### Peer reviewed publications 2013


**Treacy EP**. Invited Review for Irish Medical Times ‘Clinical Times’. Rare Diseases in Ireland and the European context, what are the plans? March 1st edition 2013


<table>
<thead>
<tr>
<th>Author(s)</th>
<th>Title</th>
<th>Year</th>
<th>Journal/Source</th>
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<tbody>
<tr>
<td>Ina Knerr, Eileen Treacy</td>
<td>Hereditary Metabolic Disorders as prototypes of treatable Rare Disorders: An Irish Perspective in an EU Context</td>
<td>2014</td>
<td>Irish Medical Times, February</td>
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<td>Fitzgerald, M., Crushell, E. and Hickey, C.</td>
<td>'Cyclic vomiting syndrome masking a fatal metabolic disease',</td>
<td>2013</td>
<td>Eur J Pediatr, 172(5), 707-10</td>
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<tr>
<td>O’Leary OA, Hawkes CP, Cody D, Hughes JAF</td>
<td>“Atypical Wolman Disease with evidence of immune dysfunction”</td>
<td>2013</td>
<td>JIMD 2013, 36: (Supplement 2), 284</td>
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**Books/chapters 2013**

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<tr>
<th>Author(s)</th>
<th>Title</th>
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<tr>
<td>Knerr I, Gibson KM, Koch HG, Häberle J.</td>
<td>Glutaminase and Glutamine Synthetase: Biochemistry, Pharmacology and Mendelian Disorder.</td>
<td>Supplement to Chapter 85: Urea Cycle Enzymes, Scriver’s Online Metabolic and Molecular Bases of Inherited Diseases (Scriver’s OMMBID), Epub March 2013</td>
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Published abstracts 2013


Presentations at International Conferences 2013


Prof. Treacy. Co-Chair for development presentation on European Network for Galactosaemia.


Prof. Treacy. Invited speaker. 12th International Congress of Inborn Errors of Metabolism. Barcelona, Spain. September 3rd, 2013. Nutrition and Dietetics meeting. ‘Galactose and Galactosaemia, when is enough really enough?’
| **Prof. Treacy.** | Invited guest speaker: Portuguese Society of Inherited Metabolic Diseases. Postgraduate education day on Galactosaemia. The utility of old (galactose metabolites) and new IgG N-glycans and others) biomarkers in monitoring efficacy of dietetic treatment. |
| **Prof. Treacy.** | The European Galactosaemia Network (EGN): an effort towards harmonizing patient treatment (with Dr. Estela Rubio-Gozalbo). |
| **Brona Fletcher.** | Evaluation of the effectiveness of a Multidisciplinary Mucopolysaccharidosis clinic in the National Centre for Inherited Metabolic Disorders. Poster presentation, Connect for Care III, The European LSD Nurses Meeting, Brussels, 14th – 15th October 2013 |
| Atypical presentation of juvenile GM1 gangliosidosis. O’Byrne J, Forman E, Crushell E, King MD. | Poster Presentation, British Paediatric Neurology Association annual meeting, Manchester 23rd – 25th Jan 2013 |
**GM Pastores.** Lysosomal storage disease, a model for diagnosis and management of rare diseases, 5th Conference of Rare Disease Day, February 26th, 2014, Rare disease Foundation, Tehran, Iran


**GM Pastores.** Lysosomal storage disease, diagnosis and management update, 10th SPDM International Symposium, March 20th, 2014, Cascais, Portugal


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**Presentations at National Conferences 2013**

**Prof. Treacy.** Invited Panel Member. IPPOSI sponsored meeting Feb. 28th, 2013. ‘Rare Diseases without Borders’, Dublin City Hall.

**Prof. Treacy.** Member of Organising Committee and Chairperson for Children’s University Hospital, Research Day for Rare Diseases Day, ‘Clinical Research in Rare Diseases. Held on March 1st, 2013

**Prof. Treacy.** Speaker: Research on Rare Diseases at Children’s University Hospital. Oral Presentation Research Day for Rare Diseases Day, Children’s University Hospital, March 1st, 2013. Knerr I, Crushell E, Hughes J, Deverell D, Mayne PD, Monavari AA, Naughten ER, Treacy EP. Four decades of newborn screening for Maple Syrup Urine Disease in the Republic of Ireland: A retrospective cohort study of the effects of early diagnosis and longitudinal metabolic control.

**Prof. Treacy.** Invited seminar. NIBRT (National Institute for Bioprocessing and Training). ‘Galactose and Galactosaemia, the how and why’, September 18th, 2013


**Prof. Treacy.** Platform presentation: DAMC Young Investigators Award, May 22nd, St. Vincent’s University Hospital. Coss KP, Doran P, Murray DW, Cotter E, Knerr I and Treacy E. Novel genomic biomarkers implicated in Classical Galactosaemia

**Prof. Treacy.** Platform presentation: Our Lady’s Children’s Hospital, Crumlin. 3rd Annual Research and Audit Meeting, May 24th. Casey J, McGettigan P, Brosnahan D, Treacy EP, Ennis S, Lynch SA. Exome sequencing confirms diagnosis where previous diagnostic testing has been normal.


**Prof. Treacy.** SSRA Student presentation, UCD, October 2013, Poster presentation, Odelia Lam, E Crushell, I Knerr, A Monavari, J Hughes, EP Treacy. Relative prevalence of HMDs in the Irish Traveller Community.


**Jane Rice.** The management of GSD type III: using a high CHO diet, evidence and practicalities. Invited speaker, The DMIMD – Dietary management of Inherited Metabolic disorders, 12th April 2013

Coss KP, Doran PP, Cotter EJ, **Knerr I**, Murray DW, **Treacy EP**. Galactosaemia, a new understanding of the pathogenesis. Poster presentation, Medical Research Charities Group Conference, Dublin, 12th November 2013

**Knerr I.** Guest lecture "Fat-burning secrets: Clinical features of Fatty Acid Oxidation Defects" Irish Paediatric Association, Annual meeting, Dublin, 8th November 2013

**Knerr I, Crushell E, Hughes J**, Deverell D, Mayne PD, **Monavari AA**, Naughten ER, **Treacy EP**. Four decades of Newborn Screening for Maple Syrup Urine Disease (MSUD) in the Republic of Ireland: A retrospective cohort study of the effects of early diagnosis and longitudinal metabolic control. Clinical Research in Rare Disease, CUH Temple Street, 1st March 2013


Forde, M. Cotter, **A.A. Monavari, I. Knerr, J. Hughes**, B. Bourke, S. Ennis, S.A. Lynch, **E. Crushell**. Clinical phenotype associated with recessive mutations in the LARS gene. Irish Paediatric Association, Dublin, November 2013 (Best presentation prize)

J. Casey, A. Green, D. Slattery, **E. Crushell**, S. Ennis, S.A. Lynch. Rare disease gene identification and its translation. Temple Street Children’s University Hospital Clinical Research in Rare Disease Day, Dublin, March 2013

J. Kutty, **E. Crushell**, M. King, **J. Hughes**. Somatic Mosaicism in X-linked PDHA1 gene, a cause of developmental delay and epilepsy. Poster presentation, Irish Paediatric Association, Annual meeting, Dublin, 8th November 2013

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**Awards/prizes 2013**

Karen Coss, PhD, SSIEM Travel award, September 2013

Dr Jim O’Byrne, Genetics trainee: Prize for best presentation in Plenary Session, Irish Paediatric Association, Annual Meeting, Dublin November 9th


Jillian Casey, Niamh Lynam-Lennon, Paul McGettigan, Jacintha O’ Sullivan, Michael McDermott, **Ahmad Monavari, Ina Knerr, Joanne Hughes**, Billy Bourke, Sean Ennis, Sally Ann Lynch, **Ellen Crushell**. Exome sequencing implicates LARS in a multisystem disorder with infantile liver failure, recurrent hepatopathy, anaemia and epilepsy. Oral presentation at British Inherited Metabolic Disease Group annual meeting, London. Dr Casey won Best Oral presentation award for this presentation.


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**Audits 2013**

**Brona Fletcher** - MPS Clinic Evaluation Form

**Celine Stenson, Maria O’Regan & Fiona Boyle** - Audit of annual blood work in PKU patients with view to revise policy