2014 Research Update

National Centre for Inherited Metabolic Disorders

Peer reviewed publications 2014
Kopajtich R, Nicholls TJ, Rorbach J, Metodiev MD, Freisinger P, Mandel H, Vanlander A, Ghezzi D, Carrozzo R, Taylor RW, Marquard K, Murayama K, Wieland T, Schwarzmayr T, Mayr JA, Pearce SF, Powell CA, Saada A, Ohtake A, Invernizzi F, Lamantea E, Sommerville EW, Pyle A, Chinnery PF, **Crushell** E, Okazaki Y, Kohda M, Kishita Y, Tokuzawa Y, Assouline Z, Rio M, Feillet F, Mousson de Camaret B, Chretien D, Munnich A, Menten B, Sante T, Smet J, Régal L, Lorber A, Khoury A, Zeviani M, Strom TM, Meitinger T, Bertini ES, Van Coster R, Klopstock T, Rötig A, Haack TB, Minczuk M, Prokisch H. Mutations in GTPBP3 cause a mitochondrial translation defect associated with hypertrophic cardiomyopathy, lactic acidosis, and encephalopathy.Am J Hum Genet. 2014 Dec 4;95(6):708-20.
Neu A, Beyer P, Bürger-Büsing J, Danne T, Etspüler J, Heidtmann B, Holl RW, Karges B, Kiess W, **Knerr I**, Kordonouri O, Lange K, Lepler R, Marg W, Näke A, Petersen M, Podeswik A, Stachow R, von Sengbusch S, Wagner V, Ziegler R, Holterhus PM. Diagnosis, Therapy and Control of Diabetes Mellitus in Children and Adolescents. Exp Clin Endocrinol Diabetes. 2014 Jul;122(7):425-34.
Coss KP, Hawkes CP, Adamczyk B, Stöckmann H, Crushell E, Saldova R, **Knerr I**, Rubio-Gozalbo ME, Monavari AA, Rudd PM, Treacy EP. N-glycan abnormalities in children with galactosemia. J Proteome Res. 2014 Feb 7;13(2):385-94.
Coss KP, Treacy EP, Cotter EJ, **Knerr I**, Murray DW, Shin YS, Doran PP. Systemic gene dysregulation in classical Galactosaemia: Is there a central mechanism? Mol Genet Metab. 2014 Nov;113(3):177-87.
Monocarboxylate Transporter 1 Deficiency and Ketone Utilization PM. van Hasselt, S Ferdinandusse, GR. Monroe, JPN. Ruiter, M Turkenburg, MJ. Geerlings, K Duran, M Harakalova, B van der Zwaag, **AA. Monavari,** I Okur, MJ. Sharrard, M Cleary, N O'Connell, M.B., Valerie Walker, E Rubio-Gozalbo, MC. de Vries, G Visser, RHJ. Houwen, JJ. van der Smagt, NM. Verhoeven-Duif, RJA. Wanders, & G van Haaften. N Engl J Med 2014; 371:1900-1907, November 13, 2014
"A Long-term Retrospective Evaluation of Functional and Radiographic Outcomes of Pediatric Hip Surgery in Hurler Syndrome" Kennedy J, Noel J, O'Meara A , Mulhall K, **Crushell E**, Fogarty E, Kelly P.*In Press* Journal of Pediatric Orthopaedics.

Other journal publications 2014
Knerr I, Crushell E: Clinical focus on Hunter syndrome- Overview of the pathophysiology and clinical manifestations of MPS II, clinical investigations and treatment modalities. The Medical Independent. 24 Jul 2014
Knerr I, Treacy E. Hereditary metabolic disorders may be under-diagnosed. Irish Medical Times. 26 Feb 2014

Books/chapters 2014
Knerr I, Vockley J, Gibson KM. Disorders of Leucine, Isoleucine and Valine Metabolism. In: Blau N, Duran M, Gibson KM, Dionisi-Vici C. Physician's Guide to the Diagnosis, Treatment and Follow-up of Inherited Metabolic Diseases. Springer, Heidelberg/New York/London, 2014

Published abstracts 2014
Knerr I, Crushell E, Hughes J, Deverell D, Mayne PD, Monavari AA, Wala A, Naughten ER, Häberle J, Treacy EP. Maple Syrup Urine Disease (MSUD): Clinical phenotypes, genotypes and treatment outcome after four decades of newborn screening in the Republic of Ireland. J Inher Metab Dis (2014), 37, Suppl 1, S80.

A. Ventzke, J. Hoffmann, E. Crushell, A. A. Monavari, P. D. Mayne, **I. Knerr**. Dihydropteridine reductase (DHPR) deficiency in Ireland: Long-term follow-up and outcome of three patients. J Inher Metab Dis (2014), 37, Suppl 1, S64.
I. Knerr, K.P. Coss, J. Kratzsch, **E. Crushell**, A. Clark, P. P. Doran, Y. S. Shin, H. Stöckmann, P. M. Rudd, E. P. Treacy. Effects of temporary low-dose galactose supplements in children of over 5 years with Classical Galactosaemia. J Inher Metab Dis (2014), 37, Suppl 1, S101.
Deverell D, Fitzsimons PE, O Shea A, Trench C, **Knerr I**, Stapleton M, Mc Sweeney N, Mayne PD. Molybdenum Cofactor Deficiency – diagnostic pitfalls on day two of life. J Inher Metab Dis (2014), 37, Suppl 1, S171.
Neville S, Sweeney B, Lynch B, **Knerr I**, O'Halloran D,Lynch SA O'Sullivan S, **Crushell E**. Friedreich's ataxia in classical galactosaemia. J Inher Metab Dis (2014), 37, Suppl 1, S104.
Huggard D, **Crushell E, Knerr I, Monavari A**. Hyperprolinaemia – a case series from Ireland. J Inher Metab Dis (2014), 37, Suppl 1, S77.
Fitzsimons PE, Borovickova I, Macken A, Murphy AM, Treacy EP, **Monavari AA**, Mayne PD. Complete dihydropyrimidine dehydrogenase deficiency: Variable presentation in the Irish population. J Inher Metab Dis (2014), 37, Suppl 1, S77.

Presentations at International Conferences 2014
Knerr I, **Crushell E, Hughes J**, Deverell D, Mayne PD, **Monavari AA**, Wala A, Naughten ER, Häberle J, Treacy EP. Maple Syrup Urine Disease (MSUD): Clinical phenotypes, genotypes and treatment outcome after four decades of newborn screening in the Republic of Ireland. SSIEM, Innsbruck, September 2014
A. Ventzke, J. Hoffmann, E. **Crushell, A. A. Monavari**, P. D. Mayne, **I. Knerr**. Dihydropteridine reductase (DHPR) deficiency in Ireland: Long-term follow-up and outcome of three patients. SSIEM, Innsbruck, September 2014
I. Knerr, K.P. Coss, J. Kratzsch, **E. Crushell**, A. Clark, P. P. Doran, Y. S. Shin, H. Stöckmann, P. M. Rudd, E. P. Treacy. Effects of temporary low-dose galactose supplements in children of over 5 years with Classical Galactosaemia. SSIEM, Innsbruck, September 2014.
Deverell D, Fitzsimons PE, O Shea A, Trench C, **Knerr I,** Stapleton M, Mc Sweeney N, Mayne PD. Molybdenum Cofactor Deficiency – diagnostic pitfalls on day two of life. J Inher Metab Dis (2014), 37, Suppl 1, S171. SSIEM, Innsbruck, September 2014.
Neville S, Sweeney B, Lynch B, **Knerr I**, O'Halloran D,Lynch SA O'Sullivan S, **Crushell E**. Friedreich's ataxia in classical galactosaemia. SSIEM, Innsbruck, September 2014.
Huggard D, **Crushell E, Knerr I, Monavari A**. Hyperprolinaemia – a case series from Ireland. SSIEM, Innsbruck, September 2014.
Neville S, Sweeney B, Lynch B, **Knerr I**, O'Halloran D,Lynch SA O'Sullivan S, **Crushell E**. Friedreich's ataxia in classical galactosaemia. Joint Irish Paediatric Association/Irish & American Paediatric Society Annual Meeting, Cork, September 2014.
J. Casey, P. McGettigan, **E. Crushell**, D. Slattery, A. Green, S. Ennis, S.A. Lynch. Lessons learned from Next-Gen studies in a consanguineous population. Genomic Disorders 2014: The Genomics of Rare Diseases, Cambridge, London, United Kingdom, March 2014

O'Connell E. Home Infusions versus Hospital Infusions. Connect for Care Copenhagen October 2014.

Presentations at National Conferences 2014
Fumarase deficiency: Clinical spectrum and diagnostic challenges. ON Oketah, **I Knerr**, **E Crushell**, PD Mayne, **J Hughes**, **AA Monavari**. Irish Paediatric Association (IPA), Cork, September 2014
Known knows, known unknowns and unknowns unknows – the challenges of exome data analysis. J Casey, **E Crushell, J Hughes**, D Cox, B Elnazir, M White, H Dorkins, S Ennis, H Murphy, SA Lynch. Temple Street Children's University Research and Audit Day, Dublin, June 2014
Lessons learned from Next-Gen studies in a consanguineous population. Genomic Disorders 2014: The Genomics of Rare Diseases, Cambridge, London, United Kingdom, March 2014. J Casey, P McGettigan, **E Crushell**, D Slattery, A Green, S Ennis, SA Lynch
Behind trhe scenes: The hidden challenges of exome sequencing in consanguineous populations. European Society of Human Genetics Conference, Milan, Italy, June 2014. J Casey, P McGettigan, **E Crushell**, D Slattery, A Green, S Ennis, SA Lynch (Poster)
Mothers experiences of caring for their child with a gastrostomy tube at home. REACH conference DCU June 2014 **Losty E** (Oral Presentation)
Known knows, known unknowns and unknowns unknows – the challenges of exome data analysis. J Casey, **E Crushell, J Hughes**, D Cox, B Elnazir, M White, H Dorkins, S Ennis, H Murphy, SA Lynch. Our Lady's Children's Hospital Research and Audit Day, Dublin, May 2014 (Poster)

Awards/prizes 2014
Best Presentation : Neville S, Sweeney B, Lynch B, **Knerr I**, O'Halloran D,Lynch SA O'Sullivan S, **Crushell E**. Friedreich's ataxia in classical galactosaemia. Joint Irish Paediatric Association/Irish & American Paediatric Society Annual Meeting, Cork, September 2014.
Audits 2014
O'Connell E, Fletcher B & Clancy C. Enzyme Replacement Therapy: vital signs audit.

Other Activities
E Crushell: Invited Speaker at Nordic and Baltic Homocystinuria Expert meeting, Gothenburg, November 2014 **E Crushell** : Invited Speaker at Irish Mucopolysaccharidosis Society Family Day Maynooth 2014 **E Crushell** : Participated in an Educational video with Irish Mucopolysaccharidosis society on Hunter Syndrome **E Crushell:** Organised and chaired a Master class in Metabolic Disorders as one of the RCPI Master class series, June 2014