: Dr Sally Ann Lynch [PI], Dr Jillian Casey, Dr Amanda McCann

Peer Reviewed Publications

Casey JP, McGettigan P, Lynam-Lennon N, McDermott M, Regan R, Conroy J, Bourke B, O' Sullivan J, Crushell E, Lynch SA, Ennis S. 2012. **Identification of a mutation in LARS as a novel cause of infantile hepatopathy.** *Mol Genet Metab.* 2012, 106(3): 351-358.

Casey JP, Nobbs M, McGettigan P, Lynch SA, Ennis S. 2012. **Recessive mutations in MCM4 / PRKDC cause a novel syndrome involving a primary immunodeficiency and a disorder of DNA repair.** *J Med Genet.* 2012, 49(4): 242-245.

Magalhaes TR, Casey JP, Conroy J, Regan R, Fitzpatrick DJ, Shah N, Sobral J, Ennis S. 2012. **HGDP and HapMap analysis by Ancestry Mapper reveals local and global population relationships.** *PLOS One* 2012, 7(11): e49438.

Lynch SA, Nguyen LS, Ng LY, Waldron M, McDonald D, Gecz J. **Broadening the phenotype associated with mutations in UPF3B: two further cases with renal dysplasia and variable developmental delay.** *Eur J Med Genet.* 2012, 55(8-9): 476-479.

O' Rourke DJ, Twomey E, Lynch SA, King MD. 2012. **Cortical dysplasia associated with the PTEN mutation in Bannayan Riley Ruvalcaba syndrome: a rare finding.** *Clin Dysmorphol.* 2012, 21(2):91-92.

Vogt J, Morgan NV, Rehal P, Faivre L, Brueton LA, Becker K, Fryns JP, Holder S, Islam L, Kivuva E, Lynch SA, Touraine R, Wilson LC, Macdonald F, Maher ER.2012. **CHRNG genotype-phenotype correlations in the multiple pterygium syndromes.** *J Med Genet.* 2012, 49(1):21-26.

Deardorff MA, Wilde JJ, Albrecht M, Dickinson E, Tennstedt S, Braunholz D, Monnich M, Yan Y, Xu W, Gil-Rodriguez MC, Clark D, Hakonarson H, Halbach S, Michelis LD, Rampuria A, Rossier E, Spranger S, Van Maldergem L, Lynch SA, Gillessen-Kaesbach G, Ludecke HJ, Ramsay RG, McKay MJ, Krantz ID, Xu H, Horsfield JA, Kaiser FJ. 2012. **RAD21 mutations cause a human cohesinopathy.** *Am J Hum Genet.* 2012, 90(6): 1014-1027.

Banka S, Veeramachaneni R, Reardon W, Howard E, Bunstone S, Ragge N, Parker MJ, Crow YJ, Kerr B, Kingston H, Metcalfe K, Chandler K, Magee A, Stewart F, McConnell VP, Donnelly DE, Berland S, Houge G, Morton JE, Oley C, Revencu N, Park SM, Davies SJ, Fry AE, Lynch SA, Gill H, Schweiger S, Lam WW, Tolmie J, Mohammed SN, Hobson E, Smith A, Blyth M, Bennett C, Vasudevan PC, García-Miñaúr S, Henderson A, Goodship J, Wright MJ, Fisher R, Gibbons R, Price SM, C de Silva D, Temple IK, Collins AL, Lachlan K, Elmslie F, McEntagart M, Castle B, Clayton-Smith J, Black GC, Donnai D. 2012. **How genetically heterogeneous is Kabuki syndrome?: MLL2 testing in 116 patients, review and analyses of mutation and phenotypic spectrum.** *Eur J Hum Genet.* 2012, 20(4): 381-388.

Schrier SA, Bodurtha JN, Burton B, Chudley AE, Chiong MA, D'Avanzo MG, Lynch SA, Musio A, Nyazov DM, Sanchez-Lara PA, Shalev SA, Deardorff MA. 2012. **The Coffin-Siris syndrome: a proposed diagnostic approach and assessment of 15 overlapping cases.** *Am J Med Genet.* A 2012, 158A(8): 1865-1876.

White M, Conroy J, Bullman H, Lever M, Crolla J, Daly E, Betts DR, Cody D, Lynch SA. **Duplication of 17q11.2 and Features of Albright Hereditary Osteodystrophy Secondary to Methylation Defects within the GNAS Cluster: Coincidence or Causal?** *Case Reports in Genetics.* (In Press) http: //dx.doi.org/10.1155/2013/764152

Nowakowska BA, de Leeuw N, Ruivenkamp CA, Sikkema-Raddatz B, Crolla JA, Thoelen R, Koopmans M, den Hollander N, van Haeringen A, van der Kevie-Kersemaekers AM, Pfundt R, Mieloo H, van Essen T, de Vries BB, Green A, Reardon W, Fryns JP, Vermeesch JR. **Parental insertional balanced translocations are an important cause of apparently de novo CNVs in patients with developmental anomalies.** *Eur J Hum Genet.* 2012 Feb;20(2):166-70. PMID: 21915152.

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Books / Chapters

O' Connell C. **Digging into the genes behind rare diseases. In: Picture of Health 2012.** Health Research Board. p.26.
Our research investigating the genetic basis of rare pediatric disorders in the Irish population was selected for inclusion in the coveted Picture of Health 2012 Report issued by the Health Research Board. This publication highlights research that has translated research discoveries into real benefits for patients and the Irish health service. The article, entitled 'Digging into the genes behind rare diseases' highlighted our identification of new disease genes causing rare disorders in Irish families and the use of our research findings to develop genetic-based diagnostic tests to improve patient health and care.

Published Abstracts

Casey J, Green A, McGettigan P, Conroy J, Regan R, Ennis A, Lynch S.A. Poster Presentation. **Next-generation diagnostics for rare disorders in the Irish population.** New Frontiers Symposium: Personal Genomics, Nijmegen, Netherlands. December 2012.

Conroy J, McGettigan P, Walsh O, McDonagh C, Moran M, O'Mahoney O, Webb D, Murphy S.M, Murphy R, King M, Ennis S, Lynch A.S. Poster Presentation. **Exome sequencing in rare neurological conditions: The advantages of linkage mapping.** Neuroscience Ireland, Dublin, Ireland. September 2012.

Conroy J, McGettigan P, Collins K, Parry-Fielder B, Moran M, O'Mahoney O, Webb D, King M, Ennis S, Lynch S.A. Poster Presentation. **Identifying the genetic basis of Landau-Kleffner Syndrome.** Irish Society of Human Genetics, Dublin, Ireland. September 2012.

Bradley L, Dabir T, Murphy C, Murray D, McGillivary A, Lynch S.A. Poster Presentation. **A retrospective analysis of the prevalence of craniosynostosis on the island of Ireland.** Irish Society of Human Genetics, Dublin, Ireland. September 2012.

Conroy J, McGettigan P, Moran M, O'Mahoney O, Webb D, King M, Ennis S, Lynch S.A. **Platform Presentation. Landau-Kleffner syndrome: CNV and exome sequencing analysis.** Neuroscience Ireland, Dublin, Ireland. September 2012.

Casey J, Green A, McGettigan P, Conroy J, Regan R, Ennis S, Lynch S.A. Poster Presentation. **An alternative approach to research: Technology driven research for rare disorders in the Irish population.** UK Clinical Research Facility Network Conference, Dublin, Ireland. July 2012.

J Casey J.P, Nobbs M, McGettigan P, Lynch S, Ennis S. Poster Presentation. **Recessive mutations in MCM4 / PRKDC cause a novel syndrome characterised by a primary immunodeficiency and impairments in DNA repair.** European Society of Human Genetics, Nurnberg, Germany. June 2012.

Casey J, McGettigan P, Alkazemi N, Maguire P, Kennedy B, Brosnahan D, Treacy E, Walsh K, Ennis S, Lynch S.A. Platform Presentation. **Exome analysis and cardiomyopathy: The Lazarus story.** Irish Society of Human Genetics, Dublin, Ireland. September 2012.

Lynch S.A, Turner J, Casey J. Poster Presentation. **Making matches: linking large Irish Traveller pedigrees as a way of helping gene identification.** Irish Society of Human Genetics, Dublin, Ireland. September 2012.

Conroy J, McGettigan P, Webb D, Walsh O, McDonagh C, Murphy S.M, Murphy R, Ennis S, Lynch S.A. Poster Presentation. **Combining linkage mapping with exome sequencing data.** Ataxia Research Conference, London, UK. November 2012.

Lynch S.A, Akram M, Goggin N, Earley M, Ennis S, Conroy J. Poster Presentation. **Ptosis, arched eyebrows, hypernasal speech, obesity & mild learning disability- a clinical & mapping study.** European Society of Human Genetics, Nurnberg, Germany. June 2012.

Palomares Bralo M, Delicado A, Mansilla E, de Torres M.L, Vallespín E, Fernández L, Martínez-Glez V, García Miñaur S, Nevado J, Santos Simarro F, Ruíz-Pérez V.L, Lynch S.A, Sharkey F.H, Thuresson A.C, Annerén G,Belligni E.F., Martínez

Fernández M.L, Bermejo E, Nowakowska B, Bocian E, Obersztyn E, Martínez Frías M.L, Hennekam R.C.M, Lapunzina P. Poster Presentation. **Characterization of a 8q21.11 microdeletion syndrome associated with intellectual disability and a recognizable phenotype.** European Society of Human Genetics, Nurnberg, Germany. June 2012.

Casey J, Nobbs M, McGettigan P, Lynch S.A, Ennis S. Platform Presentation. **A single mutation that affects two genes; MCM4 and PRKDC cause a primary immunodeficiency, disorder of DNA repair and familial glucocorticoid deficiency.** Our Lady's Children's Hospital Research and Audit Day, Dublin, Ireland. May 2012.

Conroy J, McGettigan P, Walsh O, McDonagh C, Murphy R, Webb D, Ennis S, Lynch S.A. Poster Presentation. **A novel locus for autosomal dominant episodic ataxia.** Our Lady's Children's Hospital Research and Audit Day, Dublin, Ireland. May 2012.

Casey J, Slattery D, Conroy J, Regan R, Ennis S, Lynch S.A. Platform Presentation. **Exome sequencing and the challenge of rare disorders.** National Children's Research Centre Seminar Series, Our Lady's Children's Hospital, Crumlin, Dublin, Ireland. January 2012.

Presentations at International Conferences

Lynch S.A, Turner J, Ennis S, Casey J. Poster Presentation. **Making matches- linking large Irish Traveller pedigrees to; a) aid with disease gene identification & b) understand the demographics of this endogamous community.** UK Dysmorphology Meeting. Manchester, UK. October 2012.

Lynch S.A, Turner J, Ennis S, Casey J. Platform Presentation. **Making matches- linking large Irish Traveller pedigrees to; a) aid with disease gene identification & b) understand the demographics of this endogamous community.** Irish and American Paediatric Society Meeting. Belfast, Ireland. September 2012.

Presentations at National Conferences

Conroy J, McGettigan P , Walsh O (3), McCoy B (3), Albertyn C, McGillivary A, Moran M , Goggin N, Ahmed M, O'Mahoney O, Korff C , Deonna T, Hanrahan D, Earley M , Webb D, Murphy S, Murphy R, Ennis S, King M, Lynch S.A. & Ennis S. **The hope and hype of exomic sequencing.** North Dublin Hospital Group Conference; Rotunda Hospital. November 2012; Dublin.

Local Presentaitons

Conroy J, McGettigan P, Collins K, Parry-Fielder B, Moran M, O'Mahoney O, Webb D, Ennis S, King M, Lynch S.A. **Platform Presentation. Exome sequencing – an approach to unlock the secrets of rare disorders.** Children's University Hospital Audit and Research Day, Temple Street, Dublin, Ireland. June 2012.

Awards / Prizes

Dr Jillian Casey was awarded Best Post-Doctoral Oral Presentation at the Irish Society of Human Genetics Conference 2012 for her talk entitled **'Exome analysis and cardiomyopathy: The Lazarus story.'** The research presented was partly funded by CFFH.

Dr Jillian Casey was selected as a poster award candidate at the European Society of Human Genetics Conference 2012 for her poster entitled **'Recessive mutations in MCM4 / PRKDC cause a novel syndrome characterised by a primary immunodeficiency and impairments in DNA repair.'** Only the top 2% of most highly scored abstracts were selected for poster candidate awards.

Dr Sally Ann Lynch and Dr Jillian Casey successfully secured additional funding (totaling ~€10,000) for this grant through the HRB knowledge and dissemination scheme (KEDS) in July 2012. The additional funding serves to maximize the impact of our research amongst the wider medical and public communities.

Dr Sally Ann Lynch [PI] and Dr Jillian Casey successfully secured a grant from the Children's Fund for Health, Temple Street University Hospital for€4,783.33 **Identification of the genetic basis of a syndrome causing cleft palate, microcephaly & developmental delay in an Irish Traveller family attending Temple Street Hospital.**
Applicants: Duration: August 2012-July 2013

Microbiology

Peer Reviewed Publications

Meyler KL, Meehan M, Bennett D, Cunney R, Cafferkey M. **Development of a diagnostic real-time polymerase chain reaction assay for the detection of invasive Haemophilus influenzae in clinical samples.** *Diagn Microbiol Infect Dis.* 2012 Dec;74 (4):356-62.

McDermott H, Morris D, McArdle E, O'Mahony G, Kelly S, Cormican M, Cunney R. **Isolation of NDM-1-producing Klebsiella pnemoniae in Ireland,** July 2011. *Euro Surveill.* 17, (2012).

Other Journal Publications

O'Lorcain P, Cotter S, Cunney R, O'Flanagan D, HPSC, O'Sullivan N, IMMRL, Meyler K EMBU. **Epidemiology of invasive Haemophilus influenzae disease in Ireland 2004-2011.** *Epi-Insight* 2012 May; 13(5)

Presentations at International Conferences

Mullhall R. **Nested Multi-Locus Sequence Typing (MLST) of meningococci directly from small volume of clinical samples.** Poster presentation by Mr. Robert Mullhall at the International Pathogenic Neisseria Conference 2012 in Germany.

Presentations at National Conferences

Meyler KL. **The Changing Epidemiology of Invasive Haemophilus influenzae in Ireland.** Oral presentation at conference entitled **"Vaccine Preventable Diseases in Ireland-Where are we now!"** Friday 18th May 2012 Academy of Medical Laboratory Sciences Microbiology Advisory Body, Trinity Centre for Health Sciences, St James's Hospital, Dublin.

Local Presentations

Analysis of Neisseria meningitidis serogroup B PorA genotypes and expression in Ireland. Marty Mulhall and Robert Cunney. Oral presentation at the Audit / Research Day June 2012 at the Children's University Hospital, Dublin.

National Centre for Inherited Metabolic Disorders

Initiated / Ongoing Research Projects

Four decades of Newborn Screening for Maple Syrup Urine Disease (MSUD) in the Republic of Ireland: A longitudinal study of the effects of early diagnosis and longitudinal metabolic control.
This study will significantly add to the knowledge of outcome data by compiling comprehensive data over a large period of time (1972-2011). This will highlight important aspects of therapy outcome for patients with MSUD. The study provides the opportunity to give an Irish perspective to the international experience with this rare metabolic disorder as we aim at reviewing comprehensive biochemical, molecular genetic and clinical outcome data of 17 patients detected as having MSUD by newborn screening in the Republic of Ireland.
We propose that this research will offer new insights into the long-term management of this rare disorder and also into its complex pathophysiology, including the identification of predictive factors for treatment outcome, which over time could be cost-effective by mitigating disease-related disability in patients with MSUD.
Investigators: Dr.Ina Knerr (PI), Prof. Eileen Treacy and Prof. Philip Mayne

MPSVI Clinical Surveillance Programme. March 2008 to date.
Disease specific rare disease database which monitors outcome of intervention globally and thus potentially can improve health outcomes for this and related diseases.
Investigator: Prof. Treacy PI. Funded by Biomarin Company.

Galactosaemia, a modifiable multi-system glycosylation disorder?
Potential to greatly improve the treatment of Galactosaemia by detecting improved biomarkers to understand the disease and to target specific treatments and improve current dietary treatment
Investigators: Prof. Treacy PI Co-applicants, Peter Doran, Pauline Rudd and Ina Knerr. Funded by MRCG-HRB grant.

Epidemiological study of Hereditary Metabolic Diseases in adults
Important study to plan for transition from child orientated health programme to adult Centre of Expertise for Hereditary Metabolic Diseases Prof. Treacy PI (2nd lead in Steering Committee) since 2012. Funded by European Galactosaemia Clinical and Research Consortium

Four decades of Newborn Screening for Maple Syrup Urine Disease (MSUD) in the Republic of Ireland: A longitudinal study of the effects of early diagnosis and longitudinal metabolic control.
This study will significantly add to the knowledge of outcome data by compiling comprehensive data over a large period of time (1972-2011). This will highlight important aspects of therapy outcome for patients with MSUD.
The study provides the opportunity to give an Irish perspective to the international experience with this rare metabolic disorder.
We propose that this research will offer new insights into the long-term management of this rare disorder and also into its complex pathophysiology, including the identification of predictive factors for treatment outcome, which over time could be cost-effective by mitigating disease-related disability in patients with MSUD.
Investigator: Dr Ina Knerr
Funded by CFFH-SF-2013-02 (first year); Vitaflo - International Ltd (2nd year, unrestricted research grant)

Linkage analysis to identify candidate genes for epilepsy and regression in an Irish family
€4,910 We hope to identify candidate genes that may harbour mutations resulting in the development of epilepsy with regression, post-natal microcephaly and mitochondrial dysfunction and thus provide a diagnosis for this family and may enable the development of a simple genetic test which would be of clinical use in diagnosis, management, carrier testing and prognosis of patients and their families presenting with similar features
Investigators: Ellen Crushell (PI), Judith Conroy
Part-funded by the Children's Fund for Health Seed Fund SF-2012-06

An EU rare diseases registry for Niemann Pick Disease
Awarded: €24,840 (total consortium grant = €690,793)
Investigator: Ellen Crushell.
Sponsor: DG Sanco funded European Collaborative Studies 2012
Grant Number: 308901

European network and registry for homocystinurias and methylation defects
Awarded: €27,989 (total consortium grant = €1,200,000)
Investigators: Ellen Crushell, AA Monavari
Sponsor: DG Sanco funded European Collaborative Studies 2012
Grant Number: #297298

Amino Acid Profiles versus dietary intake in MSUD.
Investigator: Jane Rice

Review of biochemical monitoring of patients with PKU.
Investigators: Maria O'Regan, Celine Stenson & Fiona Boyle.

Peer Reviewed Publications

Knerr I, Weinhold N, Vockley J, Gibson KM. **Advances and challenges in the treatment of branched-chain amino / keto acid metabolic defects.** *J Inherit Metab Dis.* 2012;35(1):29-40.

Kraus JP, Spector E, Venezia S, Estes P, Chiang PW, Creadon-Swindell G, Müllerleile S, de Silva L, Barth M, Walter M, Walter K, Meissner T, Lindner M, Ensenauer R, Santer R, Bodamer OA, Baumgartner MR, Brunner-Krainz M, Karall D, Haase C, Knerr I, Marquardt T, Steinfeld R, Beblo S, Koch HG, Konstantopoulou V, Scholl-Bürgi S, van Teeffelen-Heithoff A, Suormala T, Ugarte M, Sperl W, Superti-Furga A, Schwab KO, Grünert SC, Sass JO. **Mutation analysis in 54 propionic acidemia patients.** *J Inherit Metab Dis.* 2012;35(1):51-63.

Grünert SC, Müllerleile S, de Silva L, Barth M, Walter M, Walter K, Meissner T, Lindner M, Ensenauer R, Santer R, Bodamer OA, Baumgartner MR, Brunner-Krainz M, Karall D, Haase C, Knerr I, Marquardt T, Steinfeld R, Beblo S, Koch HG, Konstantopoulou V, Scholl-Bürgi S, van Teeffelen-Heithoff A, Suormala T, Sperl W, Kraus JP, Superti-Furga A, Schwab KO, Sass JO. **Propionic acidemia: neonatal versus selective metabolic screening.** *J Inherit Metab Dis.* 2012 ;35(1):41-49.

K P Coss; J C Byrne; D J Coman; B Adamczyk; J L Abrahams; R Saldova; A Y Brown; O Walsh; U Hendroff; C Carolan; P M Rudd; E Treacy. **IgG N-glycans as potential biomarkers for determining galactose tolerance in Classical Galactosaemia.** *Molecular Genetics and Metabolism*, 2012, 105 (2), 212-20.

Shephard EA, Treacy EP, Philips IR. **Clinical utility gene card for Trimethylaminuria.** *Eur J Hum Genet.* 2012, Mar, 20 (3).

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Coss KP, Doran PP, Owoeye C, Codd MB, Hamid N, Mayne PD, Crushell E, Knerr I, Monavari AA, Treacy EP. **Classical Galactosaemia in Ireland: Incidence, complications and outcomes of treatment.** *J Inher. Metab.* Dis. 2013, Jan; 36 (1) 21, Epub 2012, Jul 3.

Knerr I, Coss KP, Doran PP, Hughes J, Wareham N, Burling K, Treacy EP. **Leptin Levels in Children and Adults with Classic Galactosaemia Research Report:** *J. Inherit Metab Dis,* 9,125-131, 2013, Epub 2012, Nov 7.

Casey JP, McGettigan P, Lynam-Lennon N, McDermott M, Regan R, Conroy J, Bourke B, O'Sullivan J, Crushell E, Lynch S, Ennis S. I**dentification of a mutation in LARS as a novel cause of infantile hepatopathy.** *Molecular Genetics and Metabolism.* 2012 Jul;106(3):351-8.

Rigat BA, Tropak MB, Buttner J, Crushell E, Benedict D, Callahan JW, Martin DR, Mahuran DJ. **Evaluation of N-nonyl-deoxygalactonojirimycin as a pharmacological chaperone for human GM1 gangliosidosis leads to identification of a feline model suitable for testing enzyme enhancement therapy.** *Molecular Genetics and Metabolism.* 2012 Sept 107:203-212.

Crushell E, O'Leary D, Irvine AD, O'Shea A, Mayne PD, Reardon W. **Methylenetetrahydrofolate reductase (MTHFR) deficiency presenting as a rash.** *American Journal of Medical Genetics* A. 2012;158:2254-2257.

Nesbitt V, Morrison P, Crushell E, Donnelly D, Alston CL, Langping He,McFarland R and Taylor RW. **The clinical spectrum of the m.10191T>C mutation in complex I-deficient Leigh Syndrome.** *Developmental Medicine and Child Neurology* 2012; 54(6):500-506.

Fitzgerald M, Crushell E, Hickey C. **Cyclic vomiting syndrome masking a fatal metabolic disease.** *Eur J Pediatr.* 2012 Oct 11. [Epub ahead of print]

Knerr I, Weinhold N, Vockley J, Gibson KM. **Advances and challenges in the treatment of branched-chain amino / keto acid metabolic defects.** *J Inherit Metab Dis.* 2012;35(1):29-40.

Kraus JP, Spector E, Venezia S, Estes P, Chiang PW, Creadon-Swindell G, Müllerleile S, de Silva L, Barth M, Walter M, Walter K, Meissner T, Lindner M, Ensenauer R, Santer R, Bodamer OA, Baumgartner MR, Brunner-Krainz M, Karall D, Haase C, Knerr I, Marquardt T, Steinfeld R, Beblo S, Koch HG, Konstantopoulou V, Scholl-Bürgi S, van Teeffelen-Heithoff A, Suormala T, Ugarte M, Sperl W, Superti-Furga A, Schwab KO, Grünert SC, Sass JO. **Mutation analysis in 54 propionic acidemia patients.** *J Inherit Metab Dis.* 2012; 35(1):51-63.

Grünert SC, Müllerleile S, de Silva L, Barth M, Walter M, Walter K, Meissner T, Lindner M, Ensenauer R, Santer R, Bodamer OA, Baumgartner MR, Brunner-Krainz M, Karall D, Haase C, Knerr I, Marquardt T, Steinfeld R, Beblo S, Koch HG, Konstantopoulou V, Scholl-Bürgi S, van Teeffelen-Heithoff A, Suormala T, Sperl W, Kraus JP, Superti-Furga A, Schwab KO, Sass JO. **Propionic acidemia: neonatal versus selective metabolic screening.** *J Inherit Metab Dis.* 2012; 35(1):41-49.

Other Journal Publications

Neu A, Beyer P, J. Bürger-Büsing, T. Danne, J. Etspüler, B. Heidtmann, R. W. Holl, B. Karges, W. Kiess, I. Knerr I, O. Kordonouri, K. Lange, R. Lepler, W. Marg, A. Näke, M. Petersen, A. Podeswik, R. Stachow, S. von Sengbusch, V. Wagner, R. Ziegler, P. M. Holterhus. **Diagnosis and treatment of type 1 diabetes in children (in German).** *Diabetologie und Stoffwechsel* 2012; 7: S152-S162.

Books / Chapters

Vockley J, Zschocke J, Knerr I, Walsh Vockley C, Gibson KM. **Branched Chain Organic Acidurias. In: Scriver's Online Metabolic and Molecular Bases of. Inherited Disease.** McGraw-Hill, September 2012, www.ommbid.com

Vockley J, Zschocke J, Knerr I, Walsh Vockley C, Gibson KM. **Branched Chain Organic Acidurias. In: Scriver's Online Metabolic and Molecular Bases of. Inherited Disease.** McGraw-Hill, September 2012, www.ommbid.com

Published Abstracts

Knerr I, Coss CP, Doran PP, Hughes J, Treacy EP. **Leptin levels in adults and children with classic galactosaemia.** *J Inher Metab Dis* 2012, Suppl 1, S78, 2012: Birmingham, England: Annual Symposium of the Society for the Study of Inborn Errors of Metabolism (SSIEM), 4-7 September 2012

Coss KP, Doran PP, Byrne JC, Adamczyk B, Rudd PM, Treacy EP. **Classical Galactosaemia- a modifiable glycosylation defect. 2012.** *Molec. Genet.* Metab 105(3), p 290.

J Casey, P McGettigan, A Alkazemie, P Maguire, B Kennedy, D Brosnahan, EP Treacy, K Walsh, SA Lynch. **Exome analysis and cardiomyopathy: The Lazarus story.** Irish Society of Human Genetics annual meeting, Sept 3rd, 2012. Published Abstract, *Ulster Med J,* 2012, 81 (3) S01.

Coss KP, Doran PP, Byrne JC, Adamcyck B, Rudd PM, Treacy EP. **Classical Galactosaemia- a modifiable Glycosylation Disorder?** Irish Society of Human Genetics annual meeting, Sept 3rd, 2012. Published Abstract, *Ulster Med J,* 2012, 81 (3), P08.

Knerr I, Coss KP, Doran P, Hughes J, Treacy EP. **Leptin levels in adults and children with classic Galactosaemia.** *J Inherit Metab Dis* (2012) (35), suppl 1, Abs P181.

E. Jameson, B.M. Wilson, A. Broomfield, S. Santra, E. Crushell, S.A . Jones. **I-cell disease: A review of the experience of five centres.** International Symposium on MPS and Related Diseases, The Netherlands, June 2012. Conference Proceedings P008 p.70.

Presentations at International Conferences

Knerr I, Coss CP, Doran PP, Hughes J, Treacy EP. **Leptin levels in adults and children with classic galactosaemia.** Poster presentation at the Annual Symposium of the Society for the Study of Inborn Errors of Metabolism (SSIEM), September 2012, Birmingham, England

Treacy EP (with Dr. Estela Rubio-Gozalbo) Co-Chair: European Galactosaemia Consortium meeting at 2012 SSIEM meeting, Birmingham, September 6th, 2012.

Coss KP, Doran PP, Byrne JC, Adamczyk B, Rudd PM, Treacy EP. **Classical Galactosaemia- a modifiable glycosylation defect.** Annual US SIMD, US Society for Inborn Errors of Metabolism Meeting, Charlotte, North Carolina, April 1st, 2012.

Brinkley A, Keating M., Lynch A., Monavari A., Crushell, E., Treacy. E. **Revisiting IQ in adults with PKU: The impact of biochemical control.** Poster presentation: British Inherited Metabolic Diseases Group, Annual Meeting, Cardiff.

Knerr I, Coss KP, Doran P, Hughes J, Treacy EP. **Leptin levels in adults and children with classic Galactosaemia.** Poster presentation: SSIEM 2012 Annual Meeting, Birmingham, Sept 4-7, 2012.

E. Jameson, B.M. Wilson, A. Broomfield, S. Santra, E. Crushell, S.A . Jones. **I-cell disease: A review of the experience of five centres.** International Symposium on MPS and Related Diseases, June 2012, Amsterdam, The Netherlands,

Clark A. **The Treatment Challenge:Management of Emergency Regime at Home during Periods of Illness.** SSIEM Dietitians meeting 2012.

McNulty J. **The Dietary Management of GA1 – An Irish Perspective.** Oral Presentation Vitaflo Metabolic Meeting in Gothenberg.

Presentations at National Conferences

Knerr, I. **Metabolic Emergencies.** Oral presentation at the Neonatal Study Day, 27th April 2012, Limerick.

Treacy EP. Presentation: **Incidence and Outcomes of Galactosaemia.** Chairperson and Main Organiser: 'Workshop on Galactosaemia' held, at the Gresham Hotel, Dublin, January 21st, 2012.

Treacy EP. Chairperson: 'Diagnosis' Workshop, Institute of Public Health organised National Consultation Day for the Irish Rare Disease Plan, Farmleigh, June 11th, 2012.

Treacy EP. **The Irish Galactosaemia Story. (Or why sugar is sweet from birth to old age)'.** Invited speaker: North Dublin Hospital Group Collaborative Lifespan Research Day. Rotunda Hospital, Dublin, November 29th, 2012.

Costigan D, Farrell M, Treacy EP, Lawler L, Taylor R, Turnbull D, Lochmuller H. **Disabling late novel compound heterozygous mutation of the GNE gene (hereditary inclusion body myositis, IBM2).** Clinical, electromyographic, radiologic, histologic and molecular genetic features. Oral presentation: Irish Neurological Association Annual Meeting, May 12th, 2012, Cork.

Brinkley A, Keating M, Lynch A, Monavari AA, Crushell E, Guerin S, Treacy EP. **Executive functioning, IQ and concurrent phenylalanine concentrations in Irish adults with PKU.** Oral Presentaion at the Audit / Research Day June 2012 at the Children's University Hospital, Dublin.

J Casey, P McGettigan, A Alkazemie, P Maguire, B Kennedy, D Brosnahan, EP Treacy, K Walsh, SA Lynch. Platform presentation: **Exome analysis and cardiomyopathy: The Lazarus story.** Irish Society of Human Genetics annual meeting, Sept 3rd, 2012.

Coss KP, Doran PP, Byrne JC, Adamcyck B, Rudd PM, Treacy EP. **Classical Galactosaemia- a modifiable Glycosylation Disorder?** Poster presentation. Recipient of Best Poster Award at ISHG 2012 Annual meeting.

Coss KP, Hawkes CP, Cxrushell E, Adamcyck B, Saldova R, Knerr I, Monavari AA, Rudd PM, Treacy EP. **IgG N-glycan abnormalities in children with Galactosaemia** Poster presentation: 5th Annual Glycoscience Ireland Conference, 19th October 2012.

Knerr, I. **Metabolic Emergencies.** Invited Speaker: The Neonatal Study Day, 27th April 2012, Limerick.

Maria O'Regan, Celine Stenson. Presentation on **"The role of the CNS within the metabolic team in CUH"** CLIMB first National conference, Dublin, October 2012.

Maria O'Regan, Celine Stenson. **Presentation on Metabolic Disorders within the Traveller population** Sligo Traveller Primary Care, Awareness Event, Sligo, November 2012.

Local Presentations

Knerr I, Coss CP, Doran PP, Hughes J, Treacy EP. **Does leptin levels dysregulation contributre to long-term complications in patients with classic galactosaemia?** Oral presentation at the Chiildren's University Hospital Audit / Research Day, 29th June 2012.

McElligott F, Beatty E, O'Sullivan S, Hughes J, Lambert D, Cooper A, Crushell E. **Incidence of I-cell disease (mucolipidosis type II) in the Irish population.** Oral presentation at the Audit / Research Day June 2012 Children's University Hospital, Dublin.

Awards / Prizes

Coss KP, Doran PP, Byrne JC, Adamczyk B, Rudd PM, Treacy EP. **Classical Galactosaemia- a modifiable glycosylation defect'.** Annual US SIMD, US Society for Inborn Errors of Metabolism Meeting, Charlotte, North Carolina, April 1st, 2012. K. Coss: Travel award recipient and winner of overall SIMD 2012 Neil Buist Award (trainee award) for this presentation.

Coss KP, Doran PP, Byrne JC, Adamcyck B, Rudd PM, Treacy EP. Poster presentation: **Classical Galactosaemia- a modifiable Glycosylation Disorder?** Recipient of Best Poster Award at ISHG 2012 Annual meeting.

Walsh A. **Doctors Knowledge of Metabolic Disorders.** Awarded best SHO Grand Rounds Presentation TSCUH Jul-Dec 2013

O'Byrne J. **Mitochondrial Disease.** Awarded best SHO Grand Rounds Presentation. TSCUH Jan- Jul 2013.

National Paediatric Mortality Register

Initiated / Ongoing Research Projects

National Registry and research on Paediatric Mortality in the Republic of Ireland.
This project involves the development of a system for the ongoing monitoring and compiling of information on all paediatric deaths in Ireland. The core dataset provides valuable information on the current status of the national paediatric population (<15 yrs) enabling population trends and events in a number of pertinent areas such accidental and sudden unexplained deaths to be tracked accurately in a timely manner. Identification of potentially modifiable risk factors will provide the basis for future intervention strategies.
Investigators: Cliona McGarvey and Karina Hamilton

Peer Reviewed Publications

McGarvey C, O'Regan M, Cryan J, Treacy A, Hamilton K, Devaney D and Matthews T. **Sudden unexplained death in childhood (1-4yrs) in Ireland: an epidemiological profile and comparison with SIDS.** *Arch Dis Child.* 2012.;97:8 692-697 Published Online First: June 2012 doi: 10.1136 / archdischild-2011-301393.

McDonnell M, McGarvey C, O'Regan M, Matthews T. **Maternal smoking and alcohol consumption during pregnancy as risk factors for Sudden Infant Death Syndrome.** *IMJ* 2012 Vol 105 (4).

Presentations at International Conferences

McDonnell M, McGarvey C, O'Regan M, Matthews T. **Maternal smoking and alcohol consumption during pregnancy as risk factors for Sudden Infant Death Syndrome.** Oral presentation at the 2012 International Conference on Stillbirth, SIDS and Infant Survival; October 4-7, 2012: Baltimore, Maryland, USA.

Presentations at National Conferences

McGarvey C, O'Regan M, Cryan J, Treacy A, Hamilton K, Devaney D and Matthews T. **Sudden unexplained death in childhood (1-4yrs) in Ireland: an epidemiological profile and comparison with SIDS.** Poster presentation at the North Dublin Hospital Group Conference, Rotunda Hospital, Dublin November 2012.

Neonatology

Initiated / Ongoing Research Projects

3.0 Tesla MRI of the Brain at term and Neurodevelopmental exam of Infants with Intrauterine Growth Restriction.
Growth restricted infants are at increased risk of learning difficulties and concentration problems. Using MRI brain imaging and sophisticated analysis we hope to identify those at most risk within this group. In future, we hope this will help clinicians prioritise services that those at highest risk would benefit from.
Investigators: Michael Boyle (PI), Stephanie Ryan, Ailbhe Tarrant and Adrienne Foran.
Funded by the Children's Fund for Health Pac-11-53

MRI Brain in Twin-Twin Transfusion Syndrome Treated with Fetoscopic Laser Surgery
Fetoscopic laser surgery is currently the best treatment modality for Twin-Twin transfusion syndrome. We sought to assess MRI brain findings in groups of twins to investigate for MRI changes. This study supports the fact that this high risk group would benefit from routine MRI brain at term, as important findings may be missed on ultrasound alone.
Investigators: Michael Boyle (PI), Aisling Lyons, Stephanie Ryan, Fergal Malone and Adrienne Foran.

The Use of Targeted Neonatal Echocardiography to Predict Short Term Clinical Sequelae Associated with a Patent Ductus Arteriosus.
To individual subjects: The only likely benefit to your baby would be the chance finding of an undiagnosed heart or blood vessel condition. This would then exclude your baby from further participation and we would arrange for suitable follow-up and medical care.
To Society: Having a better understanding of how the heart works in preterm babies will open the door to advancement of the way we identify and treat diseases associated with the pretem heart.
Investigator: Afif EL-Khuffash

The Use of Echocardiographic and Biochemical Markers to Predict Low Cardiac Output Following PDA Ligation in Preterm Infants.
Toronto Canada. Infants undergoing PDA ligation are at risk of developing PLCS characterised by cardio-respiratory compromise. Left ventricular (LV) output (LVO)<200ml / kg / min in the immediate post operative period is predictive of PLCS and benefits from milrinone treatment. The impact of milrinone on cardiac troponin T (cTnT), LV tissue Doppler indices (TDI), and global LV longitudinal strain (GLS) is unknown.
Investigator: Afif EL-Khuffash

Non-invasive cardiac output monitoring in infants following PDA ligation.
Toronto. Canada. Preterm infants undergoing PDA ligation are at risk of developing post-op low left ventricular output (LVO) which leads to subsequent cardiorespiratory instability termed post ligation cardiac syndrome (PLCS). Echocardiography (echo) is the tool of choice currently used to identify low LVO. Prophylactic milrinone use in these high risk infants prevents PLCS. Continuous LVO monitoring using NICOM may help identify high risk infants in centers without access to echo.
Investigator: Afif EL-Khuffash

Newborn Encephalopathy Study. Help identify modifiable risk factors in pathway of HIE.
Investigators: Dr. Breda Hayes and Prof. Mary King.

Near Infra red spectroscopy in preterm babies with patent ductus arteriosus.
Assessing the impact of ibuprofen treatment for ducts on ten brain oxygenation of preterm babies. In a study of 20 preterms this study showed no effect on tissues oxygenation to either brain or gut pre or post treatment.
Investigators: Dr. Kathleen Gorman and Dr. Adrienne Foran

AnSer: A Multicentre Clinical Evaluation of a Decision Support Algorithm for Neonatal Seizure Detection, Phase1.
Collaborator Wellcome Trust Strategic Award for Multicentre national study of a seizure detection EEG algorithm for newborns Principal Investigator: Professor Geraldine Boylan UCC –E 3 million this will recruit from 7 international sites trying to develop software that detects new-born seizures. Recruitment to begin mid 2013
Investigators: Dr. Geraldine Boylan (UCC Cork), Dr. Janet Rennie (UCL London), Dr. Adrienne Foran (PI Rotunda)

To Establish Reference Ranges for Coagulation Parameters and to Characterize Plasma Thrombin Generation in Very Preterm Infants

This research project aims to establish normal ranges for plasma coagulation tests in preterm infants < 30 weeks gestational age, and to investigate the relationship of the test results with morbidity and mortality in this population.
Investigators: Prof Naomi McCallion, Dr Elaine Neary
Co-funded from the Children's Fund for Health (RPAC-12-62) and the National Children's Research Centre

Peer Reviewed Publications

EL-Khuffash A, Jain A, Dragulescu A, McNamara PJ, Mertens L. **Acute Changes in Myocardial Systolic Function in Preterm Infants Undergoing Patent Ductus Arteriosus Ligation. A Tissue Doppler and Myocardial Deformation Study.** *J Am Soc Echocardiogr.* 2012 Oct;25(10):1058-67.

Weisz D, Jain A, McNamara P, EL-Khuffash A. **Non-invasive Cardiac Output Monitoring in Neonates Using Bioreactance: A Comparison with Echocardiography.** *Neonatology.* 2012 Apr 11:102(1):61-67.

EL-Khuffash A, Mertens L. **Early Treatment of a Patent Ductus Arteriosus. Do We Know What We Are Doing?** *Pediatr Crit Care Med* 2012 May;13(3):363-4.

EL-Khuffash A, Unger S. **The Concept of Milk Kinship in Islam: Issues Raised when Offering Preterm Infants of Muslim Families Donor Human Milk.** *J Hum Lact.* 2012 2012 May;28(2):125-7.

Jain A, McNamara PJ, Ng E, EL-Khuffash A. **The Use of Targeted Neonatal Echocardiography (TnECHO) to Confirm Placement of Peripherally Inserted Central Catheters in Neonates.** *Am J Perinatol.* 2012 Feb;29(2):101-6.

Jain A, Sehgal M, EL-Khuffash A, Khadawardi E, McNamara PJ. **Use of Targeted Neonatal Echocardiography to Prevent Postoperative Cardiorespiratory Instability After PDA Ligation.** *J Pediatr.* 2012 Apr; 160(4): 584-589.e1.

Hayes BC, Cooley S, Donnelly J, Doherty E, Grehan A, Madigan C, McGarvey C, Mulvany S, Ryan S, Gillian J, Geary MP, Matthews TG, King MD. **The placenta in infants >36 weeks. Gestation with neonatal encephalopathy: a case control study.** *Arch Dis Child Fetal Neonatal Ed.* 2012 July 12 (epub ahead of print).

Hayes BC, Doherty E, Grehan A, Madigan C, McGarvey C, Mulvany S, Geary MP, Matthews TG, King MD. **Are serum markers of liver and muscle injury useful in neonatal hypoxic-ischaemic encephalopathy?** *Journal of Neonatal – Perinatal medicine.* 2012; 5: 305-310.

Geoghegan S, Foran A, Twomey A, Murphy BP, Clarke. **Regionalisation and neonatal networks.** *T. Ir Med J.* 2012 Jul-Aug; 105(7):249.

Unterscheider J, O'Byrne J, Foran A, Robinson I, Ryan S, Devaney D, Gillick J, Malone F, Breathnach F. **Prenatal identification of an accessory lower limb.** *Prenat Diagn.* 2011 Dec;31(12):1203-4. doi: 10.1002 / pd.2846. Epub 2011.

Murphy JF. **Stillbirth: The other half of Perinatal Mortality.** *Ir Med J* 2012; 105; 4.

Murphy JF. **Erasures and Suspensions from the General Medical Council.** *Ir Med J* 2012; 105:36.

Murphy JF. **Doctors Today.** *Ir Med J* 2012; 105:68.

Murphy JF. W**hat is the Role and Value of Extra-Mural Activity.** *Ir Med J* 2012; 105:100.

Murphy JF. **Providing Support to Doctors Working in Intensive Care.** *Ir Med J.* 2012; 105:132.

Murphy JF. **Performance Improvement and Health Cost Reductions.** *Ir Med J* 2012; 105:164.

Murphy JF. **Babies and Mothers- Reducing Risk through Audits and Confidential Enquiries.** *Ir Med J* 2012; 105:228.

Murphy JF. **Pertussis has Re-Emerged.** *Ir Med J* 2012; 105: 260.

Murphy JF. **Glucose Homeostasis in the Intensive Care: The end of a Cycle.** *Ir Med J* 2012; 105:292.

Murphy JF. **The Doolin Lecture.** *Ir Med J* 2012; 105:324.

Kieran E, Twomey A, Molloy E, Murphy JF, O'Donnell C. **A Randomised Trial of Nasal Prong or Facial Mask for Reaspiratory Support of Preterm Infants.** *Pediatrics* 2012; 130:1176.

McCarthy L, McKenna M, Oyefesco O, Murphy JF, Molloy E. **Vitamin D Nutritional Status in Preterm Infants and Response to Supplementation.** *British Journal of Nutrition* 2012.

Murphy JF, Nicholson A, Turner G. **The Paediatric and Neonatology Clinical Programme: Review of Paediatric and Neonatology Services 2012.**

Murphy JF. **The Burden of Disease.** *Ir Med J* 2013; 106; 4.

Murphy JF. **The Non-Consultant Hospital Doctor Hours.** *Ir Med J* **2013; 106:36.**

Murphy JF. **Clinical Networks.** *Ir Med J* 2013; 106:68.

Murphy JF. **Antibiotic resistance is becoming a Mainstream Challenge to Public Health.** *Ir Med J* 2013; 106:100.

O'Donnell, C. P. F. **Turn and face the strange - ch..ch..ch..changes to neonatal resuscitation guidelines in the past decade.** *J Paediatr Child Health* 48, 735–739 (2012).

McCarthy RA, McKenna MJ, Oyefeso O, Uduma O, Murray BF, Brady JJ, Kilbane MT, Murphy JF, Twomey A, Donnell CP, Murphy NP, Molloy EJ. **Vitamin D nutritional status in preterm infants and response to supplementation.***Br J Nutr.* 2012 Nov 27:1-8. [Epub ahead of print] PMID: 23182428

Kieran EA, Twomey AR, Molloy EJ, Murphy JF, O'Donnell CP. **Randomized trial of prongs or mask for nasal continuous positive airway pressure in preterm infants.** *Pediatrics.* 2012 Nov; 130(5):e1170-6. Epub 2012 Oct 22. PMID: 23090339

Geoghegan S, Foran A, Twomey A, Murphy BP, Clarke T. **Regionalisation and neonatal networks.** *Ir Med J.* 2012 Jul-Aug; 105(7):249. No abstract available. PMID: 23008890

Barrett MJ, Donoghue V, Mooney EE, Slevin M, Persaud T, Twomey E, Ryan S, Laffan E, Twomey A. **Isolated acute non-cystic white matter injury in term infants presenting with neonatal encephalopathy.** *Arch Dis Child Fetal Neonatal Ed.* 2012 Jul 19. [Epub ahead of print] PMID: 22813533

Leaf A, Dorling J, Kempley S, McCormick K, Mannix P, Linsell L, Juszczak E, Brocklehurst P; **Abnormal Doppler Enteral Prescription Trial Collaborative Group. Early or delayed enteral feeding for preterm growth-restricted infants: a randomized trial.** *Pediatrics.* 2012 May; 129(5):e1260-8. Epub 2012 Apr 9. PMID: 22492770

Other Journal Publications

Geoghegan S, Foran A, Twomey A, Murphy BP, Clarke T. **Regionalisation and neonatal networks.** *Ir Med J.* 2012 Jul-Aug; 105(7):249. No abstract available. PMID: 23008890

Published Abstracts

EL-Khuffash A, Jain A, Dragulescu A, McNamara PJ, Mertens L. **Acute Changes in Myocardial Systolic Function in Preterm Infants Undergoing Patent Ductus Arteriosus Ligation. A Tissue Doppler and Myocardial Deformation Study.** *American Society of Echocardiograph,* Gaylord, Washington, July 2012.

Dany E Weisz, Amish Jain, Patrick J McNamara, Afif F EL-Khuffash. **Non-invasive Cardiac Output Monitoring in Neonates Using Bioreactance: A Comparison with Echocardiography.** Paediatric Academic Society Meeting. Boston, Massachusetts, May 2012; Neonatal-Fetal-Maternal Research Day, Toronto, April 2012; Hospital for Sick Children Paediatric Research Day, May 2012.

Cecilia Herbozo, Afif F EL-Khuffash, Amish Jain, Anie Lepointe, Patrick J McNamara. **Targeted Neonatal Echocardiography (TNE) in the NICU: Experience of a Tertiary Neonatal Centre in Canada.** Neonatal-Fetal-Maternal Research Day, Toronto, April 2012; Paediatric Academic Society Meeting. Boston, Massachusetts, May 2012.

Jessie van Dyk, Anie Lepointe, Jennifer Young, Afif F EL-Khuffash, Linh Ly, Elizabeth Asztalos, and Patrick J McNamara, MD1.**Preoperative Hemodynamic Instability is Associated with Poor Neurodevelopmental Outcome or Death before Discharge in Preterm Infants Undergoing Patent Ductus Arteriosus Ligation.** Neonatal-Fetal-Maternal Research Day, Toronto, April 2012; Paediatric Academic Society Meeting. Boston, Massachusetts, May 2012.

Amish Jain, Sandra L. Merklinger, Hideyuki Kato, Afif El Khuffash, Glen S. VaArsdell MD, Patrick McNamara. **Validation study of Echocardiography-Derived indices of Pulmonary Hypertension by Correlating with Right Ventricular Hypertrophy in Newborn Mice Exposed to Mild Hypoxia.** May 2012, Paediatric Academic Society Meeting. Boston, Massachusetts.

Amish Jain. Sandra L. Merklinger. Hideyuki Kato. Afif El Khuffash, Glen S. Van Arsdell, Patrick McNamara. **Longitudinal Evaluation of Pulmonary Hypertension in a Neonatal Murine Experimental Model: Feasibility of Echoardiography.** PAS 2012. May 2012, Paediatric Academic Society Meeting. Boston, Massachusetts.

Presentations at International Conferences

Boyle M. **Placental weight and volume differ in Intra-Uterine Growth Restricted Infants according to antenatal Doppler measurements.** Oral presentation at the Irish-American Pediatric Society Meeting; September 2012: Belfast, Northern Ireland.

Boyle M. **The relationship between Levene Index and grouped gestational ages in preterm infants.** Oral presentation at the Irish-American Pediatric Society Meeting; September 2012: Belfast, Northern Ireland.

Boyle M. **Ventricular Index remains static between 24 and 34 weeks.** Oral presentation at the Irish-American Pediatric Society Meeting; September 2012: Belfast, Northern Ireland.

Boyle M. **Peripherally inserted central catheter (PICC) related Superior Vena Cava Syndrome post Patent Ductus Arteriosus (PDA) ligation.** Oral presentation at the 4th Congress of the European Academy of Paediatric Societies Meeting; October 2012: Istanbul, Turkey.

Boyle M. **A report of two cases of glucocorticoid associated cardiac dysfunction in Noonan Syndrome.** Poster presentation at the 4th Congress of the European Academy of Paediatric Societies Meeting; October 2012: Istanbul, Turkey.

Boyle M. **Expansion of Ventricular Indices to Include Extremes of Prematurity.** Oral presentation at the Neonatal Society Meeting; November 2012: London, UK.

Kinlen D, Foran A. **Auditing Compliance with Oxygen Saturation Targets in Preterm Infants in NICU Irish-American Paediatric Society Meeting; September 2012:** Belfast, Northern Ireland.(Oral Presentation)

Joy E Tan1, Naomi Mc Callion1, O Mc Mahon2, J Caird2, D Crimmins2 A Foran1. Departments of 1Neonatology & 2 Neurosurgery, Children University Hospital, Dublin. **Incidence of Neural Tube Defect and basic newborn evaluation January 2009 to December 2011** Irish-American Paediatric Society Meeting; September 2012: Belfast, Northern Ireland. (Oral Presentation)

Gorman K, Walsh O, Mc Evoy M, Foran A, Clarke T, **A pilot survey of newborn referral practices and follow up in a paediatric out-patient department (POPD).** The Rotunda Hospital, Dublin Irish-American Paediatric Society Meeting; September 2012: Belfast, Northern Ireland. (Oral Presentation)

Gorman K(1), Pinnamaneni MR(1), El-Khuffash, A(1,3) Franklin O(1,2) , Foran A(1,3). **Effects of ibuprofen on cerebral and somatic regional tissue oxygenation, using NIRS in preterm infants <1500g with a significant PDA.** 1: Rotunda Maternity Hospital, Parnell Square, Dublin 12: Our Lady's Hospital for Children, Crumlin, Dublin 12; 3: Children's University Hospital, Temple Street, Dublin 1. 4th Congress of the European Academy of Paediatric Societies Meeting; October 2012: Istanbul, Turkey. (Poster Presentation)

Presentations at National Conferences

Boyle M. **MRI Brain in Twin-Twin Transfusion Syndrome Treated with Fetoscopic Laser Surgery.** Oral presentation at the 3rd Annual Neonatal Research Symposium; RCPI, Dublin: Jan 2012.

Boyle M. **Peripherally inserted central catheter (PICC) related Superior Vena Cava Syndrome post Patent Ductus Arteriosus (PDA) ligation.** Poster presentation at the Irish Paediatric Association Meeting, Belfast: April 2012.

Boyle M. **A report of two cases of glucocorticoid associated cardiac dysfunction in Noonan Syndrome.** Poster presentation at the Irish Paediatric Association Meeting, Belfast: April 2012.

Boyle M. **Functional MRI (fMRI) analysis of the Intra-Uterine Growth Restricted Infant Brain.** Oral presentation at the inaugural North Dublin Hospital Group Meeting; Rotunda Hospital, Dublin: Nov 2012.

Boyle M. **Functional MRI (fMRI) analysis of the Intra-Uterine Growth Restricted Infant Brain.** Oral presentation at the 2nd Irish Congress of Obstetrics, Gynaecology & Perinatal Medicine; Druids Glen, Wicklow: Dec 2012.

Boyle M. **Placental weight and volume differ in Intra-Uterine Growth Restricted Infants according to antenatal Doppler measurements.** Oral presentation at the 2nd Irish Congress of Obstetrics, Gynaecology & Perinatal Medicine; Druids Glen, Wicklow: Dec 2012.

Hayes B, **Neuroimaging and the Preterm Infant.** IDNIC Ireland 2012 Conference Ashling Hotel, Dublin November 15th 2012.

Hayes B. **Asphyxia in the Term Infant. NDHG Research Conference.** Rotunda Hospital 29th November 2012.

Local Presentations

K. Gorman, D. Kinlan A. Foran. **Audit of cases of persistent pulmonary hypertension of the newborn (PPHN) in a 6 month period.** Oral presentation at the Audit / Research Day June 2012 Children's University Hospital, Dublin.

Joy E Tan1, Naomi Mc Callion1, O Mc Mahon2, J Caird2, D Crimmins2 A Foran1 **Incidence of Neural Tube Defect and basic newborn evaluation January 2009 to December 2011** Children's University Hospital Research day June 2012 (Oral Presentation)

Creally M, Muprhy J. **The Medical Management of Ileostomies in Neonates.** Oral Presentation at the Audit / Research Day December 2012 at the Children's University Hospital, Dublin.

Awards / Prizes

Boyle M. Awarded First Place for best oral presentation at the 3rd Annual Neonatal Research Symposium; RCPI, Dublin: Jan 2012.

Boyle M. Awarded Second Place for oral presentation at the Children's University Hospital Annual Research Day; Temple Street, Dublin: June 2012.

Boyle M. The Children's Fund for Health Travel Grant, € 800. **Neonatal MRI Brain following Fetoscopic Laser Surgery for Twin-Twin Transfusion Syndrome: Implications for Clinical Practice.** Poster presentation at the Society for Maternal-Fetal Medicine Meeting, San Francisco, Feb 2013.

Post Graduate Medical Education Research Award winner, University of Toronto; $9,900. July 2012.

School of Graduate Studies Conference Grant, University of Toronto: **Non-invasive Cardiac Output Monitoring in Neonates Using Bioreactance: A Comparison with Echocardiography.** $500, May 2012.

Paediatric Academic Society Young Investigator's Travel Awards Program, Boston, Massachusetts, May 2012: **Non-invasive Cardiac Output Monitoring in Neonates Using Bioreactance: A Comparison with Echocardiography.** $500, May 2012.

Winner of Best Neonatal Platform Presentation at the 11th Annual Neonatal and Maternal Fetal Medicine Research Day, Toronto, April 2012.

Collaborator Wellcome Trust Strategic Award for **Multicentre national study of a seizure detection EEG algorithm for newborns.** Principal Investigator: Professor Geraldine Boylan UCC – Euro 3 million.

Nephrology

Initiated / Ongoing Research Projects

Clinico-Pathological Features of C3 Glomerulopathy and associated acquired and genetic complement abnormalities in an Irish Population.

1. Genetic and immunological testing conducted will add to understanding of C3 Glomerulopathy.
2. Adds to understanding of the Clinico-Pathological features of C3 Glomerulopathy.
3. Potential area for development of new medications to treat kidney disease.
   Investigator: Dr. Atif Awan

Peer Reviewed Publications

Quinlan C, Bates M, Sheils A, Dolan NM, Riordan M, Awan A. **Chronic hemodialysis in children weighing less than 10 kg.** *Pediatr Nephrol* 2012 Dec 2012 Online publication (ahead of print)

Published Abstracts

Teoh CW, Bates M, Sheils A, Dolan NM, Riordan M, Waldron M, Awan A. **Vitamin D Status in children undergoing haemodialysis in Ireland.** *Pediatr Nephrol* 2012; 27: 1802 (P346)

O'Shaughnessy MM, O'Regan J, Wong L, Traynor CA, Flanagan MJ, Dorman AM, Teoh CW, Medjeral-Thomas NR, Pickering MC, Cook HT, Conlon PJ. **Clinicopathological features and predictors of ourcome in C3 glomerulopathy.** *J Am Soc Nephrol* 2012; 23: 60A (FR-OR132)

Wong L, O'Shaughnessy MM, O'Regan J, Traynor CA, Flanagan MJ, Teoh CW, Medjeral-Thomas NR, Dorman AM, Pickering MC, Cook HT, Conlon PJ. **Renal transplantation outcomes in C3 glomerulopathy: The Irish Experience.** *J Am Soc Nephrol* 2012; 23: 1075A (PUB821)

Local Presentations

Teoh CW, Sheils A, Bates M, Dolan N.M, Riordan M, Waldron M, Awan A. **Vitamin D Status of Children undergoing haemodialysis in Ireland.** Oral Presentation at the Research / Audit Day December 2012 at the Children's University Hospital, Dublin.

Hensey C, Bates M, Riordan M, Waldron M, Awan A, Dolan N. **A 10 year audit of the safety, and complications of plasma exchange in the treatment of haemolytic uraemic syndrome.** Oral Presentation at the Research / Audit Day December 2012 at the Children's University Hospital, Dublin.

McEvoy M, Teoh CW, Goghlan D, Colhoun E, Ryan M, Riordan M. **Urinary tract infections in children – are we over investigating?** Oral Presentation at the Research / Audit Day December 2012 at the Children's University Hospital, Dublin.

Awards / Prizes

Teoh CW, Bates M, Dolan NM, Riordan M, Waldron M, Awan A. **Anaemia in children undergoing haemodialysis in Ireland: An audit of new practice guidelines for intravenous iron therapy** for best oral presentation at the Research / Audit Day. December 2012 at the Children's University Hospital, Dublin.

Neurology

Initiated / Ongoing Research Projects

Identifying the genetic basis of Landau Kleffner Syndrome
Identified a number of candidate genes and the complexity of LKS. It illustrates that there is a possibility that genetic heterogeneity may cause this disorder. The first international genetic study undertaken to better understand this rare epilepsy disorder.
Investigators: Dr. Sally Ann Lynch and Prof. Mary D King
Part-funded by the Children's Fund for Health PAC 09133

Genotype–Phenotype characterisation of the undiagnosed early onset epileptic encephalopathies in an Irish cohort
By improved diagnostics, the burden of invasive, low yield, expensive testing will be reduced and improved genetic counselling for families will be achieved. Specific diagnosis should lead to improved specific treatment. Genotype-phenotype correlations reveal the true spectrum of diseases. Deciphering the genomics of severe sporadic epileptic disorders is essential for future research into potential therapies.
Investigators: Prof. Mary D King and Dr. Nicholas Allen
Funded by the Children's Fund for Health RPAC-12-60

Peer Reviewed Publications

Allen NM ,O'hici B, Anderson G, Leslie C, Nestor T, Lynch SA, King MD. **Variant Late Infantile Neuronal Ceroid Lipofuscinosis (vLINCL) due to a Novel Heterozygous CLN8 Mutation and de novo 8p23.3 Deletion.** *Clin Genet* 2012;81: 602-604.

Allen, N. M., McKeon, A., O'Rourke, D. J., O'Meara, A. & King, M. D. **Excessive blinking and ataxia in a child with occult neuroblastoma and voltage-gated potassium channel antibodies.** *Pediatrics* 129, e1348–1352 (2012).

O'Rourke DJ, Twomey E, Lynch Sa , King MD. **Cortical Dysplasia associated with PTEN mutation in Bannayan Riley Ruvalcaba syndrome: A novel finding**. *Clin Dysmorphol.* 2012; 21: 91-92.

Brunklaus A, Dorris L, Ellis R, Reavey E, Lee E, Forbes G, Appleton R, Baxter P, Cross JH, Ferrie C, Hughes I, King MD, Jollands A, Livingston J, Lynch B, Phillip S, Rittey C, Scheffer IE, Williams R, & Zuberi SS. **The clinical utility of SCN1A testing for infantile onset epilepsy.** *Dev Med Child Neurol* 2012 55:154-61.

Hayes BC, Cooley S, Donnelly J, Doherty E, Grehan A, Madigan C, McGarvey C, Mulvany S, Ryan S, Gillian J, Geary MP, Matthews TG, King MD. **The placenta in infants >36 weeks gestation with neonatal encephalopathy: a case control study.** *Arch Dis Child Fetal Neonatal Ed.* 2012 Jul 12. [Epub ahead of print].

Heinzen El, Swoboda Kj, Hitomi Y, Gurrieri F, Nicole S, De Vries B, Tiziano FD, Fontaine B, Walley NM, Heavin S, Panagiotakaki E. European AHC Genetics Consortium (I.B AHC), ENRAH for SME CONSORTIUM, Fiori S, Abiusi E, Di Pietro L, Sweney MT, Newcomb TM, Viollet L, Huff C, Jorde L, Reyna SP, Murphy KJ, Shianna Kv, Gumbs CE, Little L, Silver K, Ptáček LJ, Haan J, Ferrari MD, Bye AM, Herkes GK,. Whitelaw CM, Webb D, Lynch BJ, Uldall P, King MD, Scheffer IE, Neri G, Arzimanoglou A, Van Den Maagdenberg A M, Sisodiya SM, Mikati MA, **Goldstein DB. ATP1A3 de novo mutations in alternating hemiplegia of childhood.** *Nature Genetics* 2012;44(9):1030-4.

Van Bogaert P, King MD, Paquier P, Wetzburger C, Labasse C, Dubru JM, Deonna T. **Acquired auditory agnosia in childhood and normal sleep EEG subsequently diagnosed as Landau-Kleffner syndrome: report of 3 cases.** *Dev Med Child Neurol,* November 2012 [Epub Ahead of Print].

Hayes B, Doherty E, Grehan A, Madigan C, McGarvey, C, Mulvany S, Geary MP, Matthews TJ, King MD. **Are Biochemical Investigations Useful in Term Asphyxia?** *J Neonat-Perinat Med* 2012; 5 305-310.

Peall KJ, Waite Aj, Wardle M, Kurian MA, Hedderly T, Lin JP, Smith M, Whone A, Pall H, White C, Lux A, Jardine P, Lynch B, Lumsden De, Kirov G, O'Riordan S, Samuel M, Lynch T, King MD, Chinnery PF, Warner TT, Owen MJ, Blake DJ, Morris HR. **SGCE and Myoclonus Dystonia: genetic analysis, diagnostic criteria and contiguous gene deletion.** *Brain* 2013; 136: 294-303.

Peall KJ, Waite Aj, Wardle M, Kurian MA, Hedderly T, Lin JP, Smith M, Whone A, Pall H, White C, Lux A, Jardine P, Lynch B, Lumsden De, Kirov G, O'Riordan S, Samuel M, Lynch T, King MD, Chinnery PF, Warner TT, Owen MJ, Blake DJ, Morris HR. SGCE and Myoclonus Dystonia: genetic analysis, diagnostic criteria and contiguous gene deletion. Brain 2013; 136: 294-303.
Heinzen EL, Swoboda KJ, Hitomi Y, Gurrieri F, Nicole S, de Vries B, Tiziano FD, Fontaine B, Walley NM, Heavin S, Panagiotakaki E; European Alternating Hemiplegia of Childhood (AHC) Genetics Consortium; Biobanca e Registro Clinico per l'Emiplegia Alternante (I.B.AHC) Consortium; European Network for Research on Alternating Hemiplegia (ENRAH) for Small and Medium-sized Enterpriese (SMEs) Consortium, Fiori S, Abiusi E, Di Pietro L, Sweney MT, Newcomb TM, Viollet L, Huff C, Jorde LB, Reyna SP, Murphy KJ, Shianna KV, Gumbs CE, Little L, Silver K, Ptáček LJ, Haan J, Ferrari MD, Bye AM, Herkes GK, Whitelaw CM, Webb D, Lynch BJ, Uldall P, King MD, Scheffer IE, Neri G, Arzimanoglou A, van den Maagdenberg AM, Sisodiya SM, Mikati MA, Goldstein DB. **De novo mutations in ATP1A3 cause alternating hemiplegia of childhood.** *Nat Genet.* 2012 Sep; 44(9):1030-4.

Bloetzer C, Jeannet PY, Lynch B, Newman CJ. **Sleep disorders in boys with Duchenne muscular dystrophy.** *Acta Paediatr.* 2012 Dec;101(12):1265-9

Allen NM, Lynch B, Twomey E. **Limbic encephalitis in a boy with N-methyl-D-aspartate receptor antibodies.** *J Pediatr.* 2012 Jun; 160(6):1060.

Heinzen EL, Swoboda KJ, Hitomi Y, Gurrieri F, Nicole S, de Vries B, Tiziano FD, Fontaine B, Walley NM, Heavin S, Panagiotakaki E; European Alternating Hemiplegia of Childhood (AHC) Genetics Consortium; Biobanca e Registro Clinico per l'Emiplegia Alternante (I.B.AHC) Consortium; European Network for Research on Alternating Hemiplegia (ENRAH) for Small and Medium-sized Enterpriese (SMEs) Consortium, Fiori S, Abiusi E, Di Pietro L, Sweney MT, Newcomb TM, Viollet L, Huff C, Jorde LB, Reyna SP, Murphy KJ, Shianna KV, Gumbs CE, Little L, Silver K, Ptáček LJ, Haan J, Ferrari MD, Bye AM, Herkes GK, Whitelaw CM, Webb D, Lynch BJ, Uldall P, King MD, Scheffer IE, Neri G, Arzimanoglou A, van den Maagdenberg AM, Sisodiya SM, Mikati MA, Goldstein DB. **De novo mutations in ATP1A3 cause alternating hemiplegia of childhood.** *Nat Genet.* 2012 Sep;44(9):1030-4.

Bloetzer C, Jeannet PY, Lynch B, Newman CJ. **Sleep disorders in boys with Duchenne muscular dystrophy.** *Acta Paediatr.* 2012 Dec; 101(12):1265-9.

Published Abstracts

Jeffery E, Birch R, Reavey E, Abuarafeh I, Dale R, King MD, Macleod S, Morrison C, O'Regan M, Zuberi S. **Clusters of epileptic seizures with long seizure free period characterize epilepsy associated with mutations in the Protocadherin 19(PCDH19) gene.** *Dev Med Child Neurol* 2012 ;54(Suppl 1):8-9.

Mcgann M, O'rourke Dj, Peake D, Ni Shuibhne N, King MD, Lynch B, Crowe C. **Narcolepsy in close temporal relationship to H1N1 vaccination in Irish children.** *Dev Med Child Neurol* 2012 ; 54(Suppl 1):29.

Allen NM, O'hici B, Anderson G, Nestor T, Lynch SS, King MD. **Variant late infantile neuronal ceroid lipofuscinosis due to a novel heterozygous CLN8 mutation and de novo 8p23.3 deletion.** *Dev Med Child Neurol* 2012; 54(Suppl 1): 36.

Allen NM, Treacey EP, Moran M, King MD. **'Stiff neonate' with mitochondrial DNA depletion.** *Dev Med Child Neurol* 2012; 54(Suppl 1):36.

Illingworth MA, Younis R, Hardy C, Mcdonald F, Chong WK, Sharma S, Nemeth A, Jayawant S, Kneen R, Hughes S, Meyer E, King MD, Maher ER, Kurian MA. **Novel mutations in PLA2G6-associated neurodegeneration** *PLAN. Dev Med Child Neurol* 2012;54 (Suppl 1):38.

Peall KJ, Waite AJ, Kurian MA, Hedderly T, Smith MA, Lin JP, King MD, Warner TT, Pall H, Chinnery P, Whone A, White C, Owen MJ, Blake DJ, Morris HR. **Myoclonus dystonia: a clinical and genetic description.** *Dev Med Child Neurol* 2012; 54 (Suppl 1): 77.

Peake D, Cassidy D, Mckee S, King MD, Kurian MA. **A novel TITF1 mutation in benign hereditary chorea.** *Dev Med Child Neurol* 2012; 54 (Suppl 1): 82.

Presentations at International Conferences

Peall KJ, Waite AJ, Kurian MA, Smith M, Pall H, Nestor T, King MD, Blake DJ, Owen MJ, Morris HR. **Contiguous gene deletions involving SGCE gene: A Clinical Description.** 16TH International Gongress of Parkinson's Disease and Movement Disorders. Dublin June 17th-21, 2012.

King MD. **Movement Disorders in Children.** 16th International Gongress of Parkinson's Disease and Movement Disorders. Dublin June 17th-21, 2012.

King MD. **Neurometabolic Movement Disorders in Children.** Clinical Neurometabolic Workshop, UCL, Goodenough College London, 28thNovember 2012.

Lynch Bryan, **Greatly increased rate of diagnosis of childhood narcolepsy since 2009 at an Irish Centre: population characteristics.** International Paediatric Sleep Association Congress 2012, Manchester, UK

Lynch Bryan. **Epilepsy in Angelman's Syndrome.** International Angelman's Syndrome Conference, Croke Park, Dublin, 13 / 5 / 12.

Lynch Bryan. **Seizures in the ICU: diagnosis and treatment.** International Paediatric Critical Care Conference, Convention Centre, Dublin, 27 / 9 / 12.

Presentations at National Conferences

King MD. **Diagnostic Considerations in the Child with CP: Investigations and Differential Diagnosis.** Symposium on "Shaping the Future." Central Remedial Clinic 60 years Anniversary March 1-2nd 2012 Dublin.

Mcgann M, O'rourke D, Crowe C, Ni Shuibhne N, King MD, Lynch BJ. **Narcolepsy Post-H1N1 Vaccination in Children: The Irish Experience** Irish Neurological Association Meeting Cork, May 2012.

Conroy J, Mc Gettigan P, Collins K, Parry-fielder B, Moran M, O'Mahoney O, Hanrahan D, Deonna T, Korrf C, Webb D, Ennis S, Lynch Sa, King MD. **The Genetic Basis of Landau- Kleffner Syndrome.** Neuroscience Ireland 7th Annual Conference, Royal College of Surgeons Ireland 5-6 September 2012.

Hayes B and King MD. **Birth Asphyxia is it preventable?** Inaugural Conference on Collaborative Lifespan Research, Rotunda Hospital Dublin 29th November 2012.

Local Presentations

Mc Donald, A. King, M. Lynch B. **Sudden Unexpected Death in Epilepsy.** Oral presentation at the Audit / Research Day June 2012 at the Children's University Hospital, Dublin.

Eimear Forbes, Suzanne Crowley, Dr. Amre Shahwan, Dr.Bryan Lynch, Professor Mary King. **Ketogenic Diet Effectiveness: CUH 2011.** Oral presentation at the Audit / Research Day June 2012 at the Children's University Hospital, Dublin.

Huggars D, Slevin M, Vavasseur C. **Neurodevelopment outcome of Preterm babies of the Noughties.** Oral presentation at the Audit / Research Day December 2012 at the Children's University Hospital, Dublin.

Awards / Prizes

Conroy J, Mc Gettigan P, Collins K, Parry-Fielder B, Moran M, O' Mahoney O, Webb D, Ennis S, King MD, Lynch SA. **Exome Sequencing - an approach to unlock the secrets of rare disorders.** Audit / Research Day. Children's University Hospital Temple St. 29th June 2012 Awarded joint second prize.

Neurosurgery

Initiated / Ongoing Research Projects

All Ireland Genetic Study of Neural tube defects.
This project seeks to determine any common genetic variants in the NTD defect population in Ireland. Tissue samples from myelomeningocoel closure along with parent samples are being collected and will have microassay for copy number variants.
Investigators: Mr John Caird (PI), Prof Anne Molloy (TCD), Dr Jane Leonard, Ms Tafadzwa Mandiwanza Part-funded by the Children's Fund for Health RPAC-12-61

KIAA1549-BRAF fusion in pilocytic astrocytoma: a clinicopathologic analysis.
The BRAF fusion has been shown to only be expressed in pilocytic astrocytoma and no other brain tumours therefore it is being introduced as a diagnostic marker. This study will correlate BRAF expression and clinical outcome in the hope if determining if BRAF fusion can also be viewed as a prognostic marker.
Investigators: Mr John Caird (PI), Dr Francesca Brett (Beaumont), Ms Tafadzwa Mandiwanza
Part-funded by the Children's Fund for Health RPAC-12-63

Peer Reviewed Publications

Sweeney KJ, Caird J, Sattar MT, Allcutt D, Crimmins D. **Spinal level of myelomeningocele lesion as a contributing factor in posterior fossa volume, intracranial cerebellar volume, and cerebellar ectopia.** *J Neurosurg Pediatr.* 2013 Feb; 11(2):154- 159 Epub. 2012 Nov 16.

Other Journal Publications

O'Halloran PJ, Kaliaperumal C, Crimmins D, Allcutt D, Caird J. **Chemotherapy induced CSF malabsorption in a shunted patient: Case report and review of the literature.** *BMJ case reports* 2013 Feb 8; 2013. pii: bcr2012008255. doi: 10.1136 / bcr-2012-008255.

Smyth E, Kaliaperumal C, Leonard J, Caird J. **Acute functional deterioration in a child with Cerebral palsy.** *BMJ Case Reports* 2012. doi: 10.1136 / bcr-2012-007642.

Zakaria Z, Kaliaperumal C, Caird J, Crimmins D, Allcutt D, Sattar M. **Unilateral facial palsy in an infant- an unusual presentation of Familial multiple cavernous malformation.** *BMJ Case Reports* 2012; doi: 10.1136 / bcr-2012-007508.

Presentations at International Conferences

Sweeney K, Caird J, Sattar T, Allcutt D, Crimmins D. **Spinal level of Myelomeningocele lesion is a contributing factor in Posterior Fossa Volume, Intracranial Cerebellar Volume and Cerebellar ectopia.** Oral presentation, 2012 AANS / CNS Joint Section on Pediatric Neurosurgery St Louis USA.

Sweeney K, Crimmins D, Caird J, Sattar T, Allcutt D. **Chiari II Malformation- A correlation between anatomical characteristics and clinical symptoms and outcomes.** Oral presentation, 23rd European Society of Paediatric Neurosurgery Congress.

Sweeney K, Caird J, Crimmins D, Sattar T, Allcutt D. **Paediatric Craniopharyngiomas - A Comparison of Open Transcranial surgery V's Minimally invasive surgery reflecting a change in treatment paradigm.** Oral Presentation, 23rd European Society of Paediatric Neurosurgery Congress.

Duddy JC & Caird J. **Repair of a Large Thoracolumbar Myelomeningocele with Associated Lumbar Kyphosis.** Oral presentation at Society of British Neurological Surgeons Autumn Meeting, Royal Armouries Museum, Leeds. 26 / 9 / 2012.

Mandiwanza T, Saidlear C, Caird J, Crimmins D. **Use of CT Vs Cranial Ultrasound in children under the age of one with ventriculoperitoneal shunt.** Oral Presentation, Society of British Neurological Surgeons Autumn Meeting, Royal Armouries Museum, Leeds 26 / 9 / 12.

Khalil AA, Caird J, Allcutt D, Sattar MT, Crimmins D. **Routine Cerebrospinal fluid analysis in first time ventriculoperitoneal shunt insertion.** Poster presentation at European Association of Neurosurgery Annual Meeting, Bratislava, Slovakia, 24-27 October 2012

Nolan C, Caird J, Allcutt D, Sattar MT, Crimmins D. **Rates of infection with respect to timing of myelomeningocele closure in the newborn.** Oral presentation at European society of Pediatric Neurosurgery congress, 1-5 May 2012, VU University of Amsterdam.

Presentations at National Conferences

Duddy JC, Mandiwanza T, Okafo U, Kehoe J, Allcutt D, Caird J, Crimmins D, Sattar MT. **Paediatric Head Injury: Six Months at the National Neurosurgical Centre.** Oral presentation at Irish Paediatric Association Annual Meeting, Ramada Hotel, Belfast. 20th April 2012.

Duddy JC & Caird J. **Dura, Pia and Arachnodactyly.** Irish Institute of Clinical Neuroscience Registrar's Prize in Clinical Neuroscience Meeting, National Gallery of Ireland, Dublin. 9th November 2012.

Mandiwanza T, Duddy JC, Okafo U, Kehoe J, Caird J, Crimmins D, Allcutt D, Sattar MT. **Paediatric Head Injury: Six Months at the National Neurosurgical Centre.** Oral presentation at 48th Irish Neurological Association meeting, Irish Institute of Clinical Neuroscience, Western Gateway Building, University College Cork, 11 / 5 / 12.

Zakaria Z, Kaliaperumal C, Sattar T, Caird J, Crimmins D, Allcutt D. **'Popcorns' in An Infant's Brain.** Oral presentation at Irish Institute of Clinical Neuroscience Registrar's Prize in Clinical Neuroscience Meeting, National Gallery of Ireland, Dublin 9 / 11 / 2012.

Local Presentations

Aziz M, Kaliaperumal C, Allcutt D. **Re-audit on Paediatric Head Injury referrals to Tertiary care service in Ireland.** Oral Presentation at the Research / Audit Day December 2012 at the Children's University Hospital, Dublin.

Nursing

Initiated / Ongoing Research Projects

Systematic RV Impact of Telemedicine in health outcomes in Epilepsy.
Bridge gaps between primary, secondary and tertiary services. Provide specialist care to service users in remote areas. Reduce travelling time for patients and parents.
Investigator: Sue Crowley

Fathers of Children Living With Complex needs.
Highlights the needs and perception of Fathers caring for children with complex needs.
Investigator: Deirdre Kelleher

The exploration of mothers' experiences of caring for a child who requires gastrostomy feeding.
The study is examining mothers experiences of caring for their child at home who requires gastrostomy tube feeding.
Investigator: Ms. Eithne Losty

Breaking the cycle of visibility
The primary aim of this study is to aid patient's families disclosing the diagnosis of epilepsy.
Investigator: Veronica Lambert

Systematic R / V. The benefits of Paediatric Home Care Packages.
Will give a greater overview of provision of homecare packages in Ireland & identify strengths & weaknesses.
Investigator: Simone McCarthy

Peer Reviewed Publications

Moloney S, Dowling M. **Early Intervention and management of adrenal insufficiency in children.** *Nursing Children and Young People.* 2012 Sept; 24(7):25-28.

Quinlan C, Bates M, Sheils A, Dolan NM, Riordan M, Awan A. **Chronic hemodialysis in children weighing less than 10 kg.** *Pediatr Nephrol* 2012 Dec 2012 Online publication (ahead of print)

Published Abstracts

O' Shaughnessy MM, O'Regan J, Wong L, Traynor CA, Flanagan MJ, Dorman AM, Teoh CW, Medjeral-Thomas NR, Pickering MC, Cook HT, Conlon PJ. **Clinicopathological features and predictors of ourcome in C3 glomerulopathy.** *J Am Soc Nephrol* 2012; 23: 60A (FR-OR132)

Teoh CW, Bates M, Sheils A, Dolan NM, Riordan M, Waldron M, Awan A. **Vitamin D Status in children undergoing haemodialysis in Ireland.** *Pediatr Nephrol* 2012; 27: 1802 (P346)

Wong L, O'Shaughnessy MM, O'Regan J, Traynor CA, Flanagan MJ, Teoh CW, Medjeral-Thomas NR, Dorman AM, Pickering MC, Cook HT, Conlon PJ. **Renal transplantation outcomes in C3 glomerulopathy: The Irish Experience.** *J Am Soc Nephrol* 2012; 23: 1075A (PUB821)

Presentations at International Conferences

Brangan A. Downey D. **Inadvertent Hypothermia in Paediatric Surgery.** Poster presentation at the European Operating Room Nurses Association Congress in Lisbon; 2012: Lisbon.

Greensmith L. **Nurse's knowledge of and attitudes towards fever and fever management in one Irish Children's Hospital.** Oral presentation at the 23rd International Networking for Healthcare Education Conference; 2012: Cambridge, UK.

Presentations at National Conferences

Carey J. Carroll B. **The development of a same day surgical admission unit.** Oral presentation at the 2nd Annual Nursing Research Conference; 2012; Temple St. Children's University Hospital, Dublin.

Cullinane AM. **Valuable for my future; a review of the quality of a graduate diploma in Critical Care Nursing.** Poster presentation at the Paediatric Intensive Care Society Conference. 2012: Dublin, Ireland.

Devenney D. **Ongoing improvements in glycaemic control in children and adolescents using CSII.** Oral presentation at the 2nd Annual Nursing Research Conference; 2012; Temple St. Children's University Hospital, Dublin.

Greensmith L. **Nurse's knowledge of and attitudes towards fever and fever management in one Irish Children's Hospital.** Oral presentation at the 2nd Annual Nursing Research Conference; 2012; Temple St. Children's University Hospital, Dublin.

Greensmith L. **Nurse's knowledge of and attitudes towards fever and fever management in one Irish Children's Hospital.** Oral presentation at the 13th Annual Interdisciplinary Research Conference; 2012; Trinity College, Dublin.

Lee C. **Nurses' Attitudes Towards Computerisation in a Dublin Hospital.** Oral presentation at the 2nd Annual Nursing Research Conference; 2012; Temple St. Children's University Hospital, Dublin.

O' Connell E. **Paediatric nurses extending their scope of practice: performing skin biopsies.** Oral presentation at the 2nd Annual Nursing Research Conference; 2012; Temple St. Children's University Hospital, Dublin.

O Connor C. **Balanced Score Cards.** Oral presentation at the 2nd Annual Nursing Research Conference; 2012; Temple St. Children's University Hospital, Dublin.

Somanadhan S; Larkin P; Drennan, J.,Treacy, E.(2012) **Experience of Living and Caring for a child with Mucopolysaccharidoses (MPS); Parents' perspectives.** CUH Research / Audit day, Temple Street, 29th June 2012, (Oral Presentation)

Crowley S., Nestor T., Keily S., O'Donnell D & Carey A. **Cost saving analysis of telephone enquires to the Neurology CNS Service.** Oral presentation at the 2nd Annual Nursing Research Conference; 2012; Temple St. Children's University Hospital, Dublin.

Tuffy, H. **Children's Wound assessment & Wound assessment chart.** Oral presentation at the National Children's Wound Care Conference; 2012; Dublin.

Awards / Prizes

Greensmith L. N**urse's knowledge of and attitudes towards fever and fever management in one Irish Children's Hospital.** Awarded the prize for best presentation at the 2nd Annual Nursing Research Conference; 2012 Temple St. Children's University Hospital, Dublin.

Teoh CW, Bates M, Dolan NM, Riordan M, Waldron M, Awan A. **Anaemia in children undergoing haemodialysis in Ireland:** An audit of new practice guidelines for intravenous iron therapy for best audit presentation at the Temple Street Audit Day 2012.

Nutrition and Dietetics

Initiated / Ongoing Research Projects

Chronic hemodialysis in children weighing less than 10 kg
Our cohort demonstrates that chronic HD, while not the treatment modality of choice, is a viable management option in children <10 kg where PD is not possible as a bridge to transplantation.
Investigators: Catherine Quinlan, Marie Bates, Aishling Sheils, Niamh Dolan, Michael Riordan & Atif Awan.

Home enteral feeding – A parent's perspective.
The primary aim of this research is: (i) to improve the knowledge of health care professionals working in the area of paediatric HEN in terms achieving a better understanding of the perceived challenges facing parents and carers of children on HEN post discharge; (ii) to examine dietetic support currently available for this population group in the community and therefore highlight potential discrepancies in dietetic care for paediatric patients discharged home from a university children's hospital on HEN and (iii) to examine the provision of education and support offered to parents of children on HEN by health care professionals at various time points of the child's tube placement, therefore drawing attention to any improvements that may be required in this area.
Investigators: Suzanne Boland, Fiona Ward, Dr. Owen Hensey

Energy Intakes of Children and Adolescents with Anorexia Nervosa during In-Patient Refeeding.
This study gives us insight into how to optimally feed calories to undernourished children to allow for safe weekly weight gains.
Investigator: Marianne O' Reilly (PI), Caroline Carr

Pediasure Plus Juice Clinical Trial.
This supplement has the potential to improve nutritional status in malnourished patients. It offers an alternative to milk based supplements in the paediatric population.
Investigators: Marianne O'Reilly, Fiona Ward, Dr. D. Slattery

Peer Reviewed Publications

Quinlan C, Bates M, Sheils A, Dolan N, Riordan M & Awan A. **Chronic hemodialysis in children weighing less than 10 kg.** *Paediatric Nephrology.* 2012 Dec.

Presentations at International Conferences

Clark A. **The Treatment Challenge Management of Emergency Regime at Home during Periods of Illness.** SSIEM Dietitians Meeting 2012.

McNulty J. **The Dietaty Management of GA1 – An Irish Perspective.** Oral Presentation at the Vitaflo Metabolic Meeting in Gothenberg.

Presentations at National Conferences

Boland S, Ward F. & Hensey O. **Home Enteral Feeding Review.** Presentation at the Irish Paediatric Association Study Day; 2012: Belfast, Northern Ireland.

Ophthalmology

Peer Reviewed Publications

Forman EB, O'Byrne JJ, Capra L, McElnea E, King MD. **Idiopathic intracranial hypertension associated with iron-deficiency anaemia.** *Arch Dis Child.* 2013 Feb 14 (Epub ahead of print).

Kirwan, C., Lanigan, B. & O'Keefe, M. **Vision-related quality of life assessment using the NEI-VFQ-25 in adolescents and young adults with a history of congenital cataract.** *J Pediatr Ophthalmol Strabismus* 49, 26–31 (2012).

Ryan, A., Hartnett, C., Lanigan, B. & O'Keefe, M. **Foldable iris-fixated intraocular lens implantation in children.** *Acta Ophthalmol* 90, e458–462 (2012).

Flitcroft, D. I. **The complex interactions of retinal, optical and environmental factors in myopia aetiology.** *Prog Retin Eye Res* 31, 622–660 (2012).

Presentations at National Conferences

McElnea E, Smolarck P, Flitcroft I. **Congenital myasthenic syndromes: Diagnosis and Management.** Oral Presentation at RAMI Ophthalmology Section, 2012, Dublin.

McElnea E, Smith O, Vanderspek N, Fitzsimons S, O'Murcaigh, A, Patal L. **Bilateral retinal detachment in Infancy.** Oral Presentation at RAMI Ophtahlmology Section 2012, Dublin.

Orthopaedic Surgery

Peer Reviewed Publications

Thomason K, Stephens MM. **Tarsal Coalition**. *Current Orthopaedic Practice* 2012. May / June 23:3; 229-233.

Groarke PJ, Thomason K, Stephen MM. **Simian Foot at Skeletal Maturity: Long term Case Report follow-up.** *Foot & Ankle Int.* 2012 August 33: 8675 -677.

Kennedy, M. T., Murphy, T., Murphy, M., Laffan, E. & Connolly, P. **Whole body MRI in the diagnosis of chronic recurrent multifocal osteomyelitis.** *Orthop Traumatol Surg Res* 98, 461–464 (2012).

McKenna, P., Leonard, M., Connolly, P., Boran, S. & McCormack, D. **A comparison of pediatric forearm fracture reduction between conscious sedation and general anesthesia.** *J Orthop Trauma* 26, 550–555; discussion 555–556 (2012).

Presentations at International Conferences

Stephens MM. **Flexor Hallucis Longus Tendon Transfer for Chronic Achilles Tendon Rupture.** Oral Presentation at the 7th International Congress of the Chinese Orthopaedic Association. 2012 Beijing, China.

Stephens MM. **Neuromuscular disorders of the Foot (Pes Cavus and HSMN).** Oral presentation at the 11th Oswestry Foot & Ankle Course 2012, Oswestry UK.

Stephens MM. **How Scientific are we? A Guide to Critical Review.** Oral presentation at the European Foot & Ankle Society AGM meeting 2012, Amsterdam.

Stephens MM. **Putting the best foot forward.** Oral presentation at the Annual General Meeting at the Old Oswestrian's Club, 2012, Oswestry, UK.

Stephens MM. **Flat Foot: From Childhood to Adult.** Oral Presentation at the 13th EFORT Congress 2012, Berlin, Germany.

Stephens MM. **Flat Foot including Coalition.** Oral Presentation at the 14th EFAS Instructional Course 2012. Vienna, Austria.

Stephens MM. **The Dorsal Approach to Arthrodesis of the 1st MTP joint.** Oral Presentation at the French Foot & Ankle Society meeting, 2012, Lyon, France.

Presentations at National Conferences

Stephens MM. **The Overcorrected CTEV.** Oral Presentation at Challenges in Paediatric Orthopaedic Surgery, 2012. Cappagh National Orthopaedic Hospital.

Local Presentations

Michael K Dodds, Paul Connolly, Damian McCormack. **Transarticular Stabilization of the Immature Femoral Head. Assessment of a Novel Surgical Approach to the Dislocating Paediatric Hip in a Porcine Model.** Oral presentation at the Audit / Research Day June 2012 at the Children's University Hospital, Dublin.

Awards / Prizes

Stephens MM. Awarded the Annual Gold Medal at the Old Oswestrian's Club Annual Meeting 2012, Oswestry UK.

Paediatrics

Peer Reviewed Publications

Donnelly, J., Bimpeh, Y., Trace, F., Waters, A. & Nicholson, A. J. **Progress in reducing road-related deaths and injuries in Irish children.** *Ir Med J* 105, 108–110 (2012).

Krebit, I., Murphy, J., Awadalla, S. & Nicholson, A. J. **Frequent desaturations, Vomiting and poor weight gain in an 8 month old.** *Ir Med J* 105, 314–315 (2012).

Nicholson, A. J., Ramet, J., **Advocacy Working Group & European Academy of Paediatrics. Improving immunisation uptake across Europe.** *Ir Med J* 105, 280–282 (2012).

Barrett, M. J. & Macken, S. **Chronic constipation causing obstructive nephropathy in a delayed toddler.** *BMJ Case Rep* 2012, (2012).

Local Presentations

Seiza Abdelrahim and Alf Nicholson. **Professionalism of Undergraduate Medical Students in Paediatrics Rotation.** Oral Presentation at the Audit / Research Day June 2012 at the Children's Universtiy Hospital, Dublin.

Nicholson AJ, Doherty D, Bauer G, Day P, Patel R, Curtin M, McCarthy B, Murtagh C, Nugent L, Baker M, Hanlon A, Murphy E, Fitzsimons J. **The Paediatric Trigger Tool (PTT) – what have we found since its introduction to the Children's University Hospital?** Oral presentation at the Research / Audit Day December 2012 at the Children's University Hospital, Dublin.

Freyne B, Hamilton K, Shannon B, McGarvey C, Matthews TG, Nicholson AJ. **Unexpected deaths associated with sitting devices in infants and young children.** Oral presentation at the Research / Audit Day December 2012 at the Children's University Hospital, Dublin.

Moore CM, Doyle J, Nicholson AJ. **An audit of accidental ingestion presentations to CUH 2010-2011.** Oral presentation at the Research / Audit Day December 2012 at the Children's University Hospital, Dublin.

Freyne B, Doyle J, Mc Namara R, Nicholson AJ. **Epidemiology of high falls from windows in Irish Children.** Oral presentation at the Research / Audit Day December 2012 at the Children's University Hospital, Dublin.

Lyons A, Stafford A, Macken S. **Audit of Chromosomal Microarray use in the development outpatients.** Oral Presentation at the Audit / Research Day December 2012 at the Children's University Hospital, Dublin.

Awards / Prizes

Eva Forman, Mary Mc Kay, Sabine Maguire, AJ Nicholson. **Patterns of burns and scald injuries in young children - are Irish children any different?** Second Prize Awarded for oral presentation at the Audit / Research Day June 2012 at the Children's University Hospital, Dublin.

Plastic Surgery

Presentations at International Conferences

Collins AM, Healy C, McGillivary A, Earley MJ. **Finger sucking as a cause of palatal fistulae and how to reduce its incidence.** Oral Presentation by Dr. Anne Collins at the 12th International Congress on Cleft Lip / Palate and related craniofacial anomalies in Florida, USA.

Psychiatry

Peer Reviewed Publications

Frodl T., Carballedo A., Hughes M.M., Saleh K., Fagan A., Skokauskas N., McLoughlin D.M., Meaney J., O'Keane V., Connor J. (2012). **Reduced expression of glucocorticoid-inducible genes GILZ and SGK-1 and high IL-6 levels are associated with reduced hippocampal volumes in major depressive disorder.** *Transl Psychiatry.* 2012 March; 2(3): e88.

Skokauskas N., Meehan J., Sweeny E., Gallagher L. (2012). **Mental health problems in children with Prader-Willi Syndrome: literature review and an Irish controlled study,** *Canadian Journal of Child and Adolescent Psychiatry,* Aug; 21(3):194-203.

Skokauskas N. **(2012) Advancing Child & Adolescent Psychiatry and Mental Health throughout the World,** *ASEAN* Journal of Psychiatry, Vol. 13 (2), July, 120-123.

Skokauskas N., Doody B., Moran T., Fitzgerald M., Lawlor M., Gallagher L., Gill M. **(2012). PBL in Child and Adolescent Psychiatry at Trinity College Dublin,** *Academic Psychiatry* Jul; 1;36(4):335-9

Presentations at International Conferences

Anglim A, Barry M, Ackermann P, Kashif M, Moran A, O'Connell S, Conway E, & Guerin S. **Stability and Change in a Clinical Sample of Preschool Children with Autistic Spectrum Disorder.** Poster presentation at International Association for Child and Adolescent Psychiatry and Allied Professionals 20th World Congress, Paris 2012.

Presentations at National Conferences

Anglim A, Barry M, Ackermann P, Kashif M, Moran A, O'Connell S, Conway E, & Guerin S. **Stability and Change in a Clinical Sample of Preschool Children with Autistic Spectrum Disorder.** Poster presentation at Irish Paediatric Association Annual Meeting, Belfast, 2012.

Houlihan B. Paediatric Legislation. **Oral presentation at the Mental Health Act 2001 national review conference.** Dublin. 2012.

Awards / Prizes

Houlihan B. **Imago Dei and Disability: The evolution of a Theological concept and its implications for understanding Disability, with particular reference to Autism.** MA (Theology) thesis. Mater Dei Institute / DCU. Dublin. 2012

Psychology

Initiated / Ongoing Research Projects

The Impact of Executive Impairment on Self-care Needs in Children and Adolescents with Spina Bifida.
Identifying the areas of executive deficit that are associated with self-care needs is important for targeting treatment. Programs to support the development of independent self-care skills could then be designed to address the issue of these dysfunctional executive processes. These programs may ameliorate the personal and social consequences of failure to complete self-care skills and the potential significant medical complications (e.g. urinary tract infections, kidney failure, and / or skin ulcers).
Investigators: Dr. Cathy Madigan (PI), Dr. Sandra Hayes, Mr. Ivor Evans & Prof. Ian Robertson

A Longitudinal Mixed- Methods Study of the Psychosocial Experiences of Parents of Screening Detected Cases and Carriers, and Clinically Diagnosed Cases of CF.
This study will inform health care staff of the psychosocial implications of newborn screening for CF for parents in an Irish context and guide services in relation to supporting families through the newborn screening process.
Investigators: Dermot Ryan (PI), Dr. Barbara Dooley (UCD), Dr. Zai Edworthy

Growing up with CF- young people's experiences in their own words.
The study will further the knowledge base of young people's experience of living with CF, the impact of the illness on their development of sense of self and identity. It may inform service development, target research and interventions for this population.
Investigators: Dr. Zai Edworthy (PI), Clare Barrett, Fiona Healy, John O' Connor (TCD)

Reviewing IQ and Adaptive Behaviour in Children under 4 years with a diagnosis of ASD over time.
The study aims to provide information about the likely outcome over time for children diagnosed with an autism spectrum disorder. The results can be used to support and plan future service development and the possible benefits of receiving early intervention services as to promote best practice.
Investigators: Myra Barry (PI), Emma Conway, Dr. Suzanne Guerin (UCD)

Presentations at International Conferences

Anglim A, Barry M, Ackermann P, Kashif M, Moran A, O'Connell S, Conway E, & Guerin S. **Stability and Change in a Clinical Sample of Preschool Children with Autistic Spectrum Disorder.** Poster presentation at International Association for Child and Adolescent Psychiatry and Allied Professionals 20th World Congress, Paris 2012.

Presentations at National Conferences

Rushe H. **Living with a Sibling with Refractory Epilepsy-children's experience of having a sibling with epilepsy.** PSI Annual Conference, Cork November, 2012.

Anglim A, Barry M, Ackermann P, Kashif M, Moran A, O'Connell S, Conway E, & Guerin S. **Stability and Change in a Clinical Sample of Preschool Children with Autistic Spectrum Disorder.** Poster presentation at Irish Paediatric Association Annual Meeting, Belfast, 2012.

Physiotherapy

Initiated / Ongoing Research Projects

Applying inventive based approaches to Public Health Challenges.
Gain new knowledge regarding health of children in Ireland who are obese. Assist in usefulness of telemedicine.
Investigator: Ms. Grace O'Malley.

Peer Reviewed Publications

O'Malley G, Hussey J, Roche E. **A pilot study to profile the lower limb musculoskeletal health in obese children.** *Paediatric Physical Therapy,* 2012; 24(3).

O'Malley GC et al. **Establishing a hospital based obesity service. An Irish Experience.** *Obestiy Facts* 2012; 5 (SIO); 9.

O'Malley G etal, 15 **The temple Street W82GO Healthy lifestyles Programme effective in Reducing BMI SOS?** *Obesity Facts* 2012: 5 (516):223.

Presentations at International Conferences

Blake C, Smyth E, Keating L,. **A pilot study to investigate the validity and reliability of the Lumbar Multifidus activation test.** Poster presentation at the International Federation of Manual Therapy Congress 2012. Quebec City. Canada.

Presentations at National Conferences

Blake C. **Physiotherapy driving change in Paediatric Orthopaedics.** Oral Presentation at the Irish Society of Chartered Physiotherapist Conference 2012.

Radiology

Peer Reviewed Publications

Bracken J, Heaslip I, Ryan S, **Chloral hydrate sedation in radiology: a retrospective audit of reduced dose.** *Pediatr Radiol* (2012) 42:349-354.

Hayes B, Cooley S; Donnelly J; Doherty E; Grehan A; Madigan C; McGarvey C; Mulvany S; Ryan S; Gillian J; Geary MP; Matthews TJ; King MD. **The Placenta in Infants more than 36 weeks Gestation with Neonatal Encephalopathy: A Case Control Study.** *Arch Dis Child* Pub on line ahead of print 12 Jul 2012.

Barrett MJ, Donoghue V, Mooney EE, Slevin M, Persaud T, Twomey E, Ryan S; Laffan E; Twomey A. **Isolated acute non-cystic white matter injury in term infants presenting with neonatal encephalopathy.** *Arch Dis Child Fetal Neonatal Ed.* 2013 Mar; 98(2):F158-60. Epub 2012 Jul 19.

Stephens LR, Donoghue V, Gillick J. **Radiological versus clinical evidence of malrotation, a tortuous tale--10-year review.** *Eur J Pediatr Surg.* 2012 Jun; 22(3):238-42. Epub 2012 May 8.

Other Journal Publications

Walshe T, Browne AM, Ryan S. **Fatal infantile encephalopathy due to complex 1 deficiency – a mitochondrial DNA mutation disorder.** *Eurorad* 2012 Aug : Case 9584.

Caro P and Ryan S. **Normal Anatomy and congenital anomalies In Imaging of Gastrointestinal Tract.** Springer Verlag. Berlin Heidelberg 2012.

Presentations at International Conferences

Lyons K, Sorensen J, Twomey EL, Donoghue VB, O Riordan M, Ryan S. **Audit of Micturating Cystourethrograms performed over 1 year in a Children's Hospital.** ECR annual meeting. Vienna. March 2012.

Caro P, Donoghue V, Ryan S Monovari A. **Correlation between MR imaging of the brain and biochemical abnormalities of children with oxidative phosphorylation disorders.** European Society of Paediatric Radiology Annual Meeting, Athens. June 2012.

Presentations at National Conferences

Davies K, Ryan S and O'Dwyer T. **Injection of submandibular gland for drooling.** Irish Otoloryngology Society Meeting, Oct 2012.

Local Presentations

Donoghue V. **Paediatric Respiratory Disease.** Temple Street Children's University Hospital Study Day. Dublin. 10th February 2012.

Manana Pienaar, Veronica Donoghue, Eoghan Laffan, Eilish Twomey, Stephanie Ryan. **An audit of the yield of GP referrals for ultrasound in Children with UTI.** Oral presentation at the Audit / Research Day June 2012 Childrens University Hospital. Dublin.

Awards / Prizes

Boyle M, Meaney J, Tarrant A, Ryan S, Foran A. **3T Magnetic Resonance Imaging of the brain at term in growth restricted infants. A short –term surrogate outcome of infants enrolled in the Perinatal Ireland PORTO Study (StOOPS)** Awarded second prize. Children's University Hospital Temple Street Audit and Research Day 29 June 2012.

Invited Lectures

Donoghue V. Invited speaker on **"European School of Radiology"** Course in Sofia, Bulgaria on 17 / 06 / 12 and in Cluj, Romania on 26 / 10 / 2012 on the use of contrast media in Paediatric Radiology.

Donoghue V. Lecture on **"Congenital malformations of the GI tract"** on the Postgraduate course, European Society of Paediatric Radiology Meeting in Athens on 28 / 05 / 12.

Donoghue V. Lecture on **"How to write a Paediatric Radiology Case report"** at European Society of Paediatric radiology meeting, Athens 30 / 05 / 2012.

Donoghue V. Lecture on **"Chest Infections"** at the UKRC (Royal College) annual congress, Manchester on 27/06/2012.

Donoghue V. Lecture on **"The Newborn Chest"** at the Royal College of Radiologists Annual Scientific Meeting, London on 10/09 /2012.

Respiratory

Peer Reviewed Publications

Flanagan F, Glackin L, Slattery DM. **Successful treatment of idiopathic pulmonary capillaritis with intravenous cyclophosphamide.** *Pediatric Pulmonology* Epub 2012 May. *Pediatr Pulmonol.* 2013 483303-5. PMID22573417.

Books / Chapters

Slattery DM and Donoghue V. **Imaging in Paediatric Pulmonology: Cystic Fibrosis** *Chapter 15.* Springer New York, Heidelberg, London, 2012.

Published Abstracts

Langan N, Walsh A, Kelly L, Maye J, Healy Slattery DM. **Compliance with International Cystic fibrosis care guidelines in a Tertiary Referral paediatric clinic.** Irish Paediatric Association, 2012.

Walsh A, Lagan N, Kelly L, S Duignan Slattery DM, Healy F. **Correlation between respiratory pathogen colonisation and lung function in a paediatric Cystic Fibrosis population.** Irish Paediatric Association, 2012.

Walsh A, Lagan N, Z Edworthy, Kelly L, Slattery DM, Healy F. **Screening for Cystic Fibrosis related Complications in a paediatric population.** Irish Paediatric Association, 2012.

Cleveland R, Zurakowski D, Slattery D, Colin, A. **Assessing progression in cystic fibrosis: a novel approach.** Irish Paediatric Association, 2012.

Presentations at International Conferences

Shukla PJ, Twomey E., Wagener JS, Healy, Slattery DM. **Liver ultrasound as a screening tool for cystic fibrosis liver disease - can we do better?** American Thoracic Society 2012.

Presentations at National Conferences

G.Mujtaba, A.Doggett, F.Healy. **"TOSCA" Consults in CUH Temple Street Pulmonology Department, Children University Hospital Temple Street Dublin.** Presented in Audit day in Dec and also at the Irish Sleep Society meeting Carlton Hotel Feb, 2013.

Langan N, Walsh A, Kelly L, Maye J, Healy Slattery DM. **Compliance with International Cystic fibrosis care guidelines in a Tertiary Referral paediatric clinic.** - European Cystic Fibrosis Conference Dublin 2012.

Walsh A, Lagan N, Kelly L, S Duignan Slattery DM, Healy F. **Correlation between respiratory pathogen colonisation and lung function in a paediatric Cystic Fibrosis population.** European Cystic Fibrosis Conference, Dublin 2012.

Walsh A, Lagan N, Z Edworthy, Kelly L, Slattery DM, Healy F. S**creening for Cystic Fibrosis related Complications in a paediatric population.** European Cystic Fibrosis Conference Dublin 2012.

Cleveland R, Zurakowski D, Slattery D, Colin, A. **Assessing progression in cystic fibrosis: a novel approach.** European Society of Paediatric radiology (ESPR) May 2012 Athens.

Langan N, Walsh A, Kelly L, Maye J, Healy Slattery DM **Lung function and BMI in a paediatric cystic fibrosis population: how do we compare?** Irish Paediatric Association 2012.

Local Presentations

N. Lagan, A. Walsh, L. Kelly, J. Maye, F. Healy, D.M. Slattery. **Compliance with International Cystic Fibrosis Care Guidelines in a Tertiary Referral Paediatric Clinic.** Oral presentation at the Audit / Research Day June 2012 at the Children's University Hospital.

Awards / Prizes

A. Walsh, N. Lagan, L. Kelly, S. Deignan, D.M. Slattery, F. Healy. **Correlation between Respiratory Pathogen Colonisation and Lung Function in a Paediatric Cystic Fibrosis Population.** First Prize awarded for oral presentation at the Audit / Research Day June 2012 at the Children's University Hospital.

N Lagan, A Walsh, L Kelly, M O Reilly, M Morgan, F Healy, D Slattery. **Nutrition and Lung function in a tertiary Cystic Fibrosis Centre.** Seconf Prize awarded for oral presentation at the Audit / Research Day June 2012 at the Children's University Hospital.

Speech and Language Therapy

Initiated / Ongoing Research Projects

The establishment of normative data for Central Auditory Processing Disorders tests in Ireland
Central Auditory Processing Disorder (CAPD) results in children not understanding the sounds that they hear, despite normal hearing. Diagnostic tests for this condition were developed in the United States. The purpose of our study is to establish norms for these CAPD tests on typically developing Irish children, which is an essential step towards the development of services to assess, diagnose and treat people with CAPD in Ireland.
Investigators: Pauline Ackermann (PI), Martin Cromb, Louise Keogh, Gillian McAlister, Clare Salley
Funded by the Children's Fund for Health. PAC-11-58

Parent Led Articulation Therapy (PLAT).
Empowering parents to treat speech disorders in children with repaired cleft palate. 3 month project to investigate parents needs and develop protocol for detailed research project.
Investigators: Triona Sweeney (PI), Debbie Sell
Seed fund provided by Children's Fund for Health SF-2012-05
Funded by the Children's Fund for Health RPAC-13-01

Other Journal Publications

Sell D, Sweeney T, Harding-Bell A, John AK. **Letter to editor - Re: Measuring quality of life in cleft lip and palate patients: currently available patient-reported outcome measures.** *Plastic & Reconstructive Surgery.* 130(6):888e, December 2012.

Published Abstracts

Sweeney T., Curley J **Intra and inter rater relaibiloty of nasendoscopy analysis.** Oral presentation at Annual Conference of CFSBGI, Bristol UK April 2012.

Presentations at International Conferences

Anglim M, Barry M, Ackermann P, Kashif M, Moran A, O'Connell A, Conway E, Guerin S. **Stability and Change in a Clinical Sample of Pre school children with Autistic Spectrum Disorder.** Poster presentation at International Association for Child and Adolescent Psychiatry and Allied Professionals 20th World Congress; 2012; Paris.

Anglim M, Barry M, Ackermann P, Kashif M, Moran A, O'Connell A, Conway E, Guerin S. **Diagnosis, IQ, Adaptive Behaviour and Speech and Language functioning in pre school children with a diagnosis of ASD.** Poster presentation at Irish Paediatric Association Annual Meeting; 2012; Belfast.

Sweeney T., Curley J **Intra and inter rater relaibiloty of nasendoscopy analysis.** Oral presentation at Annual Conference of CFSBGI, Bristol UK April 2012.

Cordero, Baylis, Chapman, Dixon, Dobbelsteyn, Harding-Bell, Sell, Sweeney, Thurmes, Trost-Cardamone, Wilson. **The Americleft Speech Project: Training, Trial and Reliability Outcomes.** Co Author of 7 papers presented at American Cleft Palate Meeting, San Jose May 2012

Local Presentations

VPI training day for Speech & language Therapists, CUH, Nov 2012.

Triona Sweeney & Leeann Flood. **Assessment of Needs of Children with 22q11 Deletion Syndrome Living In Ireland.** Oral presentation at the Audit / Research Day June 2012 at the Children's University Hospital, Dublin.

Social Work

Initiated / Ongoing Research Projects

A qualitative exploration of the impact of intra familial sexual abuse by an adolescent on family functioning"

This research, undertaken by Ms. Joanne Morgan, B.A. (Hons.), M.A. explored the impact that intra familial sexual abuse (IFSA) by an adolescent has on family functioning, from the perspective of a parent. It was a qualitative exploration using interpretative phenomenological analysis. In the study intra familial sexual abuse (IFSA) was defined as sexual abuse perpetrated against a sibling, step-sibling, cousin, niece, nephew or other younger family member (Adler &Schultz, 1995); and is reported as the most common form of abuse by adolescents (Worling, 2001; Miranda & Corcoran, 2000).

Participants in the study were parents, who had attended the NIAP service and included nine participants, seven families in total, (both parents from two families participated). In exploring the impact intra familial sexual abuse by an adolescent has on family functioning, the major themes that emerged were fear, self-judgment, shame, pressure and trauma. In exploring these themes in more detail parents described their fear of exposure and judgment by others in relation to what had occurred, fear of external threat towards the young person who had abused and fear due to a lack of control in relation to decisions that might be made by various authorities (Gardai / HSE). Many expressed shame in relation to the abuse occurring and shame because of their perceived failure to protect their children. This feeling of shame was so powerful at times that social and familial isolation followed to help families to cope with this – "I didn't want to put my face outside the front door, as far as I was concerned everybody was there looking, everybody knew". They also acknowledged the pressure to closely supervise their child at a time when developmentally young people seek increased independence,

Most families disclosed the abuse to a select number of family members; this was done out of a necessity rather than by choice. For these parents the possibility of the abuse being further exposed, was a significant source of fear "No-one must find out about what's after happening, because we'll be ostracised" Other parents maintained the abuse secret from family and friends which led to a loss in the quality of their interpersonal relationships – "I had to keep everything to myself which wasn't easy. I couldn't go out and have a social life". This isolation was a significant life change for a number of parents who had previous involvement with their communities and social context. In many families, the parents / guardians assumed the responsibility for the abuse perpetrated by their child. As a result, their understanding was that they were to blame for their other child's victimisation.

The major themes that emerged from this research confirmed NIAP's experience of working with parents. In NIAP's opinion their responses, are normative when faced with a threatening and traumatic experience and highlight the importance of providing therapeutic support to parents / carers. In particular the study highlighted how important it is for parents / carers to attend the NIAP fortnightly support group to meet others who are coping with similar feelings and challenges and to explore together how to move on positively with their lives as it may not be appropriate or safe for them to share their experiences within their normal support network.

Published Abstracts

Howley J. **Coming of age in Ireland: exploring the internal colonisers.** Proceedings of the Prix Irene Conference, Raphael Institute; 2012: Prague.

Presentations at International Conferences

Howley J. **Coming of age in Ireland: exploring the internal colonisers.** Lecture presented at the Prix Irene conference, Raphael Institute; November 2012: Prague.

Presentations at National Conferences

Howley J (for Deliberate Self Harm Team) and Pavee Point Traveller Centre: **Developing a culturally sensitive service.** Cross-border conference of NIASW and IASW Social Workers in Mental Health, October 2012.

Howley J and O'Donoghue C. **A learning experience: The Circle of Security Programme Feb-April 2012.** Presented at National Meeting of Social Workers in Child and Adolescent Mental Health, November 2012.

Our Vision for the Future

As the planning starts for the new children's hospital, we envisage research as being at its epicentre. This is a unique opportunity to build from the base upwards and produce a world-class paediatric research facility linked to a world-class paediatric hospital. Within its walls clinical and translational research can be performed , clinical audit supported, population studies undertaken and specific databases set up to deal with the major disease entities and conditions affecting children and the future generations. Ireland has one of the highest birth rates in the EU, with a unique exposure to Down Syndrome, Spina Bifida, Cleft palate, Cystic Fibrosis and Metabolic Diseases (inborn errors of metabolism) to mention a few.

All great children's hospitals across the world e.g. Sick Kids in Toronto, Great Ormond Street, Melbourne and Boston Children's Hospital are measured by their research outcomes and productivity and their norms of treatment become those that are aspired to globally. We see no reason given this opportunity that we should not emulate them.

We need buy-in from the Universities, close cooperation with their silos of expertise and a realisation that this is our chance to get it right. By recognising the importance of research at this juncture we will improve the health of future generations, establish an extraordinary opportunity to be innovative and, working together, demonstrate that **'Today's Research is Tomorrow's Treatment'.**

Notes

Notes

The Department of Research is jointly supported by the Temple Street Children's University Hospital and the Children's Fund for Health Ltd.

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