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A Course in the use and interpretation of clinical laboratory tests XII Edition

DRA. FRANCES SMITH

SYNNOVIS UK // KING´S COLLEGE LONDON, UK

Education and Qualifications:

- Fellow of the Royal College of Pathologists 2025
- Doctor of Clinical Science – University of Manchester 2024
- Higher Specialist Scientific Training program in Genomics 2025
- Health Care Professions Council registration as Clinical Scientist in Molecular Genetics –March 2013 - present
- BSc Genetics 1st Class Hons Sheffield University 2005

Employment History:

Head of Laboratory and Clinical Scientist
Synnovis at King's College Hospital, Molecular Pathology, London, SE5 9RT
August 2014 - present
Oversight of the provision of molecular pathology and haemoglobinopathy genomics testing service:

SYNLAB LMP Laboratorio Médico Polanco Laboratorios Ruiz

UPAEP

COMPAC

- Management of a diverse team of fourteen technicians, scientists and bioinformaticians.
- Implementing change and improvements to enhance service delivery.
- Communicating with key stakeholders at all levels to advocate for the service through a range of media, including: talks, presentations, meetings, workshops and reports.
- Analysis, interpretation and reporting of genetic test results to meet Key Performance Indicators and turnaround time targets.
- Development and implementation of new diagnostic tests including establishing protocols to ensure quality, accuracy and safety.
- Deputising for Head of Department (Molecular Pathology).
- Recruitment and training of new staff and continuing professional development for all staff.
- Troubleshooting laboratory and staffing issues in a high-pressure environment
- Maintaining laboratory quality management systems to exacting standards
- Operational laboratory lead for the 100,000 genomes project delivery at King's College Hospital

Clinical Scientist
Institute of Cancer Research, Translational Genomics Laboratory, 15 Cotswold Rd, Sutton,
London, Surrey SM2 5NG
January 2013-August 2014
Establishment of an ISO accredited diagnostic laboratory for provision of NGS based testing for inherited cancer syndromes

- Design, validation and implementation of all diagnostic tests, including authorisation of reports
- Joint Quality Manager - establishing and maintaining a quality management system fulfilling the ISO15189 standards

Pre-registration Clinical Scientist
GSTS Pathology, The DNA Laboratory, Guy's Hospital, London, SE1 9RT
October 2011 to December 2012
Providing high quality molecular genetics testing service to clinicians:

- Receipt of samples
- Allocation of testing required based on the referral information
- Liaising with technical staff carrying out the laboratory work
- Analysis of results using various molecular genetics software; both windows and linux based
- Quality control of data
- Interpretation and reporting of results
- Responsible for the Cystic Fibrosis and Glycogen Storage Disease testing service.

Trainee Clinical Scientist
GSTS Pathology, The DNA Laboratory, Guy's Hospital, London, SE1 9RT
October 2008 - October 2011
Undertake clinical scientist training in molecular genetics and gain the knowledge, skills and experience required to practise as a clinical scientist

- Working for a period of time on each of the core diseases, which spanned a variety of inheritance patterns, disease mechanisms, mutation types, molecular testing, and referral rates. During my time working on each disease, I undertook extensive study on that particular topic, and also ran that testing service to learn the referral types, testing involved, and analysis and reporting of results.
- Undertake an extended research project focused on application of next generation sequencing to the molecular diagnosis of Glycogen Storage Diseases

Advanced Research Assistant
The Wellcome Trust Sanger Institute, Genome Campus, Hinxton, Cambridgeshire, CB10 1SA
September 2005 - September 2008
Supervising a small team of technicians producing various types of sequencing libraries for Illumina, 454, and Solid sequencers; including standard paired end, mate pair, small RNA, and mRNA libraries

- Trouble shooting, allocation of workload, training staff in new techniques, and had a general responsibility for the quality of libraries produced by the whole team
- Development of new procedures and pipelines for sample preparation and sequencing during the transfer from Sanger sequencing to new technologies.
- Producing specialist subclone sequencing libraries, such as short insert and transposon insertion libraries, YAC and BAC libraries, and fosmid libraries
- Working with the sequencing finishing teams on gap closure PCR

Publications:

- Brewin J, Clark B, Smith F, Parkin N, Nardo-Marino A, Gassas A, Deheragoda M, Hind J, Height S, Chakravorty S, Dhawan A, Rees D. Loss of Function SPTA1 Variants Causes Neonatal Liver Failure and Fetal Anemia. Am J Hematol. 2025 Sep;100(9):1682-1686. doi: 10.1002/ajh.27751. Epub 2025 Jun 26. PMID: 40569047.

- Mansour S, Josephs KS, Ostergaard P, Gordon K, Van Zanten M, Pearce J, Jeffery S, Keeley V, Riches K, Kreuter A, Wieland U, Hägerling R, Ratnam L, Sackey E, Grigoriadis D, Ho B, Smith F, Rauter E, Mortimer P, Macallan D. Redefining WILD syndrome: a primary lymphatic dysplasia with congenital multisegmental lymphoedema, cutaneous lymphovascular malformation, CD4 lymphopaenia and warts. J Med Genet. 2023 Jan;60(1):84-90. doi: 10.1136/jmedgenet-2021-107820. Epub 2021 Dec 16. PMID: 34916230; PMCID: PMC9811088.

- King-Robson J, Marshall J, Smith F, Willoughby L, Mansour S, Sztriha L. Ataxia-Pancytopenia Syndrome due to a de Novo SAMD9L Mutation. Neurol Genet. 2021 Mar 24;7(3):e580. doi: 10.1212/NXG.0000000000000580. PMID: 33884299; PMCID: PMC8054956.

- Nzelu D, Shangaris P, Story L, Smith F, Piyasena C, Alamelu J, Elmakky A, Pelidis M, Mayhew R, Sankaran S. X-linked sideroblastic anaemia in a female fetus: a case report and a literature review. BMC Med Genomics. 2021 Dec 20;14(1):296. doi: 10.1186/s12920-021- 01146-z. PMID: 34930268; PMCID: PMC8686580.

- Lazana I, Mohamedali A, Smith F, de Lavallade H, McLornan D, Raj K. Uniparental disomy (UPD) of a novel bisphosphoglycerate mutase (BPGM) mutation leading to erythrocytosis. Br J Haematol. 2021 Jan;192(1):220-223. doi: 10.1111/bjh.17223. Epub 2020 Nov 20. PMID: 33216349.

- Park H, Haller J, Smith F, Parkin N, Lythe T, Zoeller RA, Chakravorty S. Attenuation of Hemolysis Due to Glucose-6-Phosphate Isomerase Deficiency With Ketogenic Diet - A Case Report. Hemasphere. 2020 Jan 3;4(1):e328. doi: 10.1097/HS9.0000000000000328. PMID: 32072144; PMCID: PMC7000473.

- Creasey T, Biss T, Lambert J, Smith F, Clark B, Carey P. Pyridoxine-sensitive X-linked “sideroblastic” anaemia in the absence of ring sideroblasts - molecular diagnosis. Br J Haematol. 2018 Jan;180(1):10. doi: 10.1111/bjh.14909. Epub 2017 Nov 5. PMID: 29105823.

- Smith F, Hopton S, Dallabona C, Gilberti M, Falkous G, Norwood F, Donnini C, Gorman GS, Clark B, Taylor RW, Kulasekararaj AG. Sideroblastic anemia with myopathy secondary to novel, pathogenic missense variants in the YARS2 gene. Haematologica. 2018 Dec;103(12):e564-e566. doi: 10.3324/haematol.2018.194464.Epub 2018 Jul 5. PMID: 29976739; PMCID: PMC6269300.

- Cutts A, Vavoulis DV, Petrou M, Smith F, Clark B, Henderson S, Schuh A. A method for noninvasive prenatal diagnosis of monogenic autosomal recessive disorders. Blood. 2019 Oct 3;134(14):1190-1193. doi: 10.1182/blood.2019002099. Epub 2019 Aug 23. PMID: 31444163; PMCID: PMC6888147.

- Clark, B., Shooter, C., Smith, F., Brawand, D., Steedman, L., Oakley, M., Rushton, P., Rooks, H., Wang, X., Drousiotou, A., Kyrri, A., Hadjigavriel, M., Will, A., Fisher, C., Higgs, D. R., Phylipsen, M., Harteveld, C., Kleanthous, M. and Thein, S. L. (2016), Beta thalassaemia intermedia due to co-inheritance of three unique alpha globin cluster duplications characterised by next generation sequencing analysis. Br J Haematol. doi:10.1111/bjh.14294

- Ghurye RR, Sundaram K, Smith F, Clark B, Simpson MA, Fairbanks L, Adhya Z, Mufti GJ, Marsh JCW, Ibrahim MAA. Novel ADA2 mutation presenting with neutropenia, lymphopenia and bone marrow failure in patients with deficiency in adenosine deaminase 2 (DADA2). Br J Haematol. 2019 Aug;186(3):e60-e64. doi: 10.1111/bjh.15896. Epub 2019 Mar 28. PMID: 30924144.

- Dlamini N, Josifova D, Paine S, Wraige E, Pitt M, Murphy A, King A, Buk S, Kulkarni G, Smith F, Abbs S, Sewry C, Jacques T, Jungbluth H. Clinical and neuropathological features of X-linked spinal muscular atrophy (SMAX2) associated with a novel mutation in the UBA1 gene. Neuromuscular Disorders 2013 May;23(5):391-8.

- Cullup T, Kho A, Dionisi-Vici C, Brandmeier B, Smith F, Urry Z, Simpson MA, Yau S, Bertini E, McClelland V, Al-Owain M, Koelker S, Koerner C, Hoffmann G, Wijburg F, ten Hoedt A, Rogers C, Manchester D, Miyata R, Hayashi M, Said E, Soler D, Kroisel P, Windpassinger C, Filloux F, Al-Kaabi S, Hertecant J, Del Campo M, Buk S, Bodi I, Goebel H, Sewry C, Abbs S, Mohammed S, Josifova D, Gautel M, Jungbluth H. Recessive mutations in KIAA1632 cause Vici syndrome, a multisystem disorder with defective autophagy. Nature Genetics 2013 Jan;45(1):83-7.

- Paterson S, Vogwill T, Buckling A, Benmayor R, Spiers AJ, Thomson NR, Quail M, Smith F, Walker D, Libberton B, Fenton A, Hall N, Brockhurst MA. Antagonistic coevolution accelerates molecular evolution. Nature. 2010 Mar 11;464(7286):275-8.

- Wilkinson P, Waterfield NR, Crossman L, Corton C, Sanchez-Contreras M, Vlisidou I, Barron A, Bignell A, Clark L, Ormond D, Mayho M, Bason N, Smith F, Simmonds M, Churcher C, Harris D, Thompson NR, Quail M, Parkhill J, Ffrench-Constant RH. Comparative genomics of the emerging human pathogen Photorhabdus asymbiotica with the insect pathogen Photorhabdus luminescens. BMC Genomics. 2009 Jul 7;10:302.

- Liti G, Carter DM, Moses AM, Warringer J, Parts L, James SA, Davey RP, Roberts IN, Burt A, Koufopanou V, Tsai IJ, Bergman CM, Bensasson D, O'Kelly MJ, van Oudenaarden A, Barton DB, Bailes E, Nguyen AN, Jones M, Quail MA, Goodhead I, Sims S, Smith F, Blomberg A, Durbin R, Louis EJ. Population genomics of domestic and wild yeasts. Nature. 2009 Mar 19;458(7236):337-41. Epub 2009 Feb 11.

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- Pain A, Böhme U, Berry AE, Mungall K, Finn RD, Jackson AP, Mourier T, Mistry J, Pasini EM, Aslett MA, Balasubrammaniam S, Borgwardt K, Brooks K, Carret C, Carver TJ, Cherevach I, Chillingworth T, Clark TG, Galinski MR, Hall N, Harper D, Harris D, Hauser H, Ivens A, Janssen CS, Keane T, Larke N, Lapp S, Marti M, Moule S, Meyer IM, Ormond D, Peters N, Sanders M, Sanders S, Sargeant TJ, Simmonds M, Smith F, Squares R, Thurston S, Tivey AR, Walker D, White B, Zuiderwijk E, Churcher C, Quail MA, Cowman AF, Turner CM, Rajandream MA, Kocken CH, Thomas AW, Newbold CI, Barrell BG, Berriman M. The genome of the simian and Human malaria parasite Plasmodium knowlesi. Nature. 2008 Oct 9;455(7214):799-803.