Lunch meeting of the Laboratory Techniques SIG

Time: Tuesday, July 11, 12:50pm - 2:00pm
Place: Marina 6, Sheraton San Diego Hotel and Marina

Agenda

1. Welcome

2. Introduction new chair/co-chair

3. Plans on future EQAs (Dr Sandi Deans) –

55 different EQA schemes offered by NEQAS – 80% labs participating are from outside of UK. The schemes available include NIPT for aneuploidies – SD presented a summary of this scheme on Monday of this meeting. SD Invited this SIG to be involved in ensuring the EQA schemes are fit for purpose and to provide schemes that are required.

NIPT scheme – difficulties in sample collection, joint scheme between CEQAS / EMQN & UKAS due to complexities, can register via any provider. Pilot scheme – T13 18 & 21 – artificial material for pilot (not suitable to all methods). 40 participating labs. Results collated in June 2017 and will be published shortly.

Pilot 2 – patient plasma so any method can be used. Limit number of participants (60-80). Registration will open in August. EQA distribution in autumn 2017.

Requested plasma – low risk & high risk, must not be an ongoing pregnancy from labs that measure fetal fraction. Volume – at least 10mls of plasma per submitted sample as the scheme providers need to give multiple labs same samples. RAPID study resources will be available for initial samples. Contact details provided by SD for any interested participants.

Additional questions posed by SD - Is sex determination only scheme for NIPT useful?

Monogenic disorders? Microdeletion syndromes?

Updated info from labs showed a recent increase in interest for microdeletions etc.

Samples provided must be stable as they will need to be freeze thawed a few times

Aim of assessment – end to end testing? Cell free DNA extraction, method, reporting. Sample swap scheme organised by EQA providers instead of locally may be more appropriate for some NIPT schemes – microdeletions or rare autosomal trisomies.

Brynn – pointed out difficulties in upgrading the scheme to microdeletion syndromes due to the rarity of available samples. – Spiked plasma?

Additional available resource on NEQAS website. CNV – gtac tool for CNV assessment – provided on the NEQAS website. – participant charged.
4. Received proposals for pre-conference courses, conference 2018 and educational activities (see attachment)

5. Other proposals?

6. ideas on collaborative research

7. Questions

8. Closing of SIG meeting

Proposals (in brackets the proponent/s):

Pre-conference course:

- Chromosomal Mosaicism in reproduction (Joint with Genetic Counselling SIG?) (SIG):
  1. In Prenatal diagnosis of first trimester → fetoplacental mosaicsisms and risk of fetal confirmation; mosaic results on amniocytes: how to counsel?
  2. In PGS → How pre-test counsel? How to prioritize mosaic embryos? Which prenatal care for transferred mosaic embryos?
  3. In NIPT → False positive/negative results; Follow-up diagnostic testing after abnormal NIPT: CVS, amniocentesis, cordocentesis?

Rossa & Lynn suggested better as an invited presentation as not enough evidence for pre-conference course. The PGS area was suggested to not fit with the other areas (separate challenge)

Brynn – suggested there was enough information but would there be enough interest for a couple of hours but maybe not 4 hours worth of material. Brynns lectures were well attended by obs. – Follow up procedures for NIPT ‘false positives’ was suggested as a good topic.

- NIPT: technical and quality aspects
  - Frequency and significance of technical test failures with different NIPT approaches (Lieve Page-Christiaens)
  - Is there ways to further improve NIPT (prenatal testing with cfDNA) or have we reached the maximum attainable performance? (Lieve Page-Christiaens)
  - External quality control for cfDNA tests - work in progress (lecture?) (SIG)
  - Regionalization of NIPT tests: the key role of local laboratories in the on the spreading process of the tests (SIG)
  - Fetal fraction measurement: necessary? methods? Cut-off (SIG)
  - High resolution NIPT: current and future possibilities? (SIG)
  - Rare autosomal trisomies in NIPT tests: clinical utility and management options (SIG)
Brynn asked % of lab / clinical at ISPD – more clinical attendees.

All clinical points impact clinicians. Improve counselling – End a preconference course with implications for counselling? Lynn thought this would be a popular choice.

Francesca agreed. Suggestion that a bioinformatician would be helpful to explain how NIPT works. Francesca and Lynn raised doubts regarding the level of understanding of the audience if a bioinformatician explains to OBGYNs how different NIPTs work

Limitations of different methods to explain to clinicians.

Debate:

- Cell-based NIPT vs cell-free DNA NIPT (Lieve Page-Christiaens + SIG)
  Brynn suggested it was too soon to have this debate. Maybe invited presentation on why it hasn’t happened yet? -

Main lectures

- Expanded Carrier screening: cost-effectiveness, which syndromes? follow up: PGD, NIPD, prenatal diagnosis (SIG) (or preconference course with SIG reproductive screening?)

Covered in Berlin? How panels are chosen – perspectives from different countries, different companies? Discuss with Jim Goldberg – Group in Nijmegen who perform carrier screening. Whole exome carrier screening in US? Counsyl. Cost reduction allows more genes to be included in panel. Ethical aspect could be discussed. IVF centres – what do they use & why? Lynn not sure if this would be popular enough for a preconference course.

- A priori and post-test risk for chromosome/genomic abnormalities after negative test results (Grati) – re-inforce concept of risk in general prenatal & postnatal screening / test. Speaker who can give examples. Shared decision making groups – pre-conference course.

- Invasive prenatal diagnosis:
  - The use of WES/WGS for diagnostic purposes in rare undiagnosed diseases in fetuses and newborns (Lieve Page-Christiaens) – discussed a lot this year.

- New tools for pretest counselling – models to aid counselling & give correct significance. Resources for patients to look at prior to attending clinic. 15 minute video prior to NIPT – F Grati Gives good awareness to patient.