External Quality Assessment (EQA) for Genomics and Clinical Genetics - ensuring the quality of the entire genetics service

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INTRODUCTION

- GenQA offers end to end testing for all aspects of the genomics service including clinical genetics.
- Medical laboratories working to International Standard ISO15189 are required to evidence the training and ongoing competency of staff at all levels for all tasks.
- GenQA has developed Tissue-i to provide a tool to assess tumour content.
- GenQA developed the online competency tool G-TACT to provide continued assessment of laboratory and clinical staff to perform certain tasks.
- G-TACT provides evidence of competency for routine genetic laboratory duties and identifies un-met training needs within laboratory staff.
- Current continuous modules include: Sample Reception, Variant Classification and Report Authorisation.

CLASSIFICATION OF VARIANTS  (CNVs and SNVs)

- Aim is to assess an individual’s competency to determine the pathogenicity (and classification) of single nucleotide variants (SNVs).
- Free G-TACT trial module was released in June 2018 for participation and completion within two months.
- Participants were required to classify each variant (and choose the relevant classification rule for their evidence (based on Richards et al., 2015)).
- 571 participants registered, 142 used the scenario and 82 participants completed all 5 variants.
- Summary Report and Participation Certificate provided on completion of G-TACT Trial module.
- Additional free trial modules are available during 2019: BRCA variant analysis, SNV, CNV and HaemOnc.

COPY NUMBER VARIANT (CNV) and SINGLE NUCLEOTIDE VARIANT (SNV) RESULTS

- No participant provided the expected classification for all the CNV and SNV variants.
- However, most participants correctly identified whether a variant was actionable (i.e. pathogenic versus benign).

SUMMARY

- GenQA offers 94 different EQAs across the genomics specialties.
- The EQAs are either laboratory, clinical or individual based.
- The EQAs are educational.
- EQA improves the quality of the diagnostic service and identifies gaps in the genomic diagnostic service.
- There are challenges associated with variant classification and the need for education, competency assessment and standardization.
- G-TACT provides an easy, automated method for individual competency assessment.
- Tissue-i improves the clinical tumour content estimates and marked area for microdissection.
- For further information in our EQAs visit us at our STAND at ECA or email info@genqa.org

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