

National Healthcare Science School of Genetics Newsletter

Issue 3 October 2010

- | | |
|---|----------|
| 1. National Healthcare Science School of Genetics | ■ Page 1 |
| 2. Genetics Education and Training Board | ■ Page 2 |
| 3. Curriculum Development | ■ Page 2 |
| 4. Assessment | ■ Page 3 |
| 5. Pathology rotations | ■ Page 4 |
| 6. Update from the University of Nottingham | ■ Page 6 |
| 7. Train the Trainer | ■ Page 6 |
| 8. Evaluation Report | ■ Page 7 |
| 9. Modernising Scientific Careers Update | ■ Page 7 |
| 10. Registration and Regulation | ■ Page 7 |
| 11. Experiential Learning – UNIQUE Creche | ■ Page 7 |
| 12. Human Genome Strategy Group | ■ Page 8 |
| 13. Sarah Warburton | ■ Page 8 |

Modernising Scientific Careers



1. National Healthcare Science School of Genetics

Note from the Head of School



We have had a very busy year with the training programme being accepted by the Strategic Health Authority's as the modernised programme and we are now running 2 years of modernised training. One of the most important aspects of this programme is the feedback from Warwick University, from the training departments and from the trainees themselves. This is very valuable and we do make sure that it is all incorporated. Of particular importance recently was the feedback on the pathology rotations and this is influencing how these will be organised in the future – thank you to all the trainees who took so much time to formulate such an excellent document. We are also making excellent progress with the bioinformatics training development. Please don't hesitate to contact me if you want to talk about anything.



Val Davison
Head of National Healthcare Science School of Genetics

School Board

The School Board held its inaugural meeting on 19th April 2010. The Board has representation from key partners and includes nominees from the Genetics Education and Training Board of the Professional Bodies and the Royal College of Pathologists. The role of the School Board is to advise and approve the implementation of the modernised training programme for genetics, promote excellence in all aspects of the work of the School, oversee the quality, integrity and standardisation of the programme, receive reports on the quality of the training programmes and provide advice about possible improvements.

The School Board formally reports to the Modernising Scientific Careers England Implementation Board and provides updates to the Healthcare Science Education and Training subcommittee of Medical Education England (MEE).

The Board meets on a quarterly basis. Additional meetings may be called under exceptional circumstances where there are urgent issues that need discussion or approval. A subgroup of the Board gives advice on and reviews and updates the curricula (Curriculum Advisory Group).

New Releases

The School will shortly be releasing a number of documents that will outline the structure and function of the School, provide further guidance to departments delivering the training programme and further information to support trainees, they include;

- Structure & Function document
- Governance structure
- Quality Assurance Framework
- Revised PTP training manual
- Revised STP training manual
- Revised online assessment user guide
- Communications strategy

Work is also underway to develop content for a website link for the School where all documentation and information can be accessed online. Further information on this will be available shortly.

Contact Us

Access to documentation relating to the training programme or National School is available by emailing the address below;

Genetics.NWD@westmidlands.nhs.uk

2. The Genetics Education and Training Board (GETB)

The Association of Clinical Cytogeneticists (ACC) and the Clinical Molecular Genetics Society (CMGS) have now formed a joint education committee called the Genetics Education and Training Board. This is chaired by David Bourn from Newcastle and the vice Chair is Gordon Lowther from Glasgow. We will have reports from this committee in this newsletter in future.

3. Curriculum Development

As part of the ongoing review of the curriculum the School undertakes an evaluation of each module as it is completed to ensure that lessons can be learned and good practice shared. Trainees and departments receive an evaluation questionnaire and the results are collated and reviewed by the Curriculum Advisory Group.

STP Module 1 Review

To date one review has been undertaken of the first STP module and the School is currently in the process of reviewing the first PTP module.

The predominant learning taken from the review is that greater clarity is required for the competencies to ensure that evidence and level of competency is more defined. It was also recognised that greater clarity around the expectations of the MSc in Clinical Sciences is needed as is the need to recognise previous experience of trainees undertaking the programme.

These messages have been shared and reviewed by the Curriculum Advisory Group and fed back to the Curriculum Development Group for Modernising Scientific Careers.

As a result the Healthcare Science Practitioner and Healthcare Scientist training manuals have been reviewed in detail and changes made to meet the needs outlined in the evaluation.

New versions of both the manuals were released at the end of September 2010.

The School would like to thank everyone for taking such active involvement in these reviews. All comments and recommendations are making a real difference and we hope that changes made will enhance the experience for both trainees and trainers alike.

Bioinformatics

The School hosted a meeting at Nowgen in July 2010 at which members of the professional bodies bioinformatics working party, members of NGRL, DH, and other interested professionals met to discuss the future direction and integration of the discipline into the Genetics curriculum. Work is ongoing to develop the following;

- Strategy for integrating Bioinformatics in Genetics and potentially the establishment of a virtual Bioinformatics Institute.
- Work based competency training for existing staff.
- Bioinformatics modules to go into both the Practitioner and Scientist training programmes.

The DH is funding this work through the School. The next meeting will be in October – anyone who is not involved and would like to be please contact Val asap.

Further information will be made available as this work develops.

Higher Specialist Training

A national project commenced in September 2010 to develop the HSST programme for Modernising Scientific Careers led by Janet Monkman. This will look at the training necessary in addition to FRCPATH

The introduction of MSC means that a new examination for FRCPATH in genetics will be introduced in 2013. This exam will cover aspects of both cytogenetics and molecular genetics as well as a management

component as in the existing exams. A curriculum for this exam will be in place by Spring 2011. The current discipline specific examinations will continue for several years after this date to allow existing trainees to complete the exam. The Genetics SAC will confirm the date of the last discipline specific exams when they meet in December.

Fiona Macdonald

Chair, Genetics examinations panel

September 2010

Communications Training

Viv Parry, BBC journalist, radio presenter and Co Chair of the National School Board delivered some training for the Healthcare Scientists on communication and media skills in September 2010.

The training was delivered to reinforce the role of Healthcare Scientists as ambassadors for the profession and is additional to the agreed curriculum.

Industry Visits

The National School is pleased to be able to announce that a visit will be arranged for the Healthcare Science Practitioners to visit Oxford Gene Technology (OGT).

<http://www.ogt.co.uk/>

The purpose of the visit is to expose trainees to new technologies that will be critical to future service delivery in the NHS and to provide an understanding of the services available in private industry.

4. Assessment

Online Assessment Tool

As a result of the recent review of the training manuals and feedback received on the system, the online assessment tool is undergoing some significant content changes to ensure that it remains in line with the curriculum and to improve its functionality.

The School is working closely with the supplier in the development of a third version of the tool. It is hoped that this version will be released in October 2010 and will be accompanied by updated user guides.

Significantly, version 3 will also provide reporting functionality for trainees, trainers, Heads of Department and the National School.

End of Year and End of Programme Assessments

The Curriculum Advisory Group, in line with the national assessment programme for MSC has agreed that practitioner trainees will undergo both end of year progress reviews and end of programme formal exit assessments.

The end of year progress reviews are intended to provide trainees and departments with an opportunity to evaluate progress and consolidate learning but will not contribute to the final assessments. As such the outcome of the assessment will not impact on trainee progression but may highlight areas where further learning is required. The end of year progress assessment for Healthcare Science Practitioners will take place on 19th October 2010.

Healthcare Scientists will undergo an end of year progress assessment at the end of year 2.

Both Healthcare Scientists and Healthcare Science Practitioners will undergo a formal summative assessment at the end of their final year of training.

Further details on the end of year and end of programme assessments will be made available to departments and trainees shortly.

Multi Source Feedback Assessment (Healthcare Scientists)

As part of the agreed curriculum for Healthcare Scientists, all trainees are required to undertake 2 separate multi source feedback (MSF) assessments. As trainees have been in post for 12 months departments are preparing to undertake the first of these assessments in November 2010 with feedback reviews expected by early December.

MSF is an externally applied assessment which provides a sample of attitudes and opinions of colleagues on the technical performance and professional attitude of a trainee. The tool helps to provide data for reflection on performance and gives useful feedback for self evaluation.

5. Pathology Rotations

From March to June 2010, all Healthcare Scientists undertook their first pathology rotations. Trainees were placed in a variety of pathology disciplines ranging between Reproductive Science, Clinical Biochemistry, Histopathology and Immunology dependent upon local availability.

Trainees have reported the rotation to be a valuable experience in understanding the role of pathology in the delivery of healthcare and the intricate relationship these services have with Genetics. The trainees also wrote a valuable document to the school which is influencing the development of this part of the programme.

Rebecca Franses and Celia Brown at the Regional Genetics Service Laboratories at Great Ormond Street Hospital describe their experience in the Chemical Pathology Laboratory;

"I've got to be honest. The thought of spending three months doing biochemistry did not fill me with excitement: my memories of biochemistry involve never-ending first-year lectures, usually timetabled for 9am the

morning after student night and given by a lecturer who seemed to think that shorts, socks and sandals were acceptable attire. Added to that, our fellow trainees from other labs were being sent to departments such as reproductive medicine, where they could actually be involved in the creation of life itself – surely much more interesting than analysing urine samples! Fortunately, the reality of the biochemistry laboratories was very different and provided a useful experience of the structure and operation of another clinical laboratory department – and all without going near a single semen sample....

Our first four weeks in the Great Ormond Street Chemical Pathology department were spent rotating around the four sections – neonatal screening, routine testing, the enzyme laboratory, and the metabolic section. Each of these was a very different environment and had different links to our genetics training. The neonatal screening and routine laboratories, for example, have a huge number of samples to deal with on a daily basis and so the degree to which everything has to be co-ordinated in order to get the samples processed and the results sent out within turnaround time is remarkable, as is the level of automation involved. In contrast, the enzyme and metabolic laboratories carry out more specialist tests and have a smaller volume of samples and more manual lab work. In relation to genetics, it was very interesting to see how diseases we are familiar with – such as CF and congenital adrenal hyperplasia in neonatal screening, and lysosomal storage disorders in the enzyme section – were tested for biochemically, and we were able to improve our knowledge and understanding of these conditions.

After rotating round the four sections, our next two months involved carrying out mini projects in the metabolic section, which are summarised below. Though daunting at first, as both projects used high pressure liquid chromatography (HPLC) – a technique neither of us had ever used before! – the

opportunity to gain practical laboratory experience was very beneficial.

Urinary tetrasaccharide analysis for diagnosis and monitoring of Pompe disease

Pompe disease is a lysosomal storage disorder, which is inherited in an autosomally recessive manner and is caused by mutations in the gene encoding acid alpha-glucosidase. The aim of this project was to develop an HPLC-based method to test for the presence of glucose tetrasaccharide (Glc4) in urine, which is a biochemical marker of the disease. This would provide supportive evidence in diagnosis and allow monitoring of treatment response in patients undergoing enzyme replacement therapy. In order to do this, we used a standard of pure Glc4 to optimise detection by adjusting parameters such as mobile phase concentration, column temperature and detection voltage until a clear peak was produced within an acceptable time-frame for routine laboratory analysis. Urine samples from both Pompe disease patients and controls were then run, to show that it was detectable in patients but was absent in controls, and that none of the other components of normal urine interfered with its detection. Further experiments identified an internal standard and established recovery rate, and a doubling dilution demonstrated a linear relationship between Glc4 concentration and peak area within the ranges found in Pompe disease patients. This enabled accurate quantification for reporting to clinicians. The next steps will be to establish reference ranges for different patient groups by testing a large number of both control and patient urines. Once this is done, the method can be rolled out and it is hoped that it can be offered as a new service in the very near future!

Optimisation of branched chain amino acid monitoring for Maple Syrup Urine Disease

Maple syrup urine disease (MSUD) is a rare autosomal recessive disorder resulting in defective metabolism of the branch chain amino acids; leucine, isoleucine and valine.

It is caused by mutation in any one of the four genes which produce proteins that make up the branch chain oxo acid dehydrogenase multi-enzyme complex. It presents in the neonatal period with failure to feed, neurological deterioration and the presence of sweet-smelling urine. MSUD leads to severe mental and physical problems if left untreated, however management with a protein restricted diet and supplements can allow patients to lead a relatively normal life.

Management requires careful monitoring of plasma branch chain amino acid levels, which is carried out by testing blood spots via HPLC. Currently the metabolic team analyse the MSUD patient blood spots on the HPLC machine using an analysis method which takes 92 minutes per sample to run. My task was to reduce this analysis time considerably by optimising the method to elute the branch chain amino acids off the column more quickly whilst retaining the resolution of the peaks so that identifications and quantification would remain accurate. I used standard blood spot samples spiked with known concentrations of the branch chain amino acids to experiment with. The process of optimisation involved making sequential changes to the method via modification of the solvent gradient flowing through the column and the amount of time spent at each gradient step. After 16 sequential alterations I had managed to produce a 22.5 minute method which accurately measured branch chain amino acid levels in test samples. After further in house validation this test can be rolled out and hopefully result in the quicker turnaround of results to dieticians and therefore better management for patients!

Overall, both projects were very successful and we feel fortunate to have been able to have a real impact on the service of the Chemical Pathology laboratory - and hopefully patient care – during our short time in the department. We would like to thank Professor Simon Heales for all his work in making the rotation possible and for supervising us whilst we were there, as well as the rest of the Chemical Pathology

department, and we look forward to our next rotation!"

Rebecca Franses and Celia Brown
North East Thames Regional Genetics
Service Laboratory
Great Ormond Street Hospital NHS Trust,
London

The School would like to thank all the Healthcare Scientists who contributed to the production of a helpful and informative feedback document on their experiences and recommendations for future rotations. The School has shared this information with the national Modernising Scientific Careers team and is considering the recommendations made for the next rotations due in November 2010.

6. Update from The University of Nottingham

The trainees have completed the MSc modules on Healthcare Science I (Biochemistry and Reproductive Science), and Genetics of Infertility and Disorders of Sexual Differentiation. Course material is presented in the form of problem-based learning, with the trainees discussing the cases within ATutor - the online learning environment used to deliver the material. It's great to see that the discussions within the forums have been so thorough and engaging, with discussions for many cases extending over several pages.

MSc Clinical Science (Clinical Genetics)

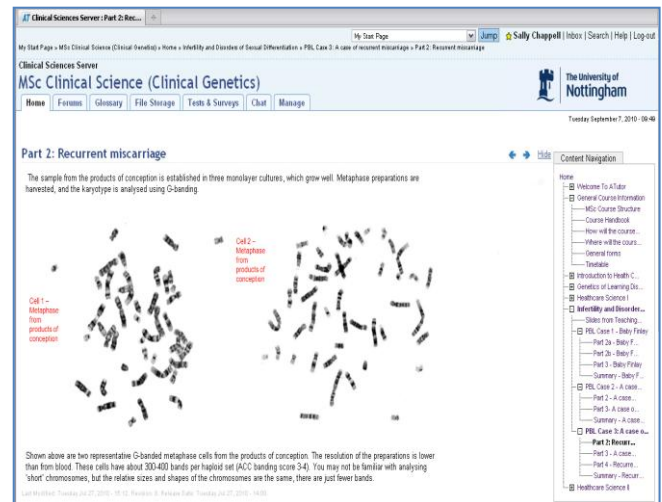
Home Forums Glossary File Storage Tests & Surveys Chat Manage

Back To: Forums | Genetics of Infertility and Sexual Differentiation | New Thread

Genetics of Infertility and Sexual Differentiation

Topic #†	Replies #†	Started By	Last Comment #†
New Post Group A- PBL 3- A case of... (Page: 2 13) (Subscribe)	20	Jonathan Ball	Fri Aug 13 15:02 ↑ ↓ ↻
New Post Group B- PBL case 3- A c... (Page: 2 13) (Subscribe)	20	Jonathan Ball	Fri Aug 13 09:42 ↑ ↓ ↻
New Post Group A- PBL2- A case of... (Page: 2 13) (Subscribe)	20	Sally Chappell	Wed Aug 4 10:33 ↑ ↓ ↻
New Post Group B- PBL2- A case of... (Page: 2 13 14) (Subscribe)	35	Sally Chappell	Mon Jul 26 15:24 ↑ ↓ ↻
New Post Group A- PBL1- Baby Finlay (Page: 2) (Subscribe)	17	Sally Chappell	Fri Jun 25 14:00 ↑ ↓ ↻
New Post Group B- PBL1- Baby Finlay (Page: 2) (Subscribe)	13	Sally Chappell	Fri Jun 25 13:27 ↑ ↓ ↻

Each PBL case gives the trainees the opportunity to think about real-life cases, and interpret data for themselves.



The first cohort of trainees are about to start Healthcare Science II (Immunology and Haematology), visiting Nottingham at the beginning of October. We are also looking forward to meeting the second cohort of trainees, who are starting the MSc course and training program in October.

7. Train the Trainer

The train the trainer programme has been agreed and is being rolled out from September 2010 – January 2011.

Departments across England and Wales taking MSc trainees for the first time in 2010 have been invited to attend together with departments with pilot trainees.

The programme will cover MSc, the Genetics curriculum, the assessment tools and online assessment system and understanding concepts in education for health professionals.

The National School will also be delivering a one day train the trainer programme for Scotland Genetics departments who are taking Healthcare Science Practitioner trainees for the first time this year.

8. Evaluation Report

The first report for the external evaluation was received by the MSC team in April 2010.

Significant lessons have been learned around standardisation, communication, timescales and recruitment processes to which both the School and the national MSC team have taken note and implemented changes.

Work continues to evaluate the role of the School and to identify any further learning as the programme progresses.

A second report is expected in October 2010.

9. MSC Update

Following the launch of Modernising Scientific Careers: The England Action Plan, in April this year, SHAs hosted a series of MSC Road Shows in July. It was the first opportunity since the launch of MSC this spring, for over 1000 healthcare scientists, SHA workforce planners and education commissioners, Trust HR managers and Higher Education Institutions to hear about the progress to date on MSC and how SHAs would be shaping its implementation. The morning sessions consisted of presentations by the Department of Health's Chief Scientific Officer's MSC team, and a panel Q and A. In the afternoon, SHAs ran their own workshops on different aspects of implementing MSC locally. Delegates chose from a number of different workshops, including workforce planning and re-profiling, training and assessment in the workplace, and healthcare scientists as leaders. NHS South East Coast will be holding its roadshow on 29 October 2010.

The latest version of the CSO bulletin is available at the following address;

http://www.dh.gov.uk/en/Aboutus/Chiefprofessionalofficers/Chiefscientificofficer/DH_118274

10. Registration and Regulation

Work is continuing with regulation and registration and as soon as there is some news we will let you know. However it is important to remember that there will be a system in place for registration when trainees qualify.

11. Experiential Learning – UNIQUE Creche

UNIQUE...but not alone



UNIQUE is a self-funded charity which provides information and support for families affected by rare chromosomal disorders. Every year, UNIQUE organise conferences and meetings for specific abnormalities. These events provide an invaluable opportunity for families and medical professionals from all over the world to get together and discuss the latest advances or share experiences.

Recently, a 3-day meeting was held in Oxford for parents of children affected by 2q37 deletions. The common features of children with 2q37 deletions are mild-to-moderate learning disabilities, short stature and joint hypermobility. Some of the more severely affected children had autistic features but most were mildly affected. It later emerged that until this weekend, not one of the families had ever met another family with a child affected by a 2q37 deletion before. As a result the families have set up their own support group including a Facebook page.

As the adults were attending lectures, the children (affected and siblings) were cared for by a specially provided crèche. The crèche was run by a group of professionally trained care assistants and trainee HCS from London, Birmingham and Newcastle. The look on the trainee HCS faces as the doors opened to release a gaggle of children full of excitement (and sugar) was priceless – the words ‘rabbits’ and ‘headlights’ spring to mind!

The children were wonderful! Our day was spent supervising paints, sandpits, train sets, Nintendos...I don't know who had more fun, the trainees or the children. One 8-year old had a particularly brilliant character. When playing shop she politely told us that she would like 2 apples, 1 carton of milk and 50g of tobacco!! Trying hard not to laugh we proceeded to ask if she had any ID as selling tobacco to a person under 16 years old was a criminal offence and could put us out of business.

The meeting was a great success as proven by the excellent feedback:

“The value of the information shared and the opportunity to attend was priceless...”

“We think it was amazing the children were so well occupied and looked after, as were families.”

The enthusiasm of the UNIQUE staff and their commitment to helping families all over the world affected by chromosomal disorders is truly priceless. We're already looking forward to the next event in October. For further information about UNIQUE please visit:

www.rarechromo.org

**Louise Johnston, Jessica Gabriel,
Institute of Human Genetics, The
Newcastle Upon Tyne Hospitals NHS
Foundation Trust**

12. Human Genome Strategy Group

The Human Genome Strategy group was established this year to look at high level strategy around the delivery of genetics services, research and innovation including IT and Education. We have an input into this committee which will be very influential in designing the future for genetics and we will report progress in this newsletter.

13. Sarah Warburton

Sarah will be retiring shortly from her valuable role as national trainer – on behalf of the genetics community we would like to thank Sarah for all her input and help and wish her all the very best for the future.

Next Issue

- **National Healthcare Science School of Genetics**
- **Curriculum Development**
- **Assessment**
- **Update from the University of Nottingham**
- **Evaluation Report**
- **Modernising Scientific Careers Update**
- **Registration and Regulation**