Introduction

The hereditary haemochromatosis (HH) EQA has been offered by the RCPAQAP since 2003. The majority of participants are in Australia, with participants ranging from Hong Kong, India, New Zealand and Oman (Figure 1). HH is an autosomal recessive disorder that is associated with variants in the HFE gene and iron overload. Three HFE gene variants (c.845G>A, c.187C>G and c.193A>T) have been characterised and associated with HH. For this EQA program, participants were provided with 8-10 DNA samples per year and are required to test each sample for c.845G>A, c.187C>G and c.193A>T if performed in their laboratory. Here we report the overall results from 2014 to 2018. Participants were assessed according to the consensus values based on a target value obtained from reference and source laboratory results.

Methods

An EQA was developed to assess inter-laboratory performance on HFE genotyping. A total of 44 DNA samples were distributed to 37 laboratories during the years 2014-2018 (Figure 2). Participants were requested to report on the gene variant detected and the zygosity status in each sample tested. The hereditary haemochromatosis (HH) EQA has been offered by the RCPAQAP since 2003. The majority of participants are in Australia, with participants ranging from Hong Kong, India, New Zealand and Oman (Figure 1). HH is an autosomal recessive disorder that is associated with variants in the HFE gene and iron overload. Three HFE gene variants (c.845G>A, c.187C>G and c.193A>T) have been characterised and associated with HH.

Results

The EQA data from 2014–2018 are presented below (Figures 4, 5 & 6) and represent the levels of genotype reporting for the three HFE gene variants; c.845G>A, c.187C>G and c.193A>T.

Participant results were assessed as:

- **Concordant**: if the participant’s result matched the consensus result.
- **Discordant**: for a participant result which does not match the consensus result or is incorrect and if the participant failed to obtain a result where one would be expected and no comparison could be made.

Conclusion

The risk of iron overload is dependent on the type of variant detected. A homozygous c.845G>A, homozygous c.187C>G and compound heterozygote c.845G>A/c.187C>G variant have varying risks of iron overload which is associated with HFE.1 The significance of c.193A>T is currently unknown.2 However, participants routinely perform this test if requested by the referring clinician. Participation in an EQA is essential to ensure that consistency of testing and reporting standards are maintained. This review has identified that over the last five years, participating laboratories have consistently produced good quality data for HH testing. Based on these data, patients are at a reduced risk of mis-diagnosis for HH.