USA Laboratory Tours, October 2023.

Rachel Carling

California Genetic Disease Screening Program, State of California Department of Health Service, Richmond Laboratory. Hosted by Rajesh Sharma, Chief, Genetic Disease Laboratory.

The California Newborn Screening Programme is the largest in the United States, screening 450,000 babies for 80 different disorders each year. DBS specimens are collected 12-48 hours after birth and sent to one of the five state-contracted laboratories. Screening is mandatory although parents can opt out for religious reasons. The programme has approximately 3,000 condition suspected results each year, of which around 900 are subsequently confirmed as true positives.

The Genetic Disease Laboratory co-ordinates the newborn screening programme for the state. The first-tier testing for all disorders is out-sourced to five Newborn and prenatal screening (NAPS) laboratories. The demographic and analytic values are transferred from the NAPS labs to the GDL via the Screening Information System (SIS). The GDL is responsible for the quality review and then releases the final results to the SIS. When new disorders are introduced (recent examples include SMA, Pompe and MPS 1) the GDL are also responsible for the validation and initial roll out of the new screening test. Once the test is well established and robustness of the assay has been demonstrated, responsibility for routine screening will be transferred to the NAPS labs.

Centre for Disease Control and Prevention (CDC). Hosted by Dr Joanne Mei, Chief, Newborn Screening Quality Assurance Program, Newborn Screening and Molecular Biology Branch.

Special thanks to Joanne Mei for arranging my visit and helping me navigate the security issues! Once I had gained access to CDC , I had a tour of the Molecular Biology laboratories and an opportunity to discuss the ins and outs of SCID screening with Rachel Lee, Franics Lee and Stanimila Nikolova. This was an invaluable and highlighted the benefits of having molecular biologists directly involved with the screening programme. For countries who are thinking of expanding the molecular aspects of their screening programmes in future, a similar approach would be recommended. Seeing the Molecular labs also emphasised the importance of having appropriate workflows set up for molecular testing.

I then went to the NSQAP labs and met Dr Kostas Petritis. The NSQAP lab prepares huge quantities of DBS QC material and it was fascinating to see some of this in progress. The difference in the scale of the DBS preparation was the first thing that struck me, and my take home message was that preparation of DBS material in my laboratory definitely needs to be improved! In the mass spec lab, there were lots of interesting projects underway including development of a screening method using LC-MS/MS rather than FIA and identification of N-acetyl-tyrosine as an indicator of TPN.

Timothy J. Garrett Laboratory, University of Florida, Gainsville. Hosted by Professor Tim Garrett.

My last lab visit was to the Timothy J. Garrett Laboratory, in the Department of Pathology, Immunology & Laboratory Medicine at the University of Florida. The research in the Garrett lab

involves the application of mass spectrometry to different areas of clinical research, including metabolomics. Tim's lab had all the interesting kit – ion mobility MS, time of flight etc and have been doing quite a lot of work on lipidomics and developing iqDBS. iqDBS involves internal standardisation of metabolites at the point of collection and has potential application to both screening and monitoring. Like any good academic lab, they also have unique ways of supporting elderly instruments…!

Big thanks to Don Chace and Fred Meindel for looking after me on my visit, ensuring I was exposed to plenty of American culture and the road trip from Atlanta to Florida – it was fun!