Example CPD records.

**Example of reflective CPD record. Continuing professional development (CPD) documentation form**

| **Date of CPD activity** | **Number of hours** | | **Type and title of CPD activity e.g. journal review, study day** | **Reflection and implications for practice**<br>**What have I learnt?**<br>**How did I use this in practice?** |
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| | **External** | **Internal** | | |

Example CPD records.

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| 15 January 2014 | 6 | | Conference – RAPID project dissemination meeting. UCL, London. | This was a very informative meeting, with up to date information about the progress of non-invasive prenatal testing. Brigitte Faas of the Netherlands spoke about use of SNP arrays in NIPT. They conducted a SNP array on maternal serum samples in 565 pregnancies. In most of these, the fetus had an ultrasound anomaly. In 75, the fetus had an inherited condition diagnosed using the array, in another 42 clinically relevant chromosomal anomalies were found. Of importance, only half of these would have been detected using a traditional karyotype, which seems to indicate that SNP arrays could be used in preference to karyotyping in cases where fetal abnormalities are detected on ultrasound. This research is now being extended to study a further 1000 pregnancies where the fetus has a structural anomaly in a study called PAGE (prenatal diagnosis of genomes and exomes). As I teach both nurses and midwives, maintaining my knowledge of what is on the horizon in prenatal testing is important.<br>Another relevant point to come from the talks at this meeting was the limitation of using NIPT where the mother is affected by the same condition. For example, NIPT is being used to detect achondroplasia in the fetus, but if the mother also has achondroplasia, it is not possible to detect. This is because the fetal DNA is not actually separated from maternal DNA, the test is focussed on finding differences between the maternal and fetal genome. Because of this same point, CF can only be excluded in the fetus if parents have different mutations: the test is for the paternal mutation.<br>In another talk on NIPT for trisomy 21 by Faas, it was emphasised that the fetal fraction was crucial in ensuring accuracy of results. Because the maternal body mass index affects the fetal fraction (lowering it), such test will not be so reliable in obese women. This will be an important issue in counselling mothers, but could be very tricky to phrase sensitively! This entire meeting was enthralling, as it included so much that is new about our field. My research and teaching both concern prenatal testing so came away with better information for my students and some ideas as to the type of research needed in future to introduce these tests appropriately. |

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| 6 December 2013 | 5 | | Human Reproduction Study Day. British Sociological Association. Plymouth, UK | During the meeting there was a presentation by a medical doctor who had been training as an obstetrician. She had experienced a stress disorder and had discontinued her training. She has a series of paintings she had done to represent her experience and feelings. These were powerful and expressive. She talked about the conflict she felt in having to perform abortions while feeling that she could not herself have had one. She agreed and supported patient choice but it was hard to perform abortions. She said she had sought to discuss the pressure and distress she felt with It was obvious that senior colleagues but felt unheard. Eventually she had become ill. This reinforced the pressure that we as professionals can feel to offer choices that conflict with our own beliefs and values. An interesting interaction occurred after the presentation. One of the audience said that she objected to this woman saying she did not personally feel she could have an abortion, identifying herself as a feminist. I was very shocked as the presenter had allowed herself to be vulnerable and received an attack. It did strongly reinforce how unacceptable it can be today to say that you don't feel entirely comfortable with termination. This has resonance for genetic counsellors as well, who may have to counsel 'neutrally' when they have values and beliefs that conflict with the choices of others and confirms the need for supervision to help us deal with those pressures without jeopardising the safety of patients or ourselves. |

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| June 8-11 2013 | 30 | | ESHG conference, Paris, France | This is a very interesting conference, combining new scientific advances with a lot of educational and clinical sessions. However, two session stood out for me. In the opening plenary, Edith Heard gave a presentation on the history of research into X-chromosome inactivation, which helped me to understand the mechanisms better. However, a real eye-opener was th talk by Jean-Laurent Casanova on infectious diseases of childhood, linking these to possible single gene variations that affect our immunity to such diseases. He presented data on how the genetics of viruses and bacteria can be studied so that the evolution of the micro-organisms, and the origin of an epidemic, can be traced using genetic testing of the organisms. This was an entirely new area for me and has broadened my view of healthcare genetics. This is important as one of the areas of genetics in healthcare signalled for development by the DH is infectious diseases, and I will need to include some material about this in my teaching.<br>In a different session, Sari Lieberman from Israel spoke about a research programme in which Israeli women from the general population were offered screening for BRCA mutations. Uptake was very high, and I wondered if the approach to breast cancer screening and surgery in Israel was different to her in the UK, and how women would accept mutation screening here. Of course, the prevalence of specific mutations makes this more feasible in Israel. Elisa Houwink talked about her oncogenetics training programme for GPs in the Netherlands. This was well-resourced and they tested the effect by sending in actors to GP surgeries afterwards to see if the GP responded appropriately to a question about FH of breast cancer. We offer training courses, so it was interesting to see that the ultimate impact, i.e. on patient care, could be tested in this way, rather than simple knowledge scores etc. The framework used for the project was Kirkpatrick's framework which I was not familiar with, but have since read about and will use in future in my assessment of learning. |

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| 5, 6,7 March 2013 | 12 | | EuroGentest 2 conference, Prague, CR. | The first session focussed on NGS testing. Prof Peter Robinson spoke about genetic databases, the importance of accurate phenotyping to enable good data to be extracted/ extrapolated. As phenotypic data are recorded by professionals, an ontology is needed to ensure we all use the same language and differentiate between terms well, e.g. atrial fibrillation and muscle fibrillation. Terminology for rare diseases now being adopted by SNOMED to enable it to be used in hospital recording systems.<br><br>David Barton spoke on the European IVD (in vitro devices) regulation and the effect of this on genetics. In general, all genetic tests must be assessed before use, using a range of stringent evaluations. However, if a genetics lab in a health institution is externally accredited (to ISO 15189) they can use their own lab developed tests. I was particularly interested in the impact on direct-to-consumer (DTC) tests as I have led the work to develop guidelines on DTC for users and health professionals. DTC tests are regarded as self-applied tests and will have to adhere to a set of standards, including more stringent evaluation before marketing is allowed and provision of adequate patient information in the national language. Importantly, DTC tests are still allowed without the input of a health professional, so the guidelines we have developed are still required. The regulation applies to all tests available in the EU, even if via the Internet. |

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| 1 November 2012 | 6 | | RAPID study dissemination meeting | During this study day, the results of the RAPID project on introduction of NIPT into clinical practice were presented. I am a member of the research group, but it was useful to hear the results from other sections of the study. For example, a study was conducted to determine the cost-effectiveness of using NIPD in aneuploidy screening. Using NIPT after initial US/Biochemical screening was effective, but the costs were actually very similar to using invasive testing. However, it is likely that the cost of NIPT will reduce considerably over the next few years and it may then be feasible to offer this instead of screening and invasive testing. It was clear that while patients prefer NIPT, health economics may dictate availability as least in the short term. The same arguments do not apply for monogenic disorder or sexing for sex-linked conditions, where it is cost effective to use NIPT. These examples made me think about the need to avoid raising patient expectation at too early a stage in development of new technologies. |
| 19 September 2012 | 1 | | Study day presentation by Prof Andrew Hattersley on monogenic diabetes, Exeter. | Andrew Hattersley spoke on the development of understanding of monogenic diabetes over a 15 year period. Although I knew quite a lot about HNF1alpha, I learnt much more about neonatal monogenic diabetes, which presents in the first year of life and has been treated as Type 1 diabetes. If this history is given while taking a family tree, neonatal diabetes should be suspected. I will include this in my lectures to midwives from now on.<br>The work on birth weight of babies with particular forms of monogenic diabetes was also interesting, as those with one mutation tend to have high birth weight regardless of which parent is affected, but the birth weight is even higher if mother affected. This can help to explain why some babies of diabetic mothers have extremely high birth weight. |

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| 19 July 2012, Plymouth University | | 3 | Presentations at Plymouth University by 2 Japanese genetic nurses: Hiroko Susaka, Sachiko Mitarai. | Two Japanese nurses doing were visiting our University to undertake advanced education in genetics nursing, both were MSc genetic nursing students from St Luke's College, Tokyo. During the week they made presentations on their own areas of interest.<br>Sachiko presented on the midwifery service in Japan and how genetic is integrated into that service. Given our own universal offer of screening for Down syndrome, it was of interest to hear that screening is not universal in Japan, but dependant on the policy in each area or hospital. Also, it is usually the obstetrician who discussed screening with the woman, although Sachiko said that women found the midwives more approachable and would often ask them questions later. Where screening is offered, there is high uptake and low acceptance of an abnormal child. Hiroko's, area of interest is neurology and she spoke about care of patients with Huntington disease, CJD and MND. As Huntington disease is my main area of interest we had a good discussion after her presentation. She felt that in her institution patients with neurodegenerative conditions are given little choices in their daily activities because of communication problems and she has felt uncomfortable about this. Also, there is very little alternative to hospital care. This experience reinforced my need to have a very open mind about the way care might be offered differently in other regions or countries, and to ensure my teaching and my writing is not UK- centric unless this is appropriate. |

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| 23-26 June 2012 | 24 | | ESHG annual conference, Nuremberg, Germany. | This was such a busy conference, with the establishment of the EBMG. I was impressed by a presentation by Marilyn Li et al on the use of NGS for clinically actionable mutations. She started by revising the types of mutations involved in cancer development, including chromosome changes, copy number variation or loss of heterozygosity. However, as well as using NGS for diagnostic testing, she explained the use of genetic testing to define prognosis and for tailoring clinical management, for example use of specific carcinogenic drugs. Genetic testing can therefore be divided into diagnostic biomarkers, prognostic biomarkers and predictive biomarkers, and she gave good examples of each. As is often the case lately, I found my ideas about genetic testing challenged and broadened. As my role involves teaching a range of health professionals, it is very important that I am aware of the opportunities to sue genetics and genomics in mainstream healthcare. I will incorporate this model into my lectures on genetics to student nurses as well as specialist nurses and counsellors. |

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| 23-24 April May 2012 | 6 | | ESRC Genomics Network Confernce. Genomics in Society | This conference is a different type to those I normally attend, as it involves a lot of legal, sociological and ethical presentations on genomics. One of the most interesting talks was by Eline Bunnik of the Netherlands, who spoke about direct-to-consumer (DTC) tests. I have been working on this topic for a while as part of the EuroGentest project, as one of our deliverables is designing guidance of professionals and patients. Bunnik spoke about informed consent in this context, suggesting a 'composite' model of consent that has been developed for biobanks. The 'tiered' aspect focusses on categories of information that the patient can choose to have, the layering differentiates between essential and non-essential information staging enables the consent process to occur over time, with opportunities for patients to alter their requirements. Although I see the point of approaching this in three ways, the operationalization of this model seems quite challenging…..and I am not sure how companies will address the needs for information and decision making support for patients. However, this has given me food for thought regarding the guidance we prepare for patients and families as part of our EuroGentest project work.. |
| 19 and 20 February 2012 | 16 | | EuroGentest2 / Techgene annual scientific meeting, Nijmegen, Netherlands | During this meeting I heard a talk by Jaris Veltman on the use of NGS to determine the causes of developmental delay and dysmorphology. This work is undertaken in Nijmegen and is pioneering. The centre had tested around 10 children, and had achieved a diagnosis in six of these. However, the level of interpretation of the data to come to each diagnosis was huge, requiring weeks of work by a trained geneticist. My main learning point from this was, although the technology is becoming more and more accessible, the human input into interpretation is increasing and therefore a shift in the balance of work in genetics is inevitable. This is important for me to understand when teaching students about the new technologies, as well as when undertaking studies on the use of genetic testing for patient benefit. The level of human resource and bio-informatic support required to make a diagnosis must be included in any economic analysis of the technology. |

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