

Dr Rhys Dore BA BMBCh MSc MRCPCH - Curriculum Vitae

<https://www.linkedin.com/in/rhysdore/>

Employment

NIHR Academic Clinical Fellow in Clinical Genetics, King's College London The Ule Lab	2023-
Specialty Registrar in Clinical Genetics, Guy's and St Thomas' NHS Foundation Trust General genetics, cancer genetics, cardiac genetics, neurogenetics, prenatal genetics and fertility, dysmorphology, mitochondrial genetics, and metabolic genetics.	2023-
NIHR Academic Clinical Fellow in Paediatrics, University College London The Ryten Lab	2021-23
Paediatric Specialty Trainee, Barts Health NHS Trust General paediatrics and neonates, tertiary neonates, community paediatrics, paediatric gastroenterology	2020-23
Academic Foundation Doctor, Brighton and Sussex University Hospitals NHS Trust Paediatrics, respiratory medicine, general surgery, stroke medicine, HIV medicine, academic paediatrics (redeployment for covid-19)	2018-20

Education

PgCert Interpretation and Clinical Application of Genomic Data	2024-
PgCert Applied Research Methods, King's College London Grade: Distinction (to date)	2023-
PgCert Artificial Intelligence, University of Essex Grade: Distinction	2023-24
PgCert Genomic Medicine, University of Exeter Grade: Distinction	2022-23
MRCPCH, Royal College of Paediatrics and Child Health	2021
MSc Paediatrics & Child Health: Advanced Paediatrics, UCL Grade: Distinction Thesis: The experience of young people with primary ciliary dyskinesia as they transition into adulthood.	2020-22
PgCert Healthcare Leadership and Commissioning, University of Brighton Grade: Distinction	2019-22
PgCert Medical Education, University of Newcastle Grade: Merit	2018-19
BM BCh Medicine & Surgery, University of Oxford Merits: biochemistry and medical genetics, applied physiology and pharmacology, psychiatry.	2012-18
BA Medical Sciences, University of Oxford Grade: 2:1 Major: neuroscience. Coursework: tuberculosis vaccine efficacy trials. Research: capillary rarefaction following pregnancy complications. Supplementary subject: the history and philosophy of science.	2012-15

Research Experience

Clinical Researcher, The Ule Lab, UK DRI, King's College London Skills: R, Bash, computational biology, RNA biology.	2023-
Clinical Researcher, The Ryten Lab, Institute of Child Health (Genetics & Genomics), UCL Skills: R, Bash, bioinformatics, public genetic databases, long-read transcriptomics.	2022-23
MSc Researcher, Institute of Child Health (Population, Policy & Practice), UCL Skills: qualitative research (interpretative phenomenological analysis), PPI, rare disease experiences	2021-22
Clinical Researcher, International Neonatology Consortium Skills: systematic review, international collaboration, virtual collaboration.	2020-23
Medical Elective Research, World Medical Fund, Malawi Skills: research in low resource settings, project planning.	2018
Undergraduate researcher, Oxford University Hospitals Neonatal Unit Skills: data collection, data analysis, economic analysis.	2016
UROF Student, Imperial College London Global eHealth Unit Skills: Python, health technology, systematic assessment.	2015
Social Impact Summer Intern, Findacure: The Fundamental Diseases Partnership Skills: Charity engagement, patient collaboration.	2014
Undergraduate Researcher, Oxford Cardiovascular Clinical Research Facility Skills: Image ProPlus, SPSS, data collection, data analysis, manuscript production.	2014

Leadership Experience

Inaugural member, UK Young Academy Elected member of a new academy which brings together various early-career professionals to allow collaboration and social impact.	2022-
Central committee member, European Society of Human Genetics – Young International representative for clinical and academic geneticists. Representative role on the ESHG Policy and Ethics Committee.	2022-
Trustee & Board Member, the McPin Foundation Providing overall strategy and vision for a charity that champions lived experience in mental health research. Additional involvement in website and brand refresh, interviewing, and standard board activities regarding financial monitoring and CEO liaison.	2021-
Scholar, Healthcare Leadership Academy 12-month scholarship to undertake educational sessions regarding leadership skills within a health-related ventures.	2022-23
Co-Lead for PPI, REACH (Research, Evaluation and Audit in Child Health) Network London Determined the plan for patient and public involvement for a multi-site collaborative research network.	2021-23
Innovation Group Member, RCPCH2040 Strategy and vision development for possible innovations within the field of paediatrics and child health.	2020-21
Foundation Doctors' Representative, Brighton and Sussex University Hospitals NHS Trust Represented medical doctors in training at senior leadership meetings.	2018-20
Academic Representative, Brighton and Sussex University Hospitals NHS Trust Negotiated working patterns during the covid-19 redeployment of academic doctors, to ensure balanced priorities.	2018-20

Student Trustee & Board Member, the Archway Foundation	2017-18
Undertook a 12-month educational process to develop appropriate skills for effective board membership. Completed a board audit which allowed targeted hiring of further trustees.	
Founder and President, Worcester Medical Society, University of Oxford	2014-15
Conceived and developed the society. Secured start-up and regular funding via private investment.	
Student Representative, British Medical Association	2014-15
Represented medical students within a national union setting.	
Board of Management Member, the Haven: an integrated health centre	2010-12
Part of a team working on a £250,000 development for a novel integrated health centre. Personally secured £16,000 of funding for internal furnishings.	

Teaching experience

MRCPCH Course Teacher, Evelina Children's Hospital	2023-
Session co-chair, ESHG Conference	2023-
Faculty member, Practicing Qualitative Research in Child and Adolescent Health, UCL	2022-
Teaching assistant, Evidence-Based Medicine BSc, UCL	2022
Study day organiser, Royal Society of Medicine	2021
Non-stipendiary lecturer, Brighton & Sussex Medical School	2018-20
Organiser, Oxford Global Health Group Conference	2017
Non-stipendiary tutor, Green Templeton College, Oxford	2016-17
Skills day organiser, Oxford Medical School Gazette	2016
Stipendiary tutor, Worcester College, Oxford	2014-16
Academic mentor & site manager, Oxford's UNIQ Summer School	2013-15

Oral Presentations

“Don’t let it hold you back”: the experience of transition to adulthood in young people with primary ciliary dyskinesia – an interpretative phenomenological analysis. Qualitative Health Research Network Conference. Virtual. 2024.

Long-read RNA-sequencing identifies novel protein coding transcripts of genes with implications for inherited and complex cardiac disease. European Society of Human Genetics Conference. Glasgow, UK. 2023.

Long-read RNA-sequencing identifies novel potentially protein coding transcripts of genes causing inherited cardiomyopathies. Young Investigator’s Forum with African Society of Human Genetics. Cape Town, South Africa. Speed talk. 2023.

Observed ranges of neonatal blood pressure by gestational age: a systematic review. Congress of joint European Neonatal Societies. Virtual. 2021.

Poster Presentations

Human synonymous codon variation is constrained within multivalent coding regions. 2nd Workshop on Codon Usage. Edinburgh, UK. 2024.

Human synonymous codon variation is constrained within multivalent coding regions. RNA Society Annual Meeting. Edinburgh, UK. 2024.

ELFN1 deficiency is associated with an autosomal recessive neurodevelopmental disorder with epilepsy. European Society of Human Genetics. Berlin, Germany. 2024.

Empowering the Next Generation of Geneticists: Collaborations and Achievements of the ESHG-Young Committee. Berlin, Germany. 2024.

European and African Society of Human Genetics-a collaboration that continues. Berlin, Germany. 2024.

The need for young geneticist societies - a survey by the European Society of Human Genetics - Young Committee (ESHG-Y). Berlin, Germany. 2024.

TX2P: a tool for integrating long-read RNA-sequencing with mass spectrometry data. ASAP Celebration of Scientific Achievement. Virtual. 2024.

Long-read RNA-sequencing identifies novel protein coding transcripts of genes with implications for inherited and complex cardiac disease. American Society of Human Genetics Conference. Washington DC, USA. 2023.

Long-read RNA-sequencing identifies novel protein coding transcripts of genes with implications for inherited and complex cardiac disease. Genomics England Research Summit. London, UK. 2023.

The European Society of Human Genetics - Young Committee - our achievements in the last 5 years and goals for the future. European Society of Human Genetics Conference. Glasgow, UK. 2023.

The ESHG-Young & the Young Investigator Forum: Partnership, networking and innovation. European Society of Human Genetics Conference. Glasgow, UK. 2023.

Paediatric trainee Experience of multi-site Audit and Research (PEAR), a cross sectional London REACH Network study. RCPCH Conference. Glasgow, UK. 2023.

“Don’t let it hold you back”: the experience of transition to adulthood in young people with primary ciliary dyskinesia. RCPCH Conference. Glasgow, UK. 2023.

Long-read RNA-sequencing identifies novel potentially protein coding transcripts of genes causing inherited cardiac disease. 14th International Congress of Human Genetics. Cape Town, South Africa. 2023.

Delivering a discussion-based approach in paediatric journal clubs – a medical education QIP. Paediatric Educators Special Interest Group – Winter Meeting. Virtual. 2023.

Dark Skin Representation within Mobile Applications for Dermatology. European Academy of Dermatology and Venereology Spring Symposium. Virtual. 2021.

Malnutrition and Malarial Infection in Rural Malawi. European Society for Paediatric Infectious Diseases. Virtual. 2020.

A preliminary report of the analysis of the literature concerning parental experiences following newborn blood spot and urinary screening. International Neonatology Association Conference. Gent, Belgium. 2018.

Comparative case reports of parental experiences within the neonatal ward paralleled to a modernised change model. International Neonatology Association Conference. Lyon, France. 2017.

Retrospective analysis of Twitter posts and their relevance to the field of neonatology. International Neonatology Association Conference. Lyon, France. 2017.

The effect of type of feed upon weight growth velocity in premature babies on a home tube feeding programme. International Neonatology Association Conference. Lyon, France. 2017.

Mobile apps to tackle high dietary salt intake: A systematic assessment of health apps. mHealth Conference. Galway, Ireland. 2017.

Growth velocity in premature babies returning home with nasogastric tubes in situ versus those remaining in hospital. International Neonatology Association Conference. Vienna, Austria. 2016.

Publications

Gustavsson EK, Sethi S, Gao Y, Brenton JW, García-Ruiz S, Zhang D, Garza R, Reynolds RH, Evans JR, Chen Z, Grant-Peters M, MacPherson H, Montgomery K, **Dore R**... Ryten M. The annotation of GBA1 has been concealed by its protein-coding pseudogene GBAP1. *Sci. Adv.* 2024;10.

Dore R, Barnes K, Bremner S, Iwami H, Apele-Freimane D, Batton B, Dempsey E, Ergenekon E, Klein A, Pesco-Koplowitz L, Dionne J, Rabe H, International Neonatal Consortium. Neonatal blood pressure by birth weight, gestational age, and postnatal age: a systematic review. *Matern Health Neonatol Perinatol.* 2024;10(9).

Dore R, Nizza IE, Mitchison HM, Lewis C. 'Don't let it hold you back' – the experience of transition to adulthood in young people with primary ciliary dyskinesia: An interpretative phenomenological analysis. *J Health Psychol.* 2024 Jan 28:13591053231223912.

Dore R, D'Souza M, Ghosh N, Carr D, Loucaides E. Paediatric Trainee Experience of Multi-site Audit and Research (PEAR), a cross sectional London REACH Network study. *London Paediatrics.* 2023;4.

Alimohamed MZ, Mnika, K, Adadey, SM, Barbosa-Matos, R, Avram, E, Nevondwe, P, Akurugu, WA, Mupfururirwa, W, de Miranda Cerqueira, JX, **Dore, R**... Mroczek, M. Advancing intercontinental collaboration in human genetics: success story of the African and European Young Investigator Forum. *Eur J Hum Genet.* 2024 Jan;32(1):3-5.

Redman MG, Aguda V, **Dore R**, Lim JO, Speight B, McVeigh T. The role of virtual consultations in cancer genetics: challenges and opportunities introduced by the COVID-19 pandemic. *BJC Rep.* 2023;1:6.

Avram E, Ding C, Cerqueira J, Johari M, da Silva AGF, Sabau D, Noor N, Kalantari S, **Dore R**, Barbosa-Matos R, Mroczek M, Tonini F. The European Society of Human Genetics - Young Committee - activities and achievements between 2019-2022. *Eur J Hum Genet.* 2023 Feb;30:383-386.

Dore R. Dark Skin Representation within Mobile Applications for Dermatology Education: A Scoping Review. *SKINmed.* 2023;21(4):242-247.

Boardman H, Lamata P, Lazdam M, Verburg A, Siepmann T, Upton R, Bilderbeck A, **Dore R**... Leeson P. Variations in Cardiovascular Structure, Function, and Geometry in Midlife Associated With a History of Hypertensive Pregnancy. *Hypertension*. 2020 Jun;75(6):1542-1550

Wood K., O'Brien F., Liddle K., **Dore R**. A neonatal short-term home nasogastric tube feeding programme. *Infant* 2020;16(2): 67-71.

Dore R. Resistin/uric acid index for weight loss prediction? *J Pediatr*. 2020 Sep;224:185-186

Dore R. Letter to the Editor Re: Effectivity of Pancreatic Enzyme Replacement Therapy in Malnourished Children. *J Pediatr Gastroenterol Nutr*. 2020 Oct;71(4):e137

Awards, Prizes, Scholarships

Dean's Commendation for Exceptional Performance, University of Exeter National	2023
Wellcome Connecting Science Bursary International	2023
European Society of Human Genetics Travel Fellowship International	2023
Special Commendation, UK Cancer Genetics Group National	2021
Michelle Zalkin Scholarship, University College London International	2021
Foundation ePortfolio Award, South Thames Foundation School Deanery Regional	2020
Leadership Award, South Thames Foundation School Deanery Regional	2019
Vice Chancellor's Social Impact Award, University of Oxford Local	2018
Presidential Essay Prize, The British Society for the Study of Vulval Disease National	2017
MDU Mini Essay Competition, The Medical Defence Union National	2015
Moritz-Heyman Scholarship, University of Oxford National	2012

Certifications

Newborn Life Support	Expires 2025
Advanced Paediatric Life Support	Expires 2025
Advanced (Adult) Life Support	Expires 2024