

Output by Programs:

Peer Reviewed Publications

Congenital Malformations (29)

Darlow JM, Dobson MG, Darlay R, Molony CM, Hunziker M, Green AJ, Cordell HJ, Puri P, and Barton DE (2014) A new genome scan for primary nonsyndromic vesicoureteric reflux emphasizes high genetic heterogeneity and shows linkage and association with various genes already implicated in urinary tract development. *Mol Genet Genomic Med.* 2(1) 7-29 PMID: 24498626

Stuart HM, Roberts NA, Hilton EN, McKenzie EA, Daly SB, Hadfield KD, Rahal JS, Gardiner NJ, Tanley SW, Lewis MA, Sites E, Angle B, Alves C, Lourenço T, Rodrigues M, Calado A, Amado M, Guerreiro N, Serras I, Beetz C, Varga RE, Silay MS, Darlow JM, Dobson MG, Barton DE, Hunziker M, Puri P, Feather SA, Goodship JA, Goodship TH, Lambert HJ, Cordell HJ; the UK VUR Study Group, Saggar A, Kinali M; the 4C Study Group, Lorenz C, Moeller K, Schaefer F, Bayazit AK, Weber S, Newman WG, Woolf AS (2014) Urinary tract effects of HPSE2 mutations. *J Am Soc Nephrol Epub ahead of print* 21 Aug. PMID: 25145936

Hunziker M, Colhoun E, Puri P. Renal cortical abnormalities in siblings of index patients with vesicoureteral reflux. *Pediatrics.* 2014;133(4):e933-7. PMID:24664090

Stuart HM, Roberts NA, Hilton EN, McKenzie EA, Daly SB, Hadfield KD, Rahal JS, Gardiner NJ, Tanley SW, Lewis MA, Sites E, Angle B, Alves C, Lourenço T, Rodrigues M, Calado A, Amado M, Guerreiro N, Serras I, Beetz C, Varga RE, Silay MS, Darlow JM, Dobson MG, Barton DE, Hunziker M, Puri P, Feather SA, Goodship JA, Goodship TH, Lambert HJ, Cordell HJ; the UK VUR Study Group, Saggar A, Kinali M; the 4C Study Group, Lorenz C, Moeller K, Schaefer F, Bayazit AK, Weber S, Newman WG, Woolf AS. Urinary Tract Effects of HPSE2 Mutations. *J Am Soc Nephrol.* 2014. pii: ASN.2013090961. PMID:25145936

Hofmann A, Gosemann JH, Takahashi T, Friedmacher F, Duess JW, Puri P. Imbalance of caveolin-1 and eNOS expression in the pulmonary vasculature of experimental diaphragmatic hernia. *Birth Defects Res B Dev Reprod Toxicol.* 2014;101(4):341-6. PMID: 25078423

Friedmacher F, Fujiwara N, Hofmann AD, Takahashi H, Alvarez LA, Gosemann JH, Puri P. Prenatal retinoic acid increases lipofibroblast expression in hypoplastic

rat lungs with experimental congenital diaphragmatic hernia. *J Pediatr Surg.* 2014;49(6):876-81; discussion 881. PMID:24888826

Darlow JM, Dobson MG, Darlay R, Molony CM, Hunziker M, Green AJ, Cordell HJ, Puri P, Barton DE. A new genome scan for primary nonsyndromic vesicoureteric reflux emphasizes high genetic heterogeneity and shows linkage and association with various genes already implicated in urinary tract development. *Mol Genet Genomic Med.* 2014;2(1):7-29. PMID: 24498626

Hofmann AD, Friedmacher F, Hunziker M, Takahashi H, Duess JW, Gosemann JH, Puri P. Upregulation of serotonin-receptor-2a and serotonin transporter expression in the pulmonary vasculature of nitrofen-induced congenital diaphragmatic hernia. *J Pediatr Surg.* 2014;49(6):871-4; discussion 874-5. PMID:24888825

Kutasy B, Friedmacher F, Pes L, Paradisi F, Puri P. Increased uptake of dietary retinoids at the maternal-fetal barrier in the nitrofen model of congenital diaphragmatic hernia. *J Pediatr Surg.* 2014;49(6):866-70. PMID:24888824

Friedmacher F, Fujiwara N, Hofmann AD, Takahashi H, Gosemann JH, Puri P. Expression of Eya1 and Six1 is decreased in distal airways of rats with experimental pulmonary hypoplasia. *J Pediatr Surg.* 2014;49(2):301-4. PMID: 24528972

Friedmacher F, Hofmann AD, Takahashi T, Takahashi H, Kutasy B, Puri P. Prenatal administration of all-trans retinoic acid upregulates leptin signaling in hypoplastic rat lungs with experimental congenital diaphragmatic hernia. *Pediatr Surg Int.* 2014;30(12):1183-90. PMID:25330951

Hofmann AD, Takahashi T, Duess JW, Gosemann JH, Puri P. Increased pulmonary vascular expression of Krüppel-like factor 5 and activated survivin in experimental congenital diaphragmatic hernia. *Pediatr Surg Int.* 2014;30(12):1191-7. PMID:25323860

Takahashi T, Friedmacher F, Takahashi H, Hofmann AD, Puri P. Myogenin gene expression is not altered in the developing diaphragm of nitrofen-induced congenital diaphragmatic hernia. *Pediatr Surg Int.* 2014;30(9):901-6. PMID:25056796

Friedmacher F, Fujiwara N, Hofmann AD, Takahashi H, Gosemann JH, Puri P. Evidence for decreased lipofibroblast expression in hypoplastic rat lungs with congenital diaphragmatic hernia. *Pediatr Surg Int.* 2014;30(10):1023-9. PMID:25023943

Duess JW, Hofmann AD, Puri P. Prevalence of Hirschsprung's disease in premature infants: a systematic review. *Pediatr Surg Int*. 2014;30(8):791-5. PMID:24986057

Hofmann AD, Duess JW, Puri P. Congenital anomalies of the kidney and urinary tract (CAKUT) associated with Hirschsprung's disease: a systematic review. *Pediatr Surg Int*. 2014;30(8):757-61. PMID: 24974188

Coyle D, Friedmacher F, Puri P. The association between Hirschsprung's disease and multiple endocrine neoplasia type 2a: a systematic review. *Pediatr Surg Int*. 2014;30(8):751-6. PMID: 24972642

Kutasy B, Pes L, Friedmacher F, Paradisi F, Puri P. Nitrofen increases total retinol levels in placenta during lung morphogenesis in the nitrofen model of congenital diaphragmatic hernia. *Pediatr Surg Int*. 2014;30(10):1017-22. PMID:24972641

Takahashi H, Friedmacher F, Fujiwara N, Hofmann A, Takahashi T, Puri P. Downregulation of p300 gene expression in airway mesenchyme of nitrofen-induced hypoplastic lungs. *Pediatr Surg Int*. 2014;30(4):431-5. PMID: 24488106

Friedmacher F, Hofmann AD, Takahashi H, Takahashi T, Gosemann JH, Puri P. Disruption of THY-1 signaling in alveolar lipofibroblasts in experimentally induced congenital diaphragmatic hernia. *Pediatr Surg Int*. 2014;30(2):129-35. PMID: 24374733

Hofmann AD, Friedmacher F, Takahashi H, Hunziker M, Gosemann JH, Puri P. Decreased apelin and apelin-receptor expression in the pulmonary vasculature of nitrofen-induced congenital diaphragmatic hernia. *Pediatr Surg Int*. 2014;30(2):197-203. PMID: 24363088

Mc Laughlin D, Murphy P, Puri P. Altered Tbx1 gene expression is associated with abnormal oesophageal development in the adriamycin mouse model of oesophageal atresia/tracheo-oesophageal fistula. *Pediatr Surg Int*. 2014;30(2):143-9. PMID: 24356861

Kutasy B, Friedmacher F, Duess JW, Puri P. Prenatal administration of retinoic acid increases the trophoblastic insulin-like growth factor 2 protein expression in the nitrofen model of congenital diaphragmatic hernia. *Pediatr Surg Int*. 2014;30(2):137-42. PMID: 24352370

Beygo J, Elbracht M, de Groot K, Begemann M, Kanber D, Platzer K, Gillissen-Kaesbach G, Vierzig A, Green A, Heller R, Buiting K, Eggermann T. Novel deletions affecting the MEG3-DMR provide further

evidence for a hierarchical regulation of imprinting in 14q32. *Eur J Hum Genet*. 2015 Feb;23(2):180-8. PMID:24801763

Seoighe DM, Gadancheva V, Regan R, McDaid J, Brenner C, Ennis S, Betts DR, Eadie PA, Lynch SA. A chromosomal 5q31.1 gain involving PITX1 causes Liebenberg syndrome. *Am J Med Genet A*. 2014 Nov;164A(11):2958-60. doi: 10.1002/ajmg.a.36712. Epub 2014 Aug 14. PubMed PMID: 25124102.

Conroy J, McGettigan PA, McCreary D, Shah N, Collins K, Parry-Fielder B, Moran M, Hanrahan D, Deonna TW, Korff CM, Webb D, Ennis S, Lynch SA, King MD. Towards the identification of a genetic basis for Landau-Kleffner syndrome. *Epilepsia*. 2014 Jun;55(6):858-65. doi: 10.1111/epi.12645. Epub 2014 May 14. PubMed PMID:24828792.

Casey JP, McGettigan PA, Healy F, Hogg C, Reynolds A, Kennedy BN, Ennis S, Slattery D, Lynch SA. Unexpected genetic heterogeneity for primary ciliary dyskinesia in the Irish Traveller population. *Eur J Hum Genet*. 2015 Feb;23(2):210-7. doi: 10.1038/ejhg.2014.79. Epub 2014 May 14. PubMed PMID:24824133; PubMed Central PMCID: PMC4297907.

Allen NM, Conroy J, Shahwan A, Ennis S, Lynch B, Lynch SA, King MD. Excellent outcome with de novo 15q13.3 microdeletion causing infantile spasms--a further patient. *Am J Med Genet A*. 2014 Jul;164A(7):1863-6. doi: 10.1002/ajmg.a.36532. Epub 2014 Apr 3. PubMed PMID: 24700477.

Conroy J, McGettigan P, Murphy R, Webb D, Murphy SM, McCoy B, Albertyn C, McCreary D, McDonagh C, Walsh O, Lynch S, Ennis S. A novel locus for episodic ataxia:UBR4 the likely candidate. *Eur J Hum Genet*. 2014 Apr;22(4):505-10. doi: 10.1038/ejhg.2013.173. Epub 2013 Aug 28. PubMed PMID: 23982692; PubMed Central PMCID: PMC3953901.

Gastroenterology (4)

Bourke B. Outcomes of children after esophagogastroduodenoscopy for chronic abdominal pain. *Clin Gastroenterol Hepatol*. 2015 Feb;13(2):409. doi: 10.1016/j.cgh.2014.08.007. Epub 2014 Aug 12. No abstract available. PMID:25130939

Rowland M, Gallagher C, Gallagher CG, Laoide RÓ, Canny G, Broderick AM, Drummond J, Grealley P, Slattery D, Daly L, McElvaney NG, Bourke B. Outcome in patients with cystic fibrosis liver disease. *J Cyst Fibros*. 2015 Jan;14(1):120-6. doi: 10.1016/j.jcf.2014.05.013. Epub 2014 Jun 7. PMID:24917116

Naughton J, Duggan G, Bourke B, Clyne M. Interaction of microbes with mucus and mucins: recent developments. *Gut Microbes*. 2014 Jan-Feb;5(1):48-52. doi: 10.4161/gmic.26680. Epub 2013 Oct 2. PMID:24149677

Stenke E, Hussey S. Ulcerative colitis: management in adults, children and young people (NICE Clinical Guideline CG166). *Arch Dis Child Educ Pract Ed*. 2014 Oct;99(5):194-7. PMID:24821990

Immunology/Inflammation (54)

Gleeson E, O'Donnell JS, Hams E, Fallon PF, Preston RJ. Activated Factor X Inhibits Leukocyte Pro-inflammatory Cytokine Release upon Toll-like Receptor Stimulation. *Haematologica*, 2014 Jan;99(1):185-93. PMID: 23872307

Matino D, Lillicrap D, Astermark J, Dolan G, Kessler C, Lambert T, Makris M, O'Donnell JS, Pipe S, Santagostino E, Saint-Remy, Iorio A. Switching clotting factor concentrates: Considerations in estimating the risk of immunogenicity. *Haemophilia*, 2014: Mar;20(2):200-6. PMID: 24533949

Kinsella JA, Tobin WO, Kavanagh GF, O'Donnell JS, McGrath RT, Tierney S, Feeley TM, Egan B, O'Neill D, Collins RD, Coughlan T, Harbison JA, Doherty CP, Madhavan P, Moore DJ, O'Neill SM, Colgan MP, Saqqur M, Murphy RP, Moran N, Hamilton G, McCabe DJ. Increased endothelial activation in recently symptomatic versus asymptomatic carotid artery stenosis and in cerebral microembolic-signal-negative patient subgroups. *European Journal of Neurology*, 2014 Jul;21(7):969-e55. PMID: 24712648

Tobin WO, Kinsella JA, Kavanagh GF, O'Donnell JS, McGrath RT, Tierney S, Egan B, Feeley TM, Coughlan T, Collins DR, Harbison JA, McCabe DJH. Longitudinal assessment of von Willebrand factor antigen and von Willebrand factor propeptide in response to alteration of antiplatelet therapy after TIA or ischaemic stroke. *Journal of Neurology*, 2014: Jul;261(7):1405-12. PMID: 24781842

Kinsella JA, Tobin WO, Kavanagh GF, O'Donnell JS, McGrath RT, Tierney S, Feeley TM, Egan B, O'Neill D, Collins DR, Coughlan T, Hamilton G, McCabe DJ. Increased thrombin generation potential in symptomatic versus asymptomatic moderate or severe carotid stenosis and relationship with cerebral microemboli. *Journal of Neurology, Neurosurgery & Psychiatry*, 2014: Jul 17. pii: jnnp-2013-307556. PMID: 25033981.

Laffan M, Lester W, O'Donnell JS, Will A, Tait RC, Goodeve A, Millar CM, Keeling D. Diagnosis and management of von Willebrand disease: A United Kingdom Haemophilia Centre Doctors Organisation guideline approved by the British Committee for Standards in Haematology. *British Journal of Haematology*, 2014;4 Nov;167(4):453-65. PMID: 25113304

Oliphant CJ, Hwang YY, Walker JA, Salimi M, Wong SH, Brewer JM, Englezakis A, Barlow JL, Hams E, Scanlon ST, Ogg GS, Fallon PG, McKenzie AN. (2014) MHCII-mediated dialog between group 2 innate lymphoid cells and CD4(+) T cells potentiates type 2 immunity and promotes parasitic helminth expulsion. *Immunity*. 2014 Aug 21;41(2):283-95. PMID:25088770

Schiering C, Krausgruber T, Chomka A, Fröhlich A, Adelmann K, Wohlfert EA, Pott J, Griseri T, Bollrath J, Hegazy AN, Harrison OJ, Owens BM, Löhning M, Belkaid Y, Fallon PG, Powrie F. (2014) The alarmin IL-33 promotes regulatory T-cell function in the intestine. *Nature*. 2014 Sep 25;513(7519):564-8. PMID:25043027

Linehan E, Dombrowski Y, Snoddy R, Fallon PG, Kissenpfennig A, Fitzgerald DC. (2014) Aging impairs peritoneal but not bone marrow-derived macrophage phagocytosis. *Aging Cell*. 2014 Aug;13(4):699-708. PMID:24813244

Doyle SL, Ozaki E, Brennan K, Humphries MM, Mulfaul K, Keaney J, Kenna PF, Maminishkis A, Kiang AS, Saunders SP, Hams E, Lavelle EC, Gardiner C, Fallon PG, Adamson P, Humphries P, Campbell M. (2014) IL-18 attenuates experimental choroidal neovascularization as a potential therapy for wet age-related macular degeneration. *Sci Transl Med*. 2014 Apr 2;6(230):230ra44. PMID:24695684

Aviello G, Corr SC, Johnston DG, O'Neill LA, Fallon PG. (2014) MyD88 adaptor-like (Mal) regulates intestinal homeostasis and colitis-associated colorectal cancer in mice. *Am J Physiol Gastrointest Liver Physiol*. 2014 May 1;306(9):G769-78. PMID:24603458

Ní Gabhann J, Hams E, Smith S, Wynne C, Byrne JC, Brennan K, Spence S, Kissenpfennig A, Johnston JA, Fallon PG, Jefferies CA. (2014) Btk regulates macrophage polarization in response to lipopolysaccharide. *PLoS One*. 2014 Jan 21;9(1):e85834. PMID:24465735

Hams E, Armstrong ME, Barlow JL, Saunders SP, Schwartz C, Cooke G, Fahy RJ, Crotty TB, Hirani N, Flynn RJ, Voehringer D, McKenzie AN, Donnelly SC,

- Fallon PG. (2014) IL-25 and type 2 innate lymphoid cells induce pulmonary fibrosis. *Proc Natl Acad Sci U S A*. 2014 Jan 7;111(1):367-72. PMID:24344271
- Mroz MS, Keating N, Ward JB, Sarker R, Amu S, Aviello G, Donowitz M, Fallon PG, Keely SJ. (2014) Farnesoid X receptor agonists attenuate colonic epithelial secretory function and prevent experimental diarrhoea in vivo. *Gut*. 2014 May;63(5):808-17. PMID:23916961
- Corr SC, Palsson-McDermott EM, Grishina I, Barry SP, Aviello G, Bernard NJ, Casey PG, Ward JB, Keely SJ, Dandekar S, Fallon PG, O'Neill LA. (2014) MyD88 adaptor-like (Mal) functions in the epithelial barrier and contributes to intestinal integrity via protein kinase C. *Mucosal Immunol*. 2014 Jan;7(1):57-67. PMID:23612054
- Aviello G, Amu S, Saunders SP, Fallon PG. (2014) A mineral extract from red algae ameliorates chronic spontaneous colitis in IL-10 deficient mice in a mouse strain dependent manner. *Phytother Res*. 2014 Feb;28(2):300-4. PMID:23554071
- Fernandes P, O'Donnell C, Lyons C, Keane J, Regan T, O'Brien S, Fallon P, Brint E, Houston A. (2014) Intestinal expression of Fas and Fas ligand is upregulated by bacterial signaling through TLR4 and TLR5, with activation of Fas modulating intestinal TLR-mediated inflammation. *J Immunol*. 2014 Dec 15;193(12):6103-13. PMID:25378591
- Hussain S, Berki DM, Choon SE, Burden AD, Allen MH, Arostegui JI, Chaves A, Duckworth M, Irvine AD, Mockenhaupt M, Navarini AA, Seyger MM, Soler-Palacin P, Prins C, Valeyrie-Allanore L, Vicente MA, Trembath RC, Smith CH, Barker JN, Capon F. (2014) IL36RN mutations define a severe autoinflammatory phenotype of generalized pustular psoriasis. *Journal of Allergy and Clinical Immunology*. 2014 Nov 12. pii: S0091-6749(14)01429-8. PMID:25458002
- Irvine AD. (2014) Commentary: Methotrexate and ciclosporin in the treatment of severe eczema in children. *British Journal of Dermatology*. 2014 Mar;170(3):499-500. doi: 10.1111/bjd.12821. PMID:24617433
- Irvine AD. (2014) Crossing barriers; restoring barriers? Filaggrin protein replacement takes a bow. *Journal of Investigative Dermatology*. 2014 Feb;134(2):313-4. PMID:24424455
- O'Donovan SM, Murray DM, Hourihane JO, Kenny LC, Irvine AD, Kiely M. (2014) Cohort profile: The Cork BASELINE Birth Cohort Study: Babies after SCOPE: Evaluating the Longitudinal Impact on Neurological and Nutritional Endpoints. *Int J Epidemiol*. 2014 Aug 7. PMID:25102856
- Kiely C, Devaney D, Fischer J, Lenane P, Irvine AD. (2014) Ichthyosis prematurity syndrome: a case report and review of known mutations. *Pediatr Dermatol*. 2014 Jul-Aug;31(4):517-8. PMID:24889544
- Cole C, Kroboth K, Schurch NJ, Sandilands A, Sherstnev A, O'Regan GM, Watson RM, McLean WH, Barton GJ, Irvine AD, Brown SJ. (2014) Filaggrin-stratified transcriptomic analysis of pediatric skin identifies mechanistic pathways in patients with atopic dermatitis. *J Allergy Clin Immunol*. 2014 Jul;134(1):82-91. PMID:24880632
- Nason GJ, Baker JF, Seoighe D, Irvine AD, McDermott M, Orr D, Capra M, Kelly PM. (2014) Congenital-infantile fibrosarcoma of the foot--avoidance of amputation. *Ir Med J*. 2014 May;107(5):148-9. PMID:24908860
- Courtney DG, Atkinson SD, Allen EH, Moore JE, Walsh CP, Pedrioli DM, MacEwen CJ, Pellegrini G, Maurizi E, Serafini C, Fantacci M, Liao H, Irvine AD, McLean WH, Moore CB. (2014) siRNA silencing of the mutant keratin 12 allele in corneal limbal epithelial cells grown from patients with Meesmann's epithelial corneal dystrophy. *Invest Ophthalmol Vis Sci*. 2014 May 6;55(5):3352-60. PMID:24801514
- Salinas CF, Irvine AD, Itin PH, Di Giovanna JJ, Schneider H, Clarke AJ, McGovern LS, Fete M. (2014) Second International Conference on a classification of ectodermal dysplasias: development of a multi-axis model. *Am J Med Genet A*. 2014 Oct;164A(10):2482-9. PMID:24700551
- Hughes R, McAleer M, Watson R, Collins S, Irvine A, White M. (2014) Rapidly involuting congenital hemangioma with pustules: two cases. *Pediatr Dermatol*. 2014 May-Jun;31(3):398-400. PMID:24689686
- Thawer-Esmail F, Jakasa I, Todd G, Wen Y, Brown SJ, Kroboth K, Campbell LE, O'Regan GM, McLean WH, Irvine AD, Kezic S, Sandilands A. (2014) South African amaXhosa patients with atopic dermatitis have decreased levels of filaggrin breakdown products but no loss-of-function mutations in filaggrin. *J Allergy Clin Immunol*. 2014 Jan;133(1):280-2.e1-2. PMID:24369804
- Foley C, Moran B, McMenamin M, McDermott R, Ormond P, Irvine AD. (2014) Spontaneous regression of cutaneous metastases of squamous cell carcinoma. *QJM*. 2014 Jan;107(1):61-3. PMID:23904518

- Conte MI, Pescatore A, Paciolla M, Esposito E, Miano MG, Lioi MB, McAleer MA, Giardino G, Pignata C, Irvine AD, Scheuerle AE, Royer G, Hadj-Rabia S, Bodemer C, Bonnefont JP, Munnich A, Smahi A, Steffann J, Fusco F, Ursini MV. (2014) Insight into IKBKG/NEMO locus: report of new mutations and complex genomic rearrangements leading to incontinentia pigmenti disease. *Hum Mutat.* 2014 Feb;35(2):165-77. PMID:24339369
- Linnane B, Vaish S, Clarke D, O'Sullivan N, McNally P. The findings of a clinical surveillance bronchoalveolar lavage programme in pre-school patients with cystic fibrosis. *Pediatr Pulmonol.* 2014 Nov 18. doi: 10.1002/ppul.23118. [Epub ahead of print] PubMed PMID: 25408378.
- Weldon S, McNally P, McAuley DF, Oglesby IK, Wohlford-Lenane CL, Bartlett JA, Scott CJ, McElvaney NG, Greene CM, McCray PB Jr, Taggart CC. miR-31 dysregulation in cystic fibrosis airways contributes to increased pulmonary cathepsin S production. *Am J Respir Crit Care Med.* 2014 Jul 15;190(2):165-74. doi: 10.1164/rccm.201311-1986OC. PubMed PMID: 24940638; PubMed Central PMCID: PMC4226050.
- O'Donovan C, Greally P, Canny G, McNally P, Hussey J. Active video games as an exercise tool for children with cystic fibrosis. *J Cyst Fibros.* 2014 May;13(3):341-6. doi: 10.1016/j.jcf.2013.10.008. Epub 2013 Nov 1. PubMed PMID: 24189057.
- Ringholz F, Devins M, McNally P. Managing end stage lung disease in children. *Paediatr Respir Rev.* 2014 Mar;15(1):75-80; quiz 80-1. doi: 10.1016/j.prrv.2013.07.002. Epub 2013 Aug 13. PubMed PMID: 23948571
- Nieto A, Wahn U, Bufe A, Eigenmann P, Halken S, Hedlin G, Høst A, Hourihane J, Just J, Lack G, Lau S, Matricardi PM, Muraro A, Papadopoulos N, Roberts G, Simpson A, Valovirta E, Weidinger S, Wickman M, Mazon A. Allergy and asthma prevention 2014. *Pediatr Allergy Immunol* 2014; 25: 516–533. DOI: 10.1111/pai.12272 PMID:25224992
- Gallagher A, Hourihane JO'B, Kenny LC, Irvine AD, Khashan AS. A longitudinal study of skin barrier function in pregnancy and the postnatal period. *Obstetric Medicine*, December 2014; 7: (4) 156-159. doi 10.1177/1753495X14547428
- Worm M, Moneret-Vautrin A, Scherer K, Lang R, Fernandez-Rivas M, Cardona V, Kowalski ML, Jutel M, Poziomkowska-Gesicka I, Papadopoulos NG, Beyer K, Mustakov T, Christoff G, Bilò MB, Muraro A, Hourihane JO, Grabenhenrich LB. First European data from the network of severe allergic reactions (NORA). *Allergy.* 2014 Oct;69(10):1397-404. doi: 10.1111/all.12475. Epub 2014 Aug 16. PMID: 24989080
- L Soller, JO'B Hourihane, A Dunngalvin. The impact of oral food challenge tests on food allergy health-related quality of life. *Allergy* 2014;69:1255–1257 PMID:24925125
- Muraro A, Dubois AEJ, DunnGalvin A, Hourihane JO'B, de Jong NW, Meyer R, Panesar SS, Roberts G, Salvilla S, Sheikh A, Worth A, Flokstra-de Blok BMJ. EAACI Food Allergy and Anaphylaxis Guidelines. Food allergy health-related quality of life measures. *Allergy* 2014; DOI:10.1111/all.12405. PMID:24785644
- N. Twomey, A. Temko, J. O'B. Hourihane, and W. P. Marnane. Automated detection of perturbed cardiac physiology during oral food allergen challenge in children *IEEE Journal of Biomedical and Health Informatics* 2014 18(3) 1051-1057. DOI 10.1109/JBHI.2013.2290706 PMID:24240032
- R. Crevel, J. Baumert, S. Luccioli, A. Baka, S. Hattersley, J.O'B Hourihane, S. Ronsmans, F. Timmermans, R. Ward, Y.J. Chung. Translating reference doses into allergen management practice: challenges for stakeholders *Food and Chemical Toxicology*, January 2014 <http://dx.doi.org/10.1016/j.fct.2014.01.033> PMID:24491260
- Al-Alawi M, Buchanan P, Verriere V, Higgins G, McCabe O, Costello RW, McNally P, Urbach V, Harvey BJ. (2014) Physiological levels of lipoxin A4 inhibit ENaC and restore airway surface liquid height in cystic fibrosis bronchial epithelium. *Physiol Rep.* 7;2(8). PMID: 25107986
- Higgins G, Buchanan P, Perriere M, Al-Alawi M, Costello R, Verriere V, McNally P, Harvey BJ and Urbach V. (2014) Activation of P2RY11 and ATP release by LXA4 restores the airway surface liquid layer and epithelial repair in cystic fibrosis. *Am J Respir Cell Mol Biol.* 51(2):178-90. PMID: 24588705
- Ringholz FC, Buchanan PJ, Clarke DT, Millar RG, McDermott M, Linnane B, Harvey BJ, McNally P and Urbach V. (2014) Reduced 15-LO2 and Lipoxin A4/Leukotriene B4 ratio in children with Cystic Fibrosis. *European Respiratory Journal.* Eur Respir J. 2014 Aug;44(2):394-404. PMID: 24696116

Higgins G, Ringholz F, Buchanan P, McNally P and Urbach V. (2014) Physiological Impact of Abnormal Lipoxin A4 Production on Cystic Fibrosis Airway Epithelium and Therapeutic Potential. Hindawi Publishing Corporation BioMed Research International. ID: 781087 (review article)

Hendrick SM, Mroz MS, Greene CM, Keely SJ, Harvey BJ (2014). Bile acids stimulate chloride secretion through CFTR and calcium-activated Cl⁻ channels in Calu-3 airway epithelial cells. *Am J Physiol Lung Cell Mol Physiol*. 2014 Sep 1;307(5):L407-18. doi: 10.1152/ajplung.00352.2013. Epub 2014 Jul 3. PMID: 24993131

Saint-Criq V, Harvey BJ (2014). Estrogen and the cystic fibrosis gender gap. *Steroids*. 2014 Mar; 81C:4-8. doi: 10.1016/j.steroids.2013.11.023. Epub 2013 Dec 14. Review. PMID:24342234

Ozaki, O., Campbell M., Doyle, S.L. (2015) Targeting the NLRP3 inflammasome in chronic inflammatory diseases: current perspectives. *Journal of Inflammation Research*. 16;8 15-27 PMID: 25653548

Stack J, Doyle SL, Connolly DJ, Reinert LS, O'Keeffe KM, McLoughlin RM, Paludan SR, Bowie AG. (2014) TRAM Is Required for TLR2 Endosomal Signaling to Type I IFN Induction. *Journal of Immunology*, 193(12):6090-102. PMID: 25385819

Yuan H, Wilson CM, Xia J, Doyle S. L., Li S, Fales AM, Liu Y, Ozaki E, Mulfaul K, Hanna G, Palmer GM, Wang LV, Grant GA, Vo-Dinh T. (2014) Plasmonics-enhanced and optically modulated delivery of gold nanostars into brain tumor. *Nanoscale* 6(8):4078-82 PMID: 24619405

Doyle SL, Ozaki E, Brennan K, Humphries MM, Mulfaul K, Keaney J, Kenna PF, Maminishkis A, Kiang AS, Saunders SP, Hams E, Lavelle EC, Gardiner C, Fallon PG, Adamson P, Humphries P, Campbell M. (2014) IL-18 attenuates experimental CNV as a potential therapy for wet AMD. *Science Translational Medicine* April 2; 6, 230ra44 PMID: 24695684

Vencken SF, Greene CM, McKiernan PJ. (2015) Non-coding RNA as lung disease biomarkers. *Thorax*. [Epub ahead of print] 2014 Dec 30. pii: thoraxjnl-2014-206193. doi:10.1136/thoraxjnl-2014-206193. PMID: 25550385

Julie Renwick, Paul McNally, Bettina John, Todd DeSantis, Barry Linnane, and Philip Murphy. 2014. The Microbial Community of The Cystic Fibrosis Airway is Disrupted in Early Life. *PLoS ONE*. 9(12): e109798. PMID: PMC4272276

Eliwan HO, Watson RW, Aslam S, Regan I, Philbin B, O'Hare FM, O'Neill A, Preston R, Blanco A, Grant T, Nolan B, Smith O, Molloy EJ. Neonatal brain injury and systemic inflammation: modulation by activated protein C ex vivo. *Clin Exp Immunol*. 2015 Mar;179(3):477-84. doi: 10.1111/cei.12453. PubMed PMID: 25204207

Obesity & Diabetes (29)

PC Scully, O Reid, AP Macken, W Cullen, D Leddin, C Dunne, CS O'Gorman. Food and beverage cues in United Kingdom and Irish children-television programming. *Archives of Diseases in Childhood*, May 2014. Epublished ahead of print July 2014 . PMID: 24982416

AP Macken, E Sasaki, A Quinn, W Cullen, D Leddin, C Dunne, CS O'Gorman. Paediatric Diabetes: Information-seeking behavior of families. *Irish Medical Journal* March 2014; 2014: 107(3):87. PMID:24757896

P Scully, AP Macken, D Leddin, W Cullen, C Dunne, CS O'Gorman. Food and beverage advertising during children's television programming. *Irish Journal of Medical Sciences*, April 2014. PMID: 24982416

Hawkes CP, Levine MA. Ketotic hypercalcemia: a case series and description of a novel entity (2014). *J Clin Endocrinol Metab*. 99(5):1531-6. PMID 24606099

Hawkes CP, Grimberg A. Measuring growth hormone and insulin-like growth factor-I in infants: what is normal? (2013) *Pediatric endocrinology reviews* : PER. 11(2):126-46. PMID 24575549

BC. Ip, AE. Hogan, BS. Nikolajczyk. Lymphocyte roles in metabolic dysfunction: of men and mice. *Trends in Endocrinology & Metabolism* 2015. PMID: 25573740

AE. Hogan, G. Gaoatswe, L. Lynch, MA. Corrigan, C. Woods, J. O'Connell, D. O'Shea. Glucagon-like peptide 1 analogue therapy directly modulates innate immune-mediated inflammation in individuals with type 2 diabetes mellitus. *Diabetologia* 2014. PMID: 24362727

McDarby V, Hevey D, Cody D (2015) An Overview of the Role of Social Network Sites in the Treatment of Adolescent Diabetes. *Diabetes Technol Ther*. 2015 Feb 5. [Epub ahead of print] PMID: 25654161

Delamater AM, de Wit M, McDarby V, Malik J and Acerini CL (2014) Psychological care of children and adolescents with type 1 diabetes. *Pediatric Diabetes* Sep 2014, Suppl 20:232-44 PMID: 25182317

Hawkes C, McDarby V and Cody C (2014) Fear of Hypoglycemia in parents of children with type 1 diabetes. *Journal of Paediatric Child Health* Aug 2014 50(8): 639-42 PMID: 24953343

Carolan E, Hogan AE, Corrigan M, Gaotswe G, O'Connell J, Foley N, O'Neill LA, Cody D, O'Shea D (2014) The impact of childhood obesity on inflammation, innate immune cell frequency, and metabolic microRNA expression. *J Clin Endocrinol Metab.* 2014 Mar;99(3) PMID: 24423308

Bath L, Cody D, Krogvold L, Sundberg F, Bangstad HJ. Annual Conference 2013 highlights. *Pediatr Diabetes.* 2014 May;15(3):257-9. doi: 10.1111/pedi.12145. Epub 2014 Apr 23 PMID: 24754431

McMorrow AM, Connaughton RM, Lithander FE, Roche HM. Adipose tissue dysregulation and metabolic consequences in childhood and adolescent obesity: potential impact of dietary fat quality. *Proc Nutr Soc.* Epub 2014 Dec 11 PMID: 25497038 [PubMed - in process]

Costello LA, Lithander FE, Gruen RL, Williams LT. Nutrition therapy in the optimisation of health outcomes in adult patients with moderate to severe traumatic brain injury: findings from a scoping review (2014) *Injury.* 2014 Dec;45(12):1834-41. PMID: 24996574

Lynam-Lennon N, Connaughton R, Carr E, Mongan AM, O'Farrell NJ, Porter RK, Brennan L, Pidgeon GP, Lysaght J, Reynolds JV, O'Sullivan J. Excess visceral adiposity induces alterations in mitochondrial function and energy metabolism in esophageal adenocarcinoma. *BMC Cancer.* 2014 Dec 3;14:907. PMID: 25471892 [PubMed - in process]

Finucane OM, Reynolds CM, McGillicuddy FC, Harford KA, Morrison M, Baugh J, Roche HM. Macrophage migration inhibitory factor deficiency ameliorates high-fat diet induced insulin resistance in mice with reduced adipose inflammation and hepatic steatosis (2014) *PLoS One.* 2014 Nov 20;9(11):e113369. PMID: 25412423

Jimenez-Gomez Y, Cruz-Teno C, Rangel-Zuñiga OA, Peinado JR, Perez-Martinez P, Delgado-Lista J, Garcia-Rios A, Camargo A, Vazquez-Martinez R, Ortega-Bellido M, Perez-Jimenez F, Roche HM, Malagon MM, Lopez-Miranda J. Effect of dietary fat modification on subcutaneous white adipose tissue insulin sensitivity in patients with metabolic syndrome (2014) *Mol Nutr Food Res.* 2014 Nov;58(11):2177-88. PMID: 25044988

Camargo A, Peña-Orihuela P, Rangel-Zuñiga OA, Pérez-Martínez P, Delgado-Lista J, Cruz-Teno C, Marín C, Tinahones F, Malagón MM, Roche HM, Pérez-Jiménez F, López-Miranda J. Peripheral blood mononuclear cells as in vivo model for dietary intervention induced systemic oxidative stress (2014) *Food Chem Toxicol.* 2014 Oct;72:178-86. PMID: 25057809

O'Gorman A, Morris C, Ryan M, O'Grada CM, Roche HM, Gibney ER, Gibney MJ, Brennan L. Habitual dietary intake impacts on the lipidomic profile (2014) *J Chromatogr B Analyt Technol Biomed Life Sci.* 2014 Sep 1;966:140-6. PMID: 24565891

Afman L, Milenkovic D, Roche HM. Nutritional aspects of metabolic inflammation in relation to health-insights from transcriptomic biomarkers in PBMC of fatty acids and polyphenols (2014) *Mol Nutr Food Res.* 2014 Aug;58(8):1708-20. PMID: 24449395

Camargo A, Meneses ME, Pérez-Martínez P, Delgado-Lista J, Rangel-Zuñiga OA, Marín C, Almadén Y, Yubero-Serrano EM, González-Guardia L, Fuentes F, Tinahones FJ, Roche HM, Malagón MM, Pérez-Jiménez F, López-Miranda J.

Dietary fat modifies lipid metabolism in the adipose tissue of metabolic syndrome patients (2014) *Genes Nutr.* 2014 Jul;9(4):409. PMID: 24895107

Draper E, DeCoursey J, Higgins SC, Canavan M, McEvoy F, Lynch M, Keogh B, Reynolds C, Roche HM, Mills KH, Loscher CE. Conjugated linoleic acid suppresses dendritic cell activation and subsequent Th17 responses (2014) *J Nutr Biochem.* 2014 Jul;25(7):741-9. PMID: 24794016

Tynan GA, Hearnden CH, Oleszycka E, Lyons CL, Coutts G, O'Connell J, Corrigan MA, Lynch L, Campbell M, Callanan JJ, Mok KH, Geoghegan J, O'Farrelly C, Allan SM, Roche HM, O'Shea DB, Lavelle EC. Endogenous oils derived from human adipocytes are potent adjuvants that promote IL-1 -dependent inflammation (2014) *Diabetes.* 2014 Jun;63(6):2037-50. PMID: 24458363

Wallace M, Morris C, O'Grada CM, Ryan M, Dillon ET, Coleman E, Gibney ER, Gibney MJ, Roche HM, Brennan L. Relationship between the lipidome, inflammatory markers and insulin resistance (2014) *Mol Biosyst.* 2014 Jun;10(6):1586-95. PMID: 24714806

O'Grada CM, Morine MJ, Morris C, Ryan M, Dillon ET, Walsh M, Gibney ER, Brennan L, Gibney MJ, Roche HM. PBMCs reflect the immune component of the

WAT transcriptome--implications as biomarkers of metabolic health in the postprandial state (2014) *Mol Nutr Food Res.* 2014 Apr;58(4):808-20. PMID: 24170299

McAllan L, Skuse P, Cotter PD, O'Connor P, Cryan JF, Ross RP, Fitzgerald G, Roche HM, Nilaweera KN. Protein quality and the protein to carbohydrate ratio within a high fat diet influences energy balance and the gut microbiota in C57BL/6J mice (2014) *PLoS One.* 2014 Feb 10;9(2):e88904. PMID: 24520424

Cadogan SL, Keane E, and Kearney PM. (2014) The effects of individual, family and environmental factors on physical activity levels in children: a cross-sectional study. *BMC Pediatrics.*14(1):107 PMID: 24745362

Keane E, Kearney PM, Perry IJ, Browne GM, and Harrington JM. (2014) Diet, Physical Activity, Lifestyle Behaviors, and Prevalence of Childhood Obesity in Irish Children: The Cork Children's Lifestyle Study Protocol. *JMIR Res Protoc.* 3(3):e44 PMID: 25138582

Keane E, Kearney PM, Perry IJ, Kelleher CC, and Harrington JM. (2014) Trends and prevalence of overweight and obesity in primary school aged children in the Republic of Ireland from 2002-2012: a systematic review. *BMC Public Health.* 14:974 PMID: 25312652

O'Donovan S.M., Y. J Zhang, Kinsella M., Murray D.M., Hourihane J.O'B., Kenny L.C., Irvine A.D. and Kiely M (2014) The C3-epimer of 25-hydroxyvitamin D3 [3-*epi*-25OHD3] is quantifiable in almost all neonates and mirrors variation in serum 25OHD3. *Nutrients*, 6: 2759-2919. doi:10.3390/nu6072759

Paediatric Cancer (16)

Bryan K, Terrile M, Bray IM, Domingo-Fernandez R, Watters KM, Koster J, Versteeg R, and Stallings RL (2014) Discovery of miRNA-mRNA Functional Modules within Integrated Data using Biclust Analysis. *Nucleic Acids Research* 42:e17. PMID:24357407

Rihani A, Van Maerken T, De Wilde B, Zeka F, Laureys G, Norga K, Tonini GP, Coco S, Versteeg R, Noguera R, Schulte JH, Eggert A, Stallings RL, Speleman F, and Vandesompele J (2014) Lack of association between MDM2 promoter SNP309 and clinical outcome in neuroblastoma patients. *Pediatric Blood and Cancer* 61:1867-1870. PMID:24391119

Planells-Ferrer L, Moubarak RS, Soriano A, Murphy D, Ferreres JC, Borrás F, Gallego S, Stallings RL, Segura MF and Cornella JX (2014) MYCN repression

of Lifeguard/FAIM2 is associated with increased cell migration and neuroblastoma poor prognosis. *Cell Death and Disease* 5e:1401. PMID:25188511

Harvey H, Piskareva O, Creevey L, Alcock LC, Buckley PG, O'Sullivan M, Segura MF, Gallego S, Stallings RL, Bray IM (2014) Modulation of Chemotherapeutic Drug Resistance in Neuroblastoma SK-N-AS Cells by the Neural Apoptosis Inhibitory Protein and miR-520f. *Inter. J. Cancer* 136:1579-1588 PMID:25137037

Lee S, Rahnenführer J, Lang M, de Preter K, Mestdagh P, Koster J, Versteeg R, Stallings RL, Varesio L, Asgharzadeh S, Schulte J, Fielitz K, Heilmann M, Morik, Schramm A (2014) Robust Selection of Survival Signatures from High-Throughput Genomic Data Using Two-Fold Subsampling. *PLoS One* 9:108818 PMID:2529552

Rihani A, De Wilde B, Zeka F, Laureys G, Francotte N, Tonini GP, Coco S, Versteeg R, Noguera R, Schulte JH, Eggert A, Stallings RL, Speleman F, Vandesompele J, and Van Maerken T (2014) CASP8 SNP D302H (rs1045485) is associated with worse survival in MYCN-amplified neuroblastoma patients. *PLoS One* 9(12):e114696 PMID:2550255

Miller-Delaney SF, Bryan K, Das S, McKiernan RC, Bray IM, Reynolds JP, Gwinn R, Stallings RL, Henshall DC (2014) Differential DNA Methylation Profiles of Coding and Noncoding Genes Define Hippocampal Sclerosis in Human Temporal Lobe Epilepsy. *Brain Epub ahead of print.* PMID:25552301

O'Meara E, Stack D, Phelan S, McDonagh N, Kelly L, Sciort R, Debiec-Rychter M, Morris T, Cochrane D, Sorensen P, O'Sullivan MJ. (2014). Identification of an MLL4-GPS2 fusion as an oncogenic driver of undifferentiated spindle cell sarcoma in a child. *Genes Chromosomes Cancer.* 53:991-8. PMID: 25139254.

Harvey H, Piskareva O, Creevey L, Alcock LC, Buckley PG, O'Sullivan MJ, Segura MF, Gallego S, Stallings RL, Bray IM (2014). Modulation of chemotherapeutic drug resistance in neuroblastoma SK-N-AS cells by the neural apoptosis inhibitory protein and miR-520f. *Int J Cancer.* 136:1579-88. PMID: 25137037.

Uwineza A, Gill H, Buckley P, Owens C, Capra M, O'Sullivan C, McDermott M, Brett F, Farrell M, Pears J, O'Sullivan MJ. (2014) Rhabdoid tumor: the Irish experience 1986-2013. *Cancer Genet.* 207:398-402. PMID: 25085603.

Lynn M, Shah N, Conroy J, Ennis S, Morris T, Betts D, O'Sullivan M. (2014) A Study of Alveolar Rhabdomyosarcoma Copy Number Alterations by

Single Nucleotide Polymorphism Analysis. *Appl Immunohistochem Mol Morphol*. 2014; 22:213-21. PMID:24614150

Karlsson J, Holmquist Mengelbier L, Ciornei CD, Naranjo A, O'Sullivan MJ, Gisselsson D. (2014) Clear cell sarcoma of the kidney demonstrates an embryonic signature indicative of a primitive nephrogenic origin. *Genes Chromosomes Cancer*.53:381-91. PMID: 24488803

Yoshimi A, van den Heuvel-Eibrink MM, Baumann I, Schwarz S, Simonitsch-Klupp I, de Paepe P, Campuzano V, Kerndrup GB, O'Sullivan M, Devito R, Leguit R, Hernandez M, Dworzak M, de Moerloose B, Stary J, Hasle H, Smith OP, Zecca M, Catala A, Schmutz M, Locatelli F, Führer M, Fischer A, Guderle A, Nöllke P, Strahm B, Niemeyer CM.(2014) Comparison of horse and rabbit antithymocyte globulin in immunosuppressive therapy for refractory cytopenia of childhood.*Haematologica*.99:656-63. PMID: 24162791

Lennon J, Bright SA, Carroll E, Butini S, Campiani G, O'Meara A, Williams DC and Zisterer DM. (2014) The novel pyrrolo-1,5-benzoxazepine, PBOX-6, synergistically enhances the apoptotic effects of carboplatin in drug sensitive and multidrug resistant neuroblastoma cells. *Biochem. Pharmacol.* 87 (4):611-24 PMID: 24406249

A study of alveolar rhabdomyosarcoma copy number alterations by single nucleotide polymorphism analysis. Lynn M, Shah N, Conroy J, Ennis S, Morris T, Betts D, O'Sullivan M. *Appl Immunohistochem Mol Morphol*. 2014 Mar;22(3):213-21. doi: 10.1097/ PubMed PMID: 24614150.

Atypical Alstrom syndrome with novel ALMS1 mutations precluded by current diagnostic criteria. *Eur J Med Genet*. Casey J, McGettigan P, Brosnahan D, Curtis E, Treacy E, Ennis S, Lynch SA. 2014 Feb;57(2-3):55-9. doi:10.1016/j.ejmg.2014.01.007. Epub 2014 Feb 3. PubMed PMID: 24503146.

Children's Clinical Research Unit

Cardiology (2)

Woods I, Flanagan TC (2014) Electrospinning of Biomimetic Scaffolds for Tissue-Engineered Vascular Grafts: Threading the Path. *Expert Review of Cardiovascular Therapy*. 12:815-32 PMID: 24903895

Computational modeling of blood flow in the aorta - insights into eccentric dilatation of the ascending aorta after surgery for coarctation.

Szopos M, Poussineau N, Maday Y, Canniffe C, Celermajer DS, Bonnet D, Ou P. *J Thorac Cardiovasc Surg*. 2014 Oct;148(4):1572-82. PMID:24521947

Orthopaedics (2)

Tarrant RC, O'Loughlin PF, Lynch S, Queally JM, Sheeran P, Moore D & Kiely PJ (2014) Timing and predictors of return to short-term functional activity in adolescent idiopathic scoliosis after posterior spinal fusion: a prospective follow-up study. *Spine* 39 (18):1471-1478. PMID: 24875955

Tarrant RC, Lynch S, Sheeran P, Harrington M, O'Loughlin P, Moore D & Kiely PJ (2014) Low body mass index in adolescent idiopathic scoliosis – relationship with pre and postsurgical factors. *Spine* 39 (2):140-148. PMID: 24153169

Autism (17)

Buxbaum JD, Bolshakova N, Brownfeld JM, Anney RJ, Bender P, Bernier R, Cook EH, Coon H, Cuccaro M, Freitag CM, Hallmayer J, Geschwind D, Klauck SM, Nurnberger JI, Oliveira G, Pinto D, Poustka F, Scherer SW, Shih A, Sutcliffe JS, Szatmari P, Vicente AM, Vieland V and Gallagher L. (2014) The Autism Simplex Collection: an international, expertly phenotyped autism sample for genetic and phenotypic analyses. *Mol Autism*. 20;5:34. PMID:25392729

Merikangas AK, Segurado R, Heron EA, Anney RJ, Paterson AD, Cook EH, Pinto D, Scherer SW, Szatmari P, Gill M, Corvin AP and Gallagher L. (2014). The phenotypic manifestations of rare genic CNVs in autism spectrum disorder. *Mol Psychiatry*. 10.1038/mp.2014.150. PMID:25421404

De Rubeis S, He X, Goldberg AP, Poultney CS, Samocha K, Cicek AE, Kou Y, Liu L, Fromer M, Walker S, Singh T, Klei L, Kosmicki J, Shih-Chen F, Aleksic B, Biscaldi M, Bolton PF, Brownfeld JM, Cai J, Campbell NG, Carracedo A, Chahrour MH, Chiacchetti AG, Coon H, Crawford EL, Curran SR, Dawson G, Dukatis E, Fernandez BA, Gallagher L, Geller E, Guter SJ, Hill RS, Ionita-Laza J, Jimenez Gonzalez P, Kilpinen H, Klauck SM, Klevzon A, Lee I, Lei I, Lei J, Lehtimäki T, Lin CF, Ma'ayan A, Marshall CR, McInnes AL, Neale B, Owen MJ, Ozaki N, Parellada M, Parr JR, Purcell S, Puura K, Rajagopalan D, Rehnström K, Reichenberg A, Sabo A, Sachse M, Sanders SJ, Schafer C, Schulte-Rüther M, Skuse D, Stevens C, Szatmari P, Tammimies

K, Valladares O, Voran A, Li-San W, Weiss LA, Willsey AJ, Yu TW, Yuen RK; DDD Study; Homozygosity Mapping Collaborative for Autism; UK10K Consortium, Cook EH, Freitag CM, Gill M, Hultman CM, Lehner T, Palotie A, Schellenberg GD, Sklar P, State MW, Sutcliffe JS, Walsh CA, Scherer SW, Zwick ME, Barrett JC, Cutler DJ, Roeder K, Devlin B, Daly MJ and Buxbaum JD (2014). Synaptic, transcriptional and chromatin genes disrupted in autism. *Nature* 515(7526):209-15. PMID: 25363760

Hadley D, Wu ZL, Kao C, Kini A, Mohamed-Hadley A, Thomas K, Vazquez L, Qiu H, Mentch F, Pellegrino R, Kim C, Connolly J; AGP Consortium, Glessner J and Hakonarson H. (2014) The impact of the metabotropic glutamate receptor and other gene family interaction networks on autism. *Nat Commun.* 13;5:4074. PMID:24927284

Seoighe DM, Gadancheva V, Regan R, McDaid J, Brenner C, Ennis S, Betts DR, Eadie PA and Lynch SA (2014) A chromosomal 5q31.1 gain involving PITX1 causes Liebenberg syndrome. *Am J Med Genet A.* 164A(11):2958-60 PMID:25124102

Morris DW, Pearson RD, Cormican P, Kenny EM, O'Dushlaine CT, Perreault LP, Giannoulatou E, Tropea D, Maher BS, Wormley B, Kelleher E, Fahey C, Molinos I, Bellini S, Pirinen M, Strange A, Freeman C, Thiselton DL, Elves RL, Regan R, Ennis S, Dinan TG, McDonald C, Murphy KC, O'Callaghan E, Waddington JL, Walsh D, O'Donovan M, Grozeva D, Craddock N, Stone J, Scolnick E, Purcell S, Sklar P, Coe B, Eichler EE, Ophoff R, Buizer J, Szatkiewicz J, Hultman C, Sullivan P, Gurling H, Mcquillin A, St Clair D, Rees E, Kirov G, Walters J, Blackwood D, Johnstone M, Donohoe G; International Schizophrenia Consortium, SGENE+ Consortium, O'Neill FA; Wellcome Trust Case Control Consortium 2, Kendler KS Gill M, Riley BP, Spencer CC and Corvin A (2014) An inherited duplication at the gene p21 Protein-Activated Kinase 7 (PAK7) is a risk factor for psychosis. *Hum Mol Genet.* 15;23(12): 3316-26. PMID: 24474471

Pinto D, Delaby E, Merico D, Barbosa M, Merikangas A, Klei L, Thiruvahindrapuram B, Xu X, Ziman R, Wang Z, Vorstman JA, Thompson A, Regan R, Pilorge M, Pellecchia G, Pagnamenta AT, Oliveira B, Marshall CR, Magalhaes TR, Lowe JK, Howe JL, Griswold AJ, Gilbert J, Duketis E, Dombroski BA, De Jonge MV, Cuccaro M, Crawford EL, Correia CT, Conroy J, Conceição IC, Chiocchetti AG, Casey JP, Cai G, Cabrol C, Bolshakova N, Bacchelli E, Anney R, Gallinger S, Cotterchio M, Casey G, Zwaigenbaum L, Wittemeyer K, Wing K, Wallace S, van Engeland H, Tryfon A, Thomson S, Soorya L, Rogé B, Roberts W, Poustka F, Mouga S, Minshew N, McInnes LA,

McGrew SG, Lord C, Leboyer M, Le Couteur AS, Kolevzon A, Jiménez González P, Jacob S, Holt R, Guter S, Green J, Green A, Gillberg C, Fernandez BA, Duque F, Delorme R, Dawson G, Chaste P, Café C, Brennan S, Bourgeron T, Bolton PF, Bölte S, Bernier R, Baird G, Bailey AJ, Anagnostou E, Almeida J, Wijsman EM, Vieland VJ, Vicente AM, Schellenberg GD, Pericak-Vance M, Paterson AD, Parr JR, Oliveira G, Nurnberger JI, Monaco AP, Maestrini E, Klauck SM, Hakonarson H, Haines JL, Geschwind DH, Freitag CM, Folstein SE, Ennis S, Coon H, Battaglia A, Szatmari P, Sutcliffe JS, Hallmayer J, Gill M, Cook EH, Buxbaum JD, Devlin B, Gallagher L, Betancur C and Scherer SW (2014). Convergence of genes and cellular pathways dysregulated in autism spectrum disorders. *Am J Hum Genet.* 94(5):677-94. PMID: 24768552

The impact of the metabotropic glutamate receptor and other gene family interaction networks on autism. Hadley D, Wu ZL, Kao C, Kini A, Mohamed-Hadley A, Thomas K, Vazquez L, Qiu H, Mentch F, Pellegrino R, Kim C, Connolly J; AGP Consortium, Glessner J, Hakonarson H. *Nat Commun.* 2014 Jun 13;5:4074. doi: 10.1038/ncomms5074. PMID:24927284

Kenny EM, Cormican P, Furlong S, Heron E, Kenny G, Fahey C, Kelleher E, Ennis S, Tropea D, Anney R, Corvin AP, Donohoe G, Gallagher L, Gill M and Morris DW (2014) Excess of rare novel loss-of-function variants in synaptic genes in schizophrenia and autism spectrum disorders. *Mol Psychiatry* 19(8):872-9 PMID:24126926

Fitzgerald J, Johnson K, Kehoe E, Bokde A, Garavan H, Gallagher L, McGrath J (2014). Disrupted functional connectivity in dorsal and ventral attention networks during attention orienting in autism spectrum disorders. *Autism Res.* 2014 Nov 26. doi: 10.1002/aur.1430. [Epub ahead of print] PMID: 25428212

Moreira DP, Griesi-Oliveira K, Bossolani-Martins AL, Lourenço NC1, Takahashi VN, da Rocha KM, Moreira ES, Vadasz E, Meira JG, Bertola D, Halloran EO, Magalhães TR, Fett-Conte AC, Passos-Bueno MR (2014) Investigation of 15q11-q13, 16p11.2 and 22q13 CNVs in autism spectrum disorder Brazilian individuals with and without epilepsy. *PLoS One.* 2014 Sep 25;9(9):e107705 PMID: PMC4177849

Pinto, D, E Delaby, D Merico, M Barbosa, A Merikangas, L Klei, B Thiruvahindrapuram, X Xu, R Ziman, Z Wang, JAS Vorstman, A Thompson, R Regan, M Pilorge, G Pellecchia, AT Pagnamenta, B Oliveira, CR Marshall, TR Magalhaes, JK Lowe, JL Howe, AJ Griswold, J Gilbert, E Duketis, BA Dombroski, MV De Jonge, M Cuccaro, EL Crawford, CT Correia, J Conroy, IC Conceição, AG Chiocchetti, JP Casey, G Cai, C Cabrol, N Bolshakova, E Bacchelli, R Anney, S

Gallinger, M Cotterchio, G Casey, L Zwaigenbaum, K Wittmeyer, K Wing, S Wallace, H van Engeland, A Tryfon, S Thomson, L Soorya, B Rogé, W Roberts, F Poustka, S Mougá, N Minshew, LA McInnes, SG McGrew, C Lord, M Leboyer, AS Le Couteur, A Kolevzon, P Jiménez González, S Jacob, R Holt, S Guter, J Green, A Green, C Gillberg, BA Fernandez, F Duque, R Delorme, G Dawson, P Chaste, C Café, S Brennan, T Bourgeron, PF Bolton, S Bölte, R Bernier, G Baird, AJ Bailey, E Anagnostou, J Almeida, EM Wijsman, VJ Vieland, AM Vicente, GD Schellenberg, M Pericak-Vance, AD Paterson, JR Parr, G Oliveira, JI Nurnberger, AP Monaco, E Maestrini, SM Klauck, H Hakonarson, JL Haines, DH Geschwind, CM Freitag, SE Folstein, S Ennis, et al. (2014) Convergence of genes and cellular pathways dysregulated in autism spectrum disorders. *Am J Hum Genet* 94, 677–694 PMID: PMC4067558

Joint analysis of psychiatric disorders increases accuracy of risk prediction for schizophrenia, bipolar disorder, and major depressive disorder. Maier R, et al. *Am J Hum Genet*. 2015 Feb 5;96(2):283-94. doi: 10.1016/j.ajhg.2014.12.006. Epub 2015 Jan 29. PubMed PMID: 25640677; PubMed Central PMCID: PMC4320268.

The impact of the metabotropic glutamate receptor and other gene family interaction networks on autism. Hadley D, et al. *Nat Commun*. 2014 Jun 13;5:4074. doi: 10.1038/ncomms5074. PubMed PMID: 24927284; PubMed Central PMCID: PMC4059929.

TDP2 protects transcription from abortive topoisomerase activity and is required for normal neural function. Gómez-Herreros F, Schuurs-Hoeijmakers JH, McCormack M, Grealley MT, Rulten S, Romero-Granados R, Counihan TJ, Chaila E, Conroy J, Ennis S, Delanty N, Cortés-Ledesma F, de Brouwer AP, Cavalleri GL, El-Khamisy SF, de Vries BB, Caldecott KW. *Nat Genet*. 2014 May;46(5):516-21. doi: 10.1038/ng.2929. Epub 2014 Mar 23. PubMed PMID: 24658003.

An inherited duplication at the gene p21 Protein-Activated Kinase 7 (PAK7) is a risk factor for psychosis. Morris DW, Pearson RD, Cormican P, Kenny EM, O'Dushlaine CT, Perreault LP, Giannoulatou E, Tropea D, Maher BS, Wormley B, Kelleher E, Fahey C, Molinos I, Bellini S, Pirinen M, Strange A, Freeman C, Thiselton DL, Elves RL, Regan R, Ennis S, Dinan TG, McDonald C, Murphy KC, O'Callaghan E, Waddington JL, Walsh D, O'Donovan M, Grozeva D, Craddock N, Stone J, Scolnick E, Purcell S, Sklar P, Coe B, Eichler EE, Ophoff R, Buizer J, Szatkiewicz J, Hultman C, Sullivan P, Gurling H, Mcquillin A, St Clair D, Rees E, Kirov G, Walters J, Blackwood D, Johnstone M, Donohoe G; International Schizophrenia Consortium,

SGENE+ Consortium, O'Neill FA; Wellcome Trust Case Control Consortium 2, Kendler KS, Gill M, Riley BP, Spencer CC, Corvin A. *Hum Mol Genet*. 2014 Jun 15;23(12):3316-26. doi: 10.1093/hmg/ddu025. Epub 2014 Jan 28. PubMed PMID: 24474471; PubMed Central PMCID: PMC4030770.

Excess of rare novel loss-of-function variants in synaptic genes in schizophrenia and autism spectrum disorders. Kenny EM, Cormican P, Furlong S, Heron E, Kenny G, Fahey C, Kelleher E, Ennis S, Tropea D, Anney R, Corvin AP, Donohoe G, Gallagher L, Gill M, Morris DW. *Mol Psychiatry*. 2014 Aug;19(8):872-9. doi: 10.1038/mp.2013.127. Epub 2013 Oct 15. PubMed PMID: 24126926.

Neonatology (13)

Kieran EA, O'Donnell CPF(2014) Accuracy of length measurements on neonatal umbilical catheters. *Arch Dis Child Fetal Neonatal Ed* 99: F520-F521 PMID: 25249190

Kieran EA, O'Callaghan N, O'Donnell CPF(2014) Unlicensed and off-label drug use in an Irish neonatal intensive care unit. *Acta Paediatr* 2014; 103: e139-142 PMID:24397831

Congenital diaphragmatic hernia surveillance. Chuckwu J, Cunney R, Molloy E. *Ir Med J*. 2014 Sep;107(8):261. No abstract available. PMID: 25282980

Neonatal Brain Injury and Systemic Inflammation: modulation by Activated Protein c ex vivo. E. *Clin Exp Immunol*. 2014 Sep 10. doi: 10.1111/cei.12453. [Epub ahead of print] PMID: 25204207

Eliwan H, Watson R, Aslam S, Regan I, Philbin B, O'Hare F, O'Neill A, Preston R, Blanco A, Grant T, Nolan B, Smith O, Molloy Easily missed, potentially fatal complication in an extremely preterm infant. Geoghegan SF, Vavasseur C, Donoghue V, Molloy EJ. *BMJ Case Rep*. 2014 Jul 22;2014. pii: bcr2013201397. doi: 10.1136/bcr-2013-201397. PMID: 25053692

Carroll A, Onwuneme C, McKenna MJ, Mayne PD, Molloy EJ, Murphy NP Vitamin D status in Irish children and adolescents: value of fortification and supplementation. *Clin Pediatr (Phila)*. 2014 Dec;53(14):1345-51. doi: 10.1177/0009922814541999. Epub 2014 Jul 7. PMID: 25006113

Horan M, Gibney E, Molloy E, McAuliffe F Methodologies to assess paediatric adiposity. *Ir J Med Sci*. 2014 May 4. [Epub ahead of print] PMID: 24791970

Freyne B, Stafford A, Knowles S, O'Hora A, Molloy E. Perinatal *Treponema pallidum*: evidence based guidelines to reduce mother to child transmission. *Ir Med J*. 2014 Jan;107(1):14-6. PMID: 24592640

Waters A, Jennings K, Fitzpatrick E, Coughlan S, Molloy EJ, De Gascun CF, Hall WW, Knowles SJ. Incidence of congenital cytomegalovirus infection in Ireland: implications for screening and diagnosis. *J Clin Virol*. 2014 Mar;59(3):156-60. doi: 10.1016/j.jcv.2013.12.007. Epub 2014 Jan 3. PMID: 24461765

Donnelly JM, Walsh JM, Byrne J, Molloy EJ, McAuliffe FM. Impact of maternal diet on neonatal anthropometry: a randomized controlled trial. *Pediatr Obes*. 2015 Feb;10(1):52-6. doi: 10.1111/j.2047-6310.2013.00216.x. Epub 2014 Jan 20. PMID: 24443392

The prevalence of syphilis at childbirth in Ireland: a six-year review. Lutomski JE, Shiely F, Molloy EJ. *J Matern Fetal Neonatal Med*. 2014 Nov;27(17):1823-5. doi: 10.3109/14767058.2013.876985. Epub 2014 Jan 13. PMID: 24354591

Rebolledo J, Igoe D, O'Donnell J, Domegan L, Boland M, Freyne B, McNAMARA A, Molloy E, Callaghan M, Ryan A, O'Flanagan D. Influenza in hospitalized children in Ireland in the pandemic period and the 2010/2011 season: risk factors for paediatric intensive-care-unit admission. *Epidemiol Infect*. 2014 Sep;142(9):1826-35. doi: 10.1017/S0950268813002732. Epub 2013 Nov 11. PMID: 24229618

Neary E, NíÁinle F, Cotter M, McCallion N (2014). Coagulation Values in Extreme Premature Infants. *Transfusion*. 2014;54(8):2134. PMID: 25130336

Emergency Medicine (7)

Gannon B, Jones C, Wakai A, O'Sullivan R. The cost of data collection for performance monitoring in hospitals: protocol for a systematic review. *Systematic Reviews* 2014 Jun 16;3:65. PMID: 24934212

McCabe A, Crispino G, Nally S, O'Sullivan, Wakai A. Feasibility analysis of emergency department key performance indicators in Ireland: an interim analysis. In: Top Ten Abstracts. *Irish Journal of Medical Science* 2014 Oct;183 Suppl 10:476. PMID: 25305087

McCabe A, Crispino G, Nally S, O'Sullivan, Wakai A. Feasibility Analysis of Emergency Key Performance Indicators in Ireland: An Interim Analysis. In: 2014 annual meeting issue. *Academic Emergency Medicine* 2014 May;21 Suppl 1:S315. PMID: 24787373

Murphy, A., O'Sullivan, R., Wakai, A., Grant, T. S., Barrett, M. J., Cronin, J., McCoy, S. C., Hom, J. & Kandamany, N. 2014. Intranasal fentanyl for the management of acute pain in children. *Cochrane Database Syst Rev*, 10, Cd009942 PMID25300594.

S McCoy, A Murphy, P J Larkin, M Brenner, A Wakai, R G O'Sullivan. Safe and Effective change: Critical Elements of Introducing A Successful Paediatric Procedural Sedation Programme in The Emergency Department. *Academic Emergency Medicine* 2014; 21(5): S135 (no PMID available)

McCoy S, M. Lyttle, S. Hartshorn, P. Larkin, M. Brenner, R. O'Sullivan. A baseline study of procedural sedation practices in paediatric emergency medicine: a Paediatric Emergency Research United Kingdom & Ireland (PERUKI) network study. *International Emergency Nursing* 2014; 22(4):285 doi: 10.1016/j.ienj.2014.08.006 (no PMID available)

Murphy A, Barrett M, Cronin J, McCoy S, Larkin P, Brenner M, Wakai A, O'Sullivan R. A qualitative study of the barriers to prehospital management of acute pain in children. *Emerg Med J*. 2014 Jun;31(6):493-8. doi: 10.1136/emered-2012-202166. Epub 2013 Mar 21.

Burns Unit (1)

Brenner M, Hilliard C, Peel G, Crispino G, Geraghty R & O'Callaghan G (2014) Management of pediatric skin-graft donor sites: a randomized controlled trial of three wound care products. *The Journal of Burn Care & Research*, Published online 2nd September 2014. PMID: 25185932