PAUL GISSEN - UNIVERSITY COLLEGE LONDON

Personal Details:

Professor Paul Gissen

Present Appointment: Wellcome Trust Senior Research Fellow in Clinical Sciences

Head of "Genetics and Genomic Medicine" Academic Programme

UCL Great Ormond Street Institute of Child Health

Group Leader

MRC Laboratory for Molecular Cell Biology

Honorary Consultant in Paediatric Metabolic Medicine

Great Ormond Street Hospital for Children

Date of Appointment: 01 03 2011

GMC: 4186762

Education / Qualifications

2011	Fellow of Royal College of Paediatrics and Child Health	RCPCH
2008	Certificate of Completion of Training in Paediatrics with the Special Interest in Inherited Metabolic Disorders	GMC
2002-2005	PhD Human Molecular Genetics	University of Birmingham

Professional History

Dates	Detail of position held	Institution
01 *Mar 2006*-28 Feb 2011	GSK Clinician Scientist Fellow (Senior Lecturer)	School of Clinical and Experimental Medicine, College of Medical and Dental Sciences, University of Birmingham.
01 *Apr 2008*-28 Feb 2011	Honorary Consultant in Paediatric Metabolic Medicine	Birmingham Children's Hospital
01 *Mar 2006*-31 Mar 2008	Honorary SpR in Inherited Metabolic Diseases	Birmingham Children's Hospital
05 Mar 2005-05 Mar 2006	WellChild Clinical Lecturer in Paediatrics and Child Health, Honorary SpR in Inherited Metabolic Diseases,	University of Birmingham, Birmingham Children's Hospital
01 Mar 2002 - 06 Mar 2005	RCPCH and WellChild research training fellow	Section of Medical and Molecular Genetics, Department of Paediatrics, University of Birmingham
01 Mar 2001 - 28 Feb 2002	Clinical Research Fellow in Paediatric Hepatology	Liver Unit, Birmingham Children's Hospital
01 Sep 2000– 28 February 2001	Specialist Registrar in Paediatrics (core neonatology)	Sharoe Green Hospital/ Royal Preston Hospital
02 Mar 2000 – 31 Aug 2000	Specialist Registrar in General Paediatrics	Fairfield General Hospital, Bury.
01 Aug 1997 – 30 Aug	Senior House Office in Paediatrics and Neonatology	Royal Manchester Children's Hospital, Salford Royal Hospital, Sheffield

1999		Children's Hospital).
01 Aug 1996 – 30 July 1997	Senior House Officer in Obstetrics and Gynaecology.	Queen Mother's Hospital, Glasgow, Western Infirmary, Glasgow
01 Aug 1995-30 July 1996	Junior House Officer in Medicine and Surgery	Stobhill General Hospital, Glasgow, Crosshouse General Hospital, Kilmarnock

Other Appointments And Affiliations

Membership of learned societies

Society for Study of Inborn Errors of Metabolism
British Society for Gene and Cell Therapy
British Inherited Metabolic Diseases Group
American Society of Cell Biology
American Society for Human Genetics
British Society for Genetic Medicine

Committee memberships

Board and annual meeting organising committee member "British Society for Gene and Cell Therapy"
Member of "The AADC Research Trust Medical & Scientific Advisory Board"
Member of the "UCL Rare Disease Steering Committee"
Member of "Niemann Pick Research Foundation Advisory Committee".
Member of "Batten's Disease Family Association Medical & Scientific Advisory Board"

Consultancies

"Synageva/Alexion" from September 2012.
"Dipharma" from July 2016

Prizes, Awards And Other Honours

Dates	Detail of prize, award or honour	Awarding/electing body
2013	Starting Award	European Research Council
2011	Wellcome Trust Senior Research Fellowship in Clinical Sciences	Wellcome Trust
2011	Fellow of the Royal College of Paediatrics and Child Health	Royal College of Paediatrics and Child Health (RCPCH)
2006	RCPCH/WellChild Best Researcher Award"	Royal College of Paediatrics and Child Health
2006	"Promega Young Geneticist of the Year" second prize	The Genetics Society
2006	GSK Clinician Scientist Fellowship	GSK
2004	Pfizer Academic Travel Award	Pfizer
2004	"Alex Mowat Prize" for the best presentation in hepatology	World Congress in Paediatric Gastroenterology, Hepatology and Nutrition
2004	Travelling Scholarship to attend annual Society for Study of Inborn Errors of Metabolism meeting	British Inherited Metabolic Diseases Group
2004	Young Investigator Award to attend World Congress of Paediatric Gastroenterology, Hepatology and Nutrition	WCPGHAN
2002-2005	RCPCH and WellChild Research Training Fellowship	WellChild children's charity and Royal College of Paediatrics and Child Health
1995	MB ChB with Commendation	University of Glasgow Medical School

Current Funding

1) MRC Biomedical Catalyst: DPFS scheme; PI Gissen, co-PIs Thrasher, Waddington, Alexander; "AAV-mediated Gene Therapy for Ornithine Transcarbamylase Deficiency"; £1.9M; project coordination and

supervision of a clinician scientist Dr Julien Baruteau and a postdoctoral scientist (to be appointed) in preclinical studies; 1/04/2016-1/04/2018, (2 hours per week).
2) Vtesse pharma; UK CI Gissen; Pivotal trial of two-weekly VTS270 intrathecal infusions in patients with Niemann Pick type C disease; ~£400,000; supervise a team of nurses and clinical fellows to run the clinical trial in 4 patients;1/1/2016-1/7/2017, (2 hours per week clinical time).
3) Batten Disease Family Association; PI Gissen, Co-PIs Robin Ketteler, Sara Mole, Michael Devine; "Development of a high content imaging molecule screen in neuronally differentiated iPS cells from CLN5 patients"; £20,000; supervise a postdoctoral scientist Daniel Little and a masters in cell biology student Olivier Pezzini-Picart developing high content imaging assays in iPSc derived neurons appropriate for drug screening in CLN5 type Batten disease, July 2015- July 2016, (2 hours per week).
4) Biomarin Pharma; UK CI Gissen; "Phase 1 extension trial of intra-cerebro-ventricular infusions of enzyme replacement therapy for CLN2 type Batten disease"; ~£3M; supervise a team of nurses and clinical fellows to run the clinical trial in 4 patients; September 2015- January 2020, (2 hours per week clinical time).
5) European Research Council Starting Grant; PI Gissen; "Cultured Liver Organoids in Cholestasis"; £1.3M; supervise a team including postdoctoral scientists Maelle Lorvellec and Dipok Dhar and a research technician Rebeca Fiadeiro developing in vitro liver model based on decellularised liver scaffolds and iPS cell derived liver cells; January 2014 - January 2019, (4 hours per week).
6) Innovative Medicines Initiative consortium; project coordinator and lead academic applicant Zameel Cader, University of Oxford, Pharma lead Martin Graf, Roche, UCL PIs Gissen, Mills, Heales and Skuse; "Stem cells for Biological Assays of Novel drugs and predictive toxicology"; contribution to Gissen lab 500,000 Euro; supervise a postdoctoral scientist Daniel Little and a research technician Olukunbi Mosaku developing high content imaging assays in iPSc derived neuronal cells and perform CRISPR Cas9 based mutation correction to create isogenic controls; December 2012 - January 2017, (2 hours per week).
7) Wellcome Trust Senior Fellowship; PI Gissen; "Involvement of trafficking proteins VPS33B and VIPAR in development and disease"; £1.3M; supervise a technician Anna Straatman-Iwanowska and postdoctoral scientist Blerida Banushi; September 2011 – August 2017 ;(full-time).

Academic Supervision

PhD student supervision

Previous PhD students (all successfully awarded PhD within 4 years).

A. As a primary supervisor:

1) Andrew Cullinane: 2006-2009 (funded by a Children Liver Disease Foundation Fellowship). Currently Associate Professor at Howard University Washington DC, USA.
2) Holly Smith: 2008-2011(funded by a MRC DTA studentship). Currently a Senior Research Facilitator, University of Birmingham.
3) Dr James Davison: 2009-2012 (funded by a BCH Research Foundation Springboard Fellowship and SPARKS Research Training Fellowship). Currently Consultant in Paediatric Inherited Metabolic Diseases at Great Ormond Street Hospital for Children.
4) Blerida Banushi: 2010-2013 (Marie Curie Early Career Scientist Fellowship). Currently a postdoc in Queensland, Australia.
5) Kirsten McKay: 2009-2016 (part-time NHS-funded research project at the University of Birmingham).

Current PhD students:

A. As a primary supervisor:

1) Clare Rogerson: started in 2013 (MRC LMCB PhD programme), aim to submit in April 2017.
2) Kunbi Mosaku: 2014-2017 (part-time PhD student funded by STEMBANCC) aim to submit in March 2017.

Current research summary

The aim of my research is to improve disease understanding and treatment of Inherited Metabolic Disorders (IMD).

1. *Basic cell pathology in IMD*

Wellcome Trust Senior Fellowship (2011-2016) funds research into the mechanism of Arthrogryposis, Renal Dysfunction and Cholestasis syndrome, a multi-system disorder with wide range of clinical problems including

bleeding, cholestasis, bone and kidney disease. We discovered a novel protein complex that regulates intracellular trafficking of proteins, establishment of cell polarity and organellar biogenesis.
2. *Induced pluripotent stem cells (iPSc) as models of disease.*
I am one of the UCL PIs for the "Innovative Medicines Initiative" funded StemBANCC consortium led by Oxford University (2012-2017). This work allows active collaboration with Drs Robin Ketteler (LMCB) and Kevin Mills (ICH) to develop high-content imaging assays in differentiated patient-derived iPSc. The assays test metabolic cell processes such as mitochondrial and lysosomal functions and will be used for drug screening.
The European Research Council starting award (2015-2019) funds development in my lab of 3D "liver organoids" that combine decellularised liver extracellular matrix and patient iPSc derived liver cells to test potential treatments for IMD. This is a collaboration with Prof Paolo De Coppi (ICH).
3. *AAV based gene therapy (AAV-GT) to treat liver based IMD (LIMD).*
Close collaborations with the UCL colleagues Professor Thrasher and Dr Simon Waddington and Prof Ian Alexander (Sydney University) allowed us to initiate "proof of concept" studies of AAV-GT in LIMD. "Action Medical Research" Clinical Research Fellowship for Dr Julien Baruteau, project grants from the GOSH/ICH NIHR Biomedical Research Centre and "Children's Liver Disease Foundation" funded the initial studies.
4. *Clinical trials in IMD.*

I lead several clinical trials as a PI and CI including industry-funded "first in human trials" of novel therapies in IMD such as hepatocyte progenitor cell therapy for urea cycle disorders.

Current Teaching Activity

I lecture on various masters courses at UCL. I do the following lectures:
- "Liver development", 2 hours;
- "Genetics of Inherited Metabolic Disorders": 2 hours;
- "Novel therapies in Inherited Metabolic Disorders": 2 hours.

I regularly give invited lectures, including two recent "Batten Disease and promising therapies" and "Liver Directed Gene Therapy" given at the British Society for Gene and Cell Therapy.

Publications

1. Hanley J, Kumar Dhar D, Mazzacuva F, Fiadeiro R, Burden JJ, Lyne AM, Smith H, Straatman-Iwanowska A, Banushi B, Virasami A, Mills K, Lemaigre FP, Knisely AS, Howe S, Sebire N, Waddington S, Paulusma CC, Clayton P, **Gissen P**. Vps33b is Crucial for Structural and Functional Hepatocyte Polarity. J Hepatol. 2017 (*In Press*).
2. Baruteau J, Waddington SN, Alexander IE, **Gissen P**. Delivering efficient liver-directed AAV-mediated gene therapy. Gene Ther. 2017 (*In Press*).
3. Carss KJ, Arno G, Erwood M, Stephens J, Sanchis-Juan A, Hull S, Megy K, Grozeva D, Dewhurst E, Malka S, Plagnol V, Penkett C, Stirrups K, Rizzo R, Wright G, Josifova D, Bitner-Glindzicz M, Scott RH, Clement E, Allen L, Armstrong R, Brady AF, Carmichael J, Chitre M, Henderson RH, Hurst J, MacLaren RE, Murphy E, Paterson J, Rosser E, Thompson DA, Wakeling E, Ouwehand WH, Michaelides M, Moore AT; NIHR-BioResource Rare Diseases Consortium., Webster AR, Raymond FL. Comprehensive Rare Variant Analysis via Whole-Genome Sequencing to Determine the Molecular Pathology of Inherited Retinal Disease. Am J Hum Genet. 2017 (*In Press*).
4. Gruber R, Rogerson C, Windpassinger C, Banushi B, Straatman-Iwanowska A, Hanley J, Forneris F, Strohal R, Ulz P, Crumrine D, Menon GK, Blunder S, Schmuth M, Müller T, Smith H, Mills K, Kroisel P, Janecke AR, **Gissen P**. Autosomal recessive Keratoderma-Ichthyosis-Deafness (ARKID) syndrome is caused by VPS33B mutations affecting Rab protein interaction and collagen modification. J Invest Dermatol. 2016 (*In Press*).
5. Meyer E, Carss KJ, Rankin J, Nichols JM, Grozeva D, Joseph AP, Mencacci NE, Papandreou A, Ng J, Barral S, Ngoh A, Ben-Pazi H, Willemsen MA, Arkadir D, Barnicoat A, Bergman H, Bhate S, Boys A, Darin N, Foulds N, Gutowski N, Hills A, Houlden H, Hurst JA, Israel Z, Kaminska M, Limousin P, Lumsden D, McKee S, Misra S, Mohammed SS, Nakou V, Nicolai J, Nilsson M, Pall H, Peall KJ, Peters GB, Prabhakar P, Reuter MS, Rump P, Segel R, Sinnema M, Smith M, Turnpenny P, White SM,

Wieczorek D, Wiethoff S, Wilson BT, Winter G, Wragg C, Pope S, Heales SJ, Morrogh D; UK10K Consortium.; Deciphering Developmental Disorders Study.; NIHR BioResource Rare Diseases Consortium., Pittman A, Carr LJ, Perez-Dueñas B, Lin JP, Reis A, Gahl WA, Toro C, Bhatia KP, Wood NW, Kamsteeg EJ, Chong WK, **Gissen P**, Topf M, Dale RC, Chubb JR, Raymond FL, Kurian MA. Mutations in the histone methyltransferase gene KMT2B cause complex early-onset dystonia. Nat Genet. 2017 (*In Press*).

6. Rogerson C, **Gissen P**. The CHEVI tethering complex: facilitating special deliveries. J Pathol. 2016 Nov;240(3):249-252.
7. Reid E, Papandreou A, Drury S, Boustred C, Yue W, Wedatilake Y, Beesley C, Jacques T, Anderson G, Abulhoul L, Broomfield A, Cleary MA, Grunewald S, Varadkar SM, Lench N, Rahman S, Gissen P, Clayton PT, Mills PB. Advantages and pitfalls of utilizing an extended gene panel for investigating patients with complex neurometabolic phenotypes. Brain (*In Press*).
8. Johnson B, Lowe GC, Futterer J, Lordkipanidze' M, MacDonald D, Simpson MA, Sanchez Guiu' I, Drake S, Bem D, Leo V, Fletcher SJ, Dawood B, Rivera J, Allsup D, Biss T, Bolton-Maggs PH, Collins P, Curry N, Grimley C, James B, Makris M, Motwani J, Pavord S, Talks K, Thachil J, Wilde J, Williams M, Harrison P, **Gissen P**, Mundell S, Mumford A, Daly ME, Watson SP, Morgan NV; UK Genotyping and Phenotyping of Platelets Study Group. Whole exome sequencing identifies genetic variants in inherited thrombocytopenia with secondary qualitative function defects. Haematologica. 2016 Jun 16. pii: haematol.2016.146316. [Epub ahead of print]
9. Banushi B, Forneris F, Straatman-Iwanowska A, Strange A, Lyne AM, Rogerson C, Burden JJ, Heywood WE, Hanley J, Doykov I, Straatman KR, Smith H, Bem D, Kriston-Vizi J, Ariceta G, Risteli M, Wang C, Ardill RE, Zaniew M, Latka-Grot J, Waddington SN, Howe SJ, Ferraro F, Gjinovci A, Lawrence S, Marsh M, Girolami M, Bozec L, Mills K, **Gissen P**. Regulation of post-Golgi LH3 trafficking is essential for collagen homeostasis. Nat Commun. 2016 Jul 20;7:12111.
10. Vanier MT, **Gissen P**, Bauer P, Coll MJ, Burlina A, Hendriksz CJ, Latour P, Goizet C, Welford RW, Marquardt T, Kolb SA. Diagnostic tests for Niemann-Pick disease type C (NP-C): A critical review. Mol Genet Metab. 2016 Aug;118(4):244-54.
11. Tuschl K, Meyer E, Valdivia LE, Zhao N, Dadswell C, Abdul-Sada A, Hung CY, Simpson MA, Chong WK, Jacques TS, Woltjer RL, Eaton S, Gregory A, Sanford L, Kara E, Houlden H, Cuno SM, Prokisch H, Valletta L, Tiranti V, Younis R, Maher ER, Spencer J, Straatman-Iwanowska A, **Gissen P**, Selim LA, Pintos-Morell G, Coroleu-Lletget W, Mohammad SS, Yoganathan S, Dale RC, Thomas M, Rihel J, Bodamer OA, Enns CA, Hayflick SJ, Clayton PT, Mills PB, Kurian MA, Wilson SW. Mutations in SLC39A14 disrupt manganese homeostasis and cause childhood-onset parkinsonism-dystonia. Nat Commun. 2016 May 27;7:11601.
12. Mazzacuva F, Mills P, Mills K, Camuzeaux S, **Gissen P**, Nicoli ER, Wassif C, Te Vruchte D, Porter FD, Maekawa M, Mano N, Iida T, Platt F, Clayton PT. Identification of novel bile acids as biomarkers for the early diagnosis of Niemann-Pick C disease. FEBS Lett. 2016 Jun;590(11):1651-62. doi: 10.1002/1873-3468.12196. Epub 2016 May 27.
13. Maghsoudlou P, Georgiades F, Smith H, Milan A, Shangaris P, Urbani L, Stavros P. Loukogeorgakis[1], Benedetta Lombardi[3], Giuseppe Mazza[4], Neil J. Sebire[5], Mark Turmaine[6], Simon Eaton[1],Jasminka Godovac-Zimmermann[3], Massimo Pinzani[4], **Gissen P** and De Coppi P. Optimization of liver decellularization maintains extracellular matrix micro-architecture and composition predisposing to effective cell seeding. PLOS One [*In Press*].
14. Papandreu A, **Gissen P**. Diagnostic work-up and management of patients with suspected Niemann-Pick disease type C. Therapeutic Advances in Neurological Disorders. 2016 (*In Press*).
15. Aflatounian M, Smith H, Farahani F, Tofighi A, Straatman-Iwanowska A, Khatri U, Tajeddini P, Fallahi GH, **Gissen P**, Rezaei N. Novel VIPAS39 Mutation in a Syndromic Patient with Arthrogryposis, Renal Tubular Dysfunction and Intrahepatic Cholestasis.European Journal of Medical Genetics. 2016 (*In Press*).
16. Deas E, Cremades N, Angelova PR, Ludtmann M, Yao Z, Chen S, Horrocks M, Banushi B, Little D, Devine M, **Gissen P**, Klenerman D, Dobson C, Wood N, Gandhi S, Abramov AY. Alpha-synuclein oligomers interact with metal ions to induce oxidative stress and neuronal death in Parkinson's disease. Antioxid Redox Signal. 2015 Nov 12. [Epub ahead of print]
17. Heywood WE, Camuzeaux S, Doykov I, Patel N, Preece RL, Footitt E, Cleary M, Clayton P, Grunewald S, Abulhoul L, Chakrapani A, Sebire NJ, Hindmarsh P, de Koning TJ, Heales S, Burke D,

Gissen P, Mills K. Proteomic Discovery and Development of a Multiplexed Targeted MRM-LC-MS/MS Assay for Urine Biomarkers of Extracellular Matrix Disruption in Mucopolysaccharidoses I, II, and VI. Anal Chem. 2015 Dec 15;87(24):12238-44.

18. **Gissen P**, Arias IM. Structural and functional hepatocyte polarity and liver disease. J Hepatol. 2015 (*In Press*).
19. Mazza G, De Coppi P, **Gissen P**, Pinzani M. Hepatic Regenerative Medicine. Journal of Hepatology. 2015 63:523-4.
20. Stödberg T. McTague A, Ruiz A, Hirata H, Zhen J, Long P, Farabella I, Meyer E, Kawahara A, Vassallo G, Stivaros S, Bjursell M, Stranneheim H, Tigerschiöld S, Persson B, Bangash I, Das K, Hughes D, Lesko N, Lundeberg J, Scott R, Poduri A, Scheffer I, Smith H, **Gissen P**, Schorge S, Reith M, Topf M, Kullmann D, Harvey R, Prof. Wedell and Kurian, M. Loss-of-function mutations in SLC12A5, encoding the neuronal-specific potassium-chloride co-transporter KCC2, in autosomal recessive migrating partial seizures of infancy (MPSI). Nature Communications. 2015 6:8038.
21. Bem D, Smith H, Banushi B, Burden JJ, White IJ, Hanley J, Jeremiah N, Rieux-Laucat F, Bettels R, Ariceta G, Mumford AD, Thomas SG, Watson SP, **Gissen P.** VPS33B regulates protein sorting into and maturation of α-granule progenitor organelles in mouse megakaryocytes. Blood. 2015 126:133-43.
22. Hegarty R, Hadzic N, Gissen P, Dhawan A. Inherited metabolic disorders presenting as acute liver failure in newborns and young children: King's College Hospital experience. Eur J Pediatr. 2015 Oct;174(10):1387-92.
23. Leo VC, Morgan NV, Bem D, Jones ML, Lowe GC, Lordkipanidzé M, Drake S, Simpson MA, **Gissen P**, Mumford A, Watson SP, Daly ME; UK GAPP Study Group.Use of next-generation sequencing and candidate gene analysis to identify underlying defects in patients with inherited platelet function disorders. J Thromb Haemost. 2015 13:643-50.
24. Patterson MC, Mengel E, Vanier MT, Schwierin B, Muller A, Cornelisse P, Pineda M; NPC Registry investigators. Stable or improved neurological manifestations during miglustat therapy in patients from the international disease registry for Niemann-Pick disease type C: an observational cohort study. Orphanet J Rare Dis. 2015 10:65.
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27. Hersheson J, Burke D, Clayton R, Anderson G, Jacques TS, Mills P, Wood NW, **Gissen P**, Clayton P, Fearnley J, Mole SE, Houlden H. Cathepsin D deficiency causes juvenile-onset ataxia and distinctive muscle pathology. Neurology. 2014 Nov 11;83(20):1873-5.
28. McKay Bounford K, **Gissen P**. Genetic and laboratory diagnostic approach in Niemann Pick disease type C. J Neurol. 261 Suppl 2:S569-75. 2014
29. Mills PB, Camuzeaux SS, Footitt EJ, Mills KA, **Gissen P**, Fisher L, Das KB, Varadkar SM, Zuberi S, McWilliam R, Stödberg T, Plecko B, Baumgartner MR, Maier O, Calvert S, Riney K, Wolf NI, Livingston JH, Bala P, Morel CF, Feillet F, Raimondi F, Del Giudice E, Chong WK, Pitt M, Clayton PT. Epilepsy due to PNPO mutations: genotype, environment and treatment affect presentation and outcome. Brain. May. 2014
30. Dai D, Mills PB, Footitt E, **Gissen P**, McClean P, Stahlschmidt J, Coupry I, Lavie J, Mochel F, Goizet C, Mizuochi T, Kimura A, Nittono H, Schwarz K, Crick PJ, Wang Y, Griffiths WJ, Clayton PT. Liver disease in infancy caused by oxysterol 7α-hydroxylase deficiency: successful treatment with chenodeoxycholic acid. J Inherit Metab Dis. Mar 22. 2014
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37. Holme A, Hurcombe JA, Straatman-Iwanowska A, Inward CI, **Gissen P**, Coward RJ. Glomerular involvement in the arthrogryposis, renal dysfunction and cholestasis syndrome. Clin Kidney J. 2013 Apr;6(2):183-8.
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