1. Welcome (Elaine Holgado)

2. Review of previous year’s activity:
   
   An overview is presented by Elaine Holgado (see attachment)

3. Demonstrating competency in the Genetics Laboratory (talk by Dr Sandi Deans)

   60 different countries currently participate in UKNEQAS and CEQAS (GenQA) Individuals working in genetic laboratories are increasingly becoming required (ISO15189!) to demonstrate training and competency for different roles they fulfill within the laboratory. Therefore GenQA developed an online training and competency tool to provide a source of external review and evidence continual professional development for the individual. It involves entering a virtual genetics laboratory where the participant can navigate between workstations. Registration enables unlimited annual access to the four scenarios: Sample Reception, Variant interpretation, Result analysis and Report Authorization. Next month, also CNV interpretation will be added. Sandi demonstrates how it works. Uptake: quite a lot of pathologists participate.

4. Proposals for pre-conference courses, conference 2019 and educational activities

Communication from the board by Prof. Lynn Chitty: in previous years, the SIGs were involved in proposing ideas for preconference courses and organising them. This year, the board decided that for the coming year, there only will be 4 preconference courses. These will be determined by board and local committee (no role for SIGs anymore). The 5 SIGs will have a breakfast-meeting (7.45h-9h) on 3 mornings (2 on Monday, 2 on Tuesday and one on Wednesday). These will be short business meetings with room for lectures from invited speakers as well as own members. The purpose of the board is to bring more structure in these meetings. There also will be the opportunity of recording presentations that can be used for educational purposes. Choice of one or more topics.
5. Ideas on collaborative research

Dr. Francesca Grati proposes to launch a survey to map guidelines in different countries for cfDNA test and CMA. The idea is to also include the type of the applied technology and the reimbursement regimen. The purpose is to make a simple table that can be filled easily and to include countries from USA, Europe and Asia that are all represented within the SIG. Dr. Sandi Deans is willing to circulate the surveys. This will be a collaboration of ISPD and GenQA.

Prof. Brynn Levy wants to collect data on how CMA is reported in different countries. There is variation and the goal is to unify reporting practices. He proposes to start with the controversial finding of a susceptibility locus (with variable phenotype and incomplete penetrance). Is it reported prenatally and how does this compare to the postnatal situation? UKNEQAS can help since they receive many reports from the EQA-schemes. Everyone who is interested can contact Brynn.

-if there are other ideas for collaborations, please let us know? See for contact details on the website

-ideas for next year SIG meeting/webinars:
  o consider presentation of case reports during the SIG-meeting
  o technical challenges in the lab
  o How to choose between different techniques for one application?
  o View of different companies on a specific technology

6. Questions

7. Closing of SIG meeting