Participant Survey 2019 Summary

GenQA GENOMICS QUALITY ASSESSMENT

Member of UK NEQAS *consortium*

Summary of feedback from 2019 GenQA Participant survey

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UKAS PROFICIENCY TESTING

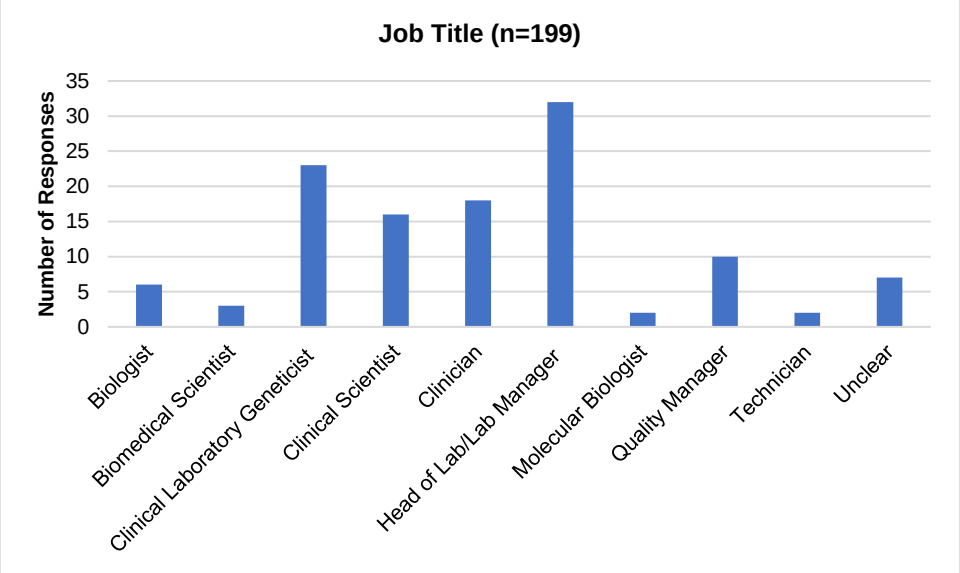
Introduction

The survey was sent to ~2500 participants during October 2019, of which 252 (~10%) participants responded to the survey. Not all individuals answered all questions. For some questions, only a very small number of participants responded (e.g. twelve or less). The survey included questions on:

Membership
Registration
EQA Participation
Sample quality
EQA Distribution
Submission
Appeals
ILRs and Summary reports
Additional EQAs requested
Further improvements

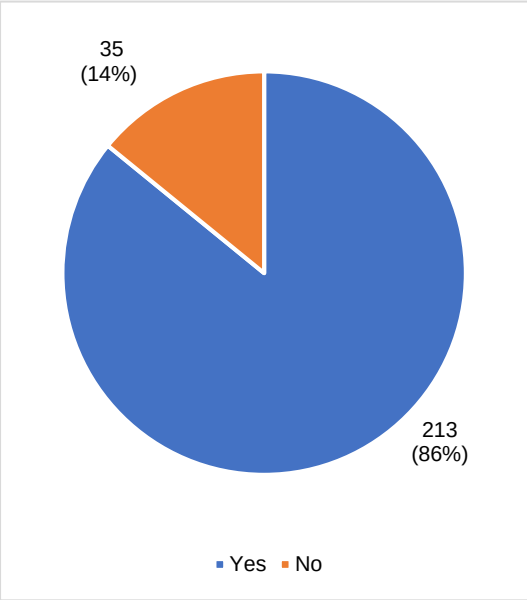
Data is presented in various formats including pie charts and graphs). This data was collected from a slider, where participants were asked to indicate their satisfaction from 0-100. Each spot on the graph represents a response. Where there are a lot of responses at 0, it is difficult to know whether this is a true response, or an individual declined to respond. Consequently, to improve the participant feedback, GenQA will use a different measure of satisfaction in future surveys.

The responders described their role below:

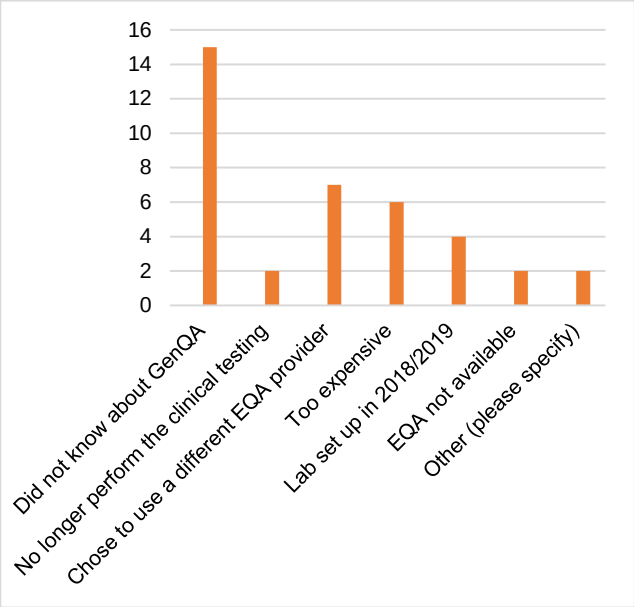


1. Membership

Did your laboratory participate in GenQA EQA in 2018? (n=248)

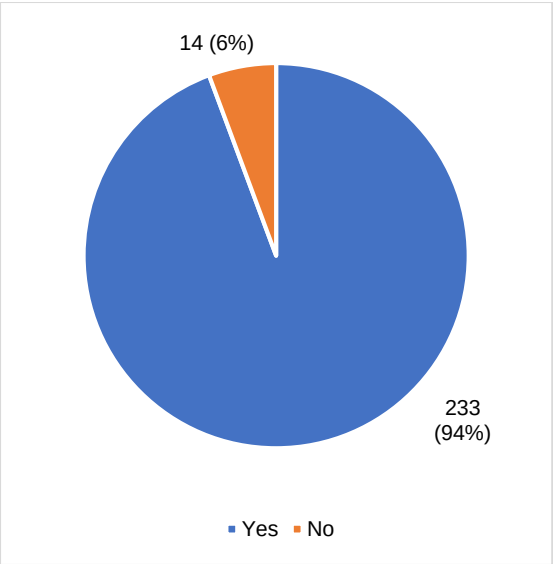


If not, then please indicate your reason(s):

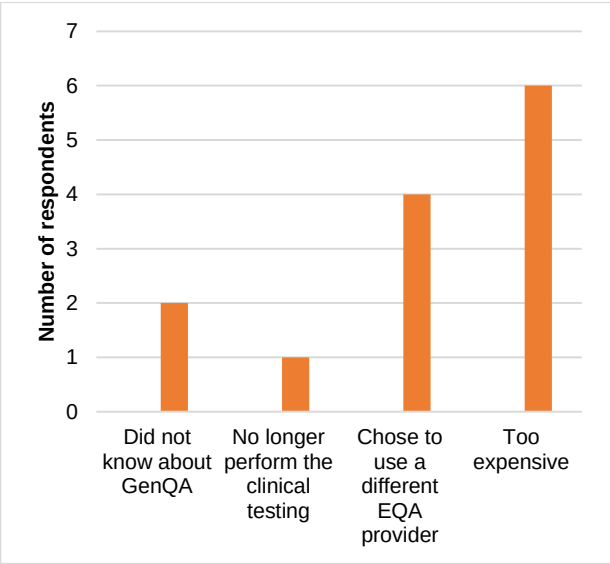


Other: due to lab relocation, and unknown decision of laboratory head.

Is your laboratory registered to participate in GenQA EQAs during 2019? (n=247)



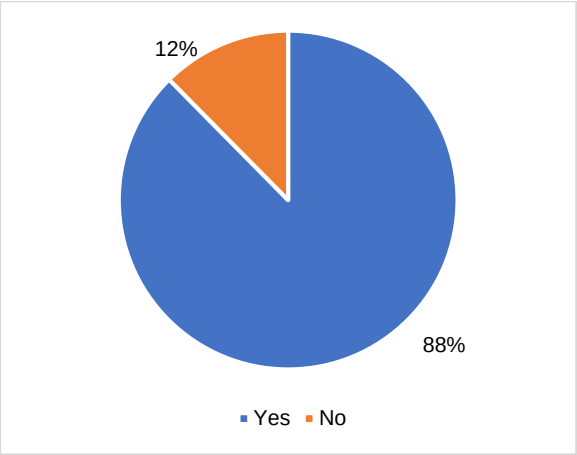
If not, then please indicate why: (n=12)



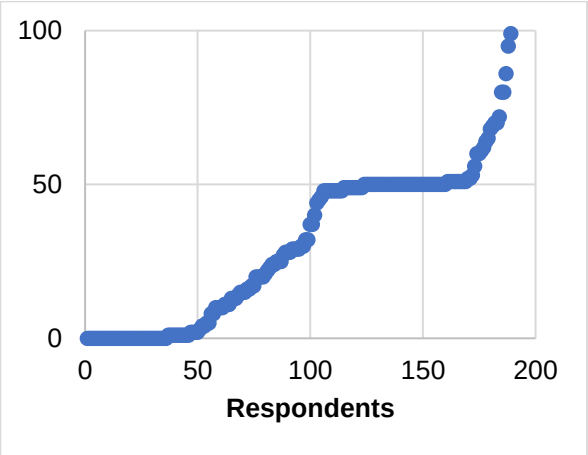
Although a small number of responses, the cost of participation is an issue. GenQA does keep the costs as low as possible and runs as a self-funding, not for profit organisation. In addition, GenQA offers additional educational workshops, advice and technical support to participants to provide a service which is value for money.

B. Registration

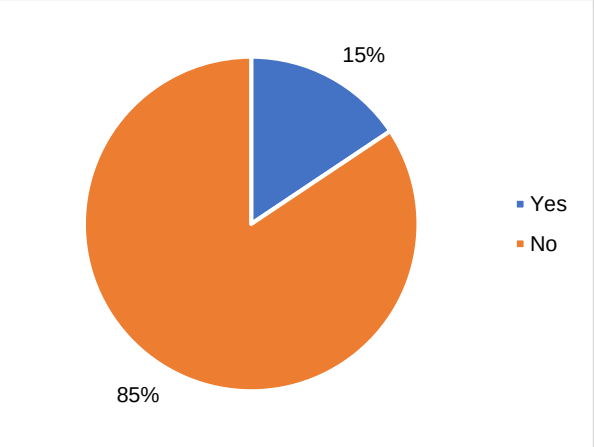
Did you complete the online registration for your laboratory? (n=242)



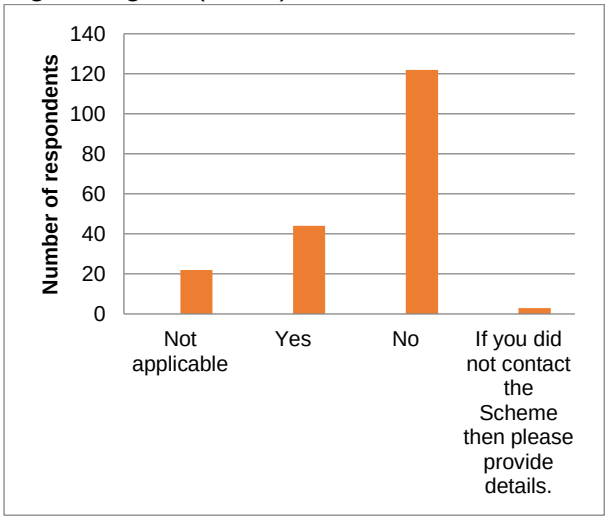
How would you rate the ease of registering? (n=189)



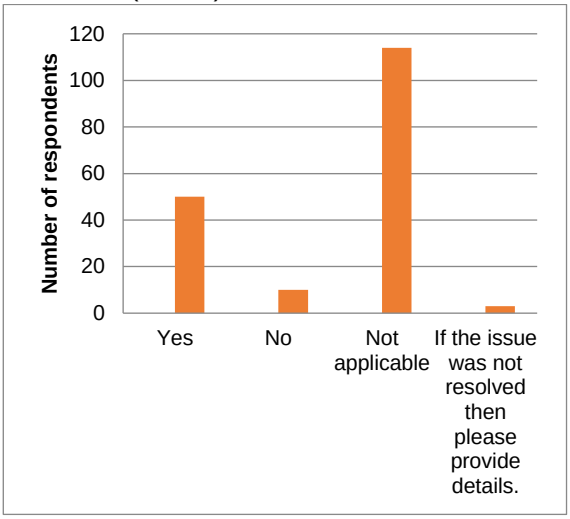
Did you experience any issues when registering? (n=192)



Did you contact GenQA for help when registering? (n=191)



Did the help you received resolve the issue? (n=174)

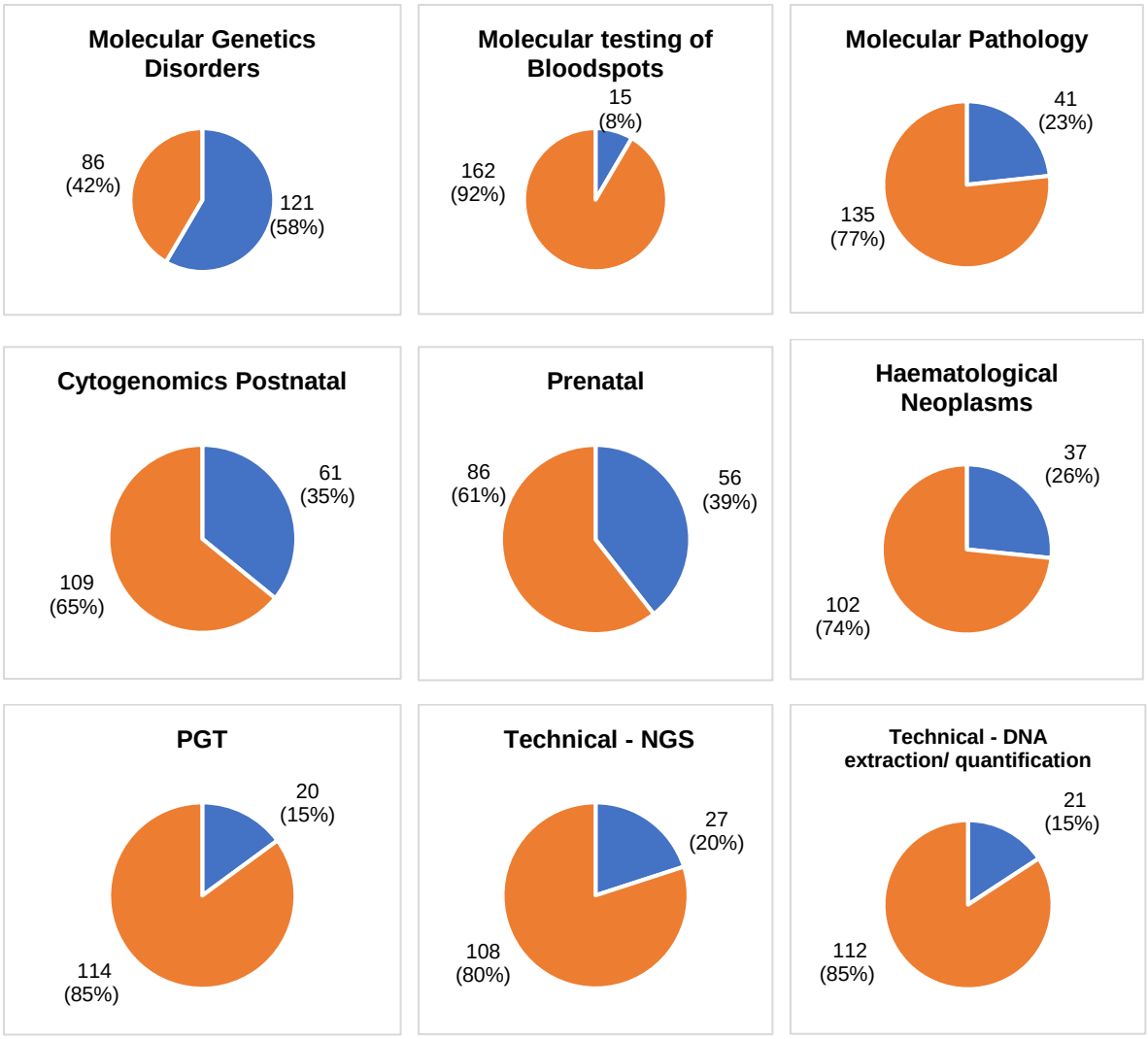


Participant comments regarding registration:
Comments were mainly regarding IT issues and communication:

	Participant comments regarding registration	GenQA response
1	Request for immediate confirmation of registration, not when registration is closed.	GenQA provide confirmation of an order, once it has been accepted, within a few days. The order is also visible through the Orders section of the Participant page. However, we are unable to confirm registration until registration is closed, as GenQA needs to ensure that adequate participants have registered to proceed with an EQA.
2	Selection of an EQA, but the system would then return to the top of the list	This issue has been resolved. Thank you for bringing this issue to our attention.
3	Communication regarding merging of EQAs and name changes	For the 2020 EQAs, where an EQA has merged or been updated the participants in the previous EQA were contacted by email to inform them of the changes. GenQA has also produced several leaflets this year providing information on both new and updated EQAs for 2020.

C. EQA participation

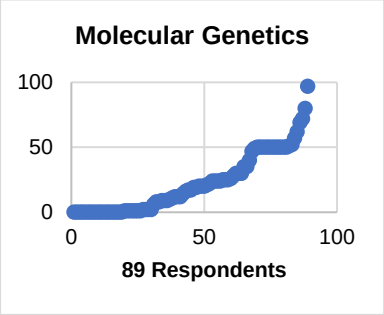
Does your laboratory participate in these EQAs? Yes: No:

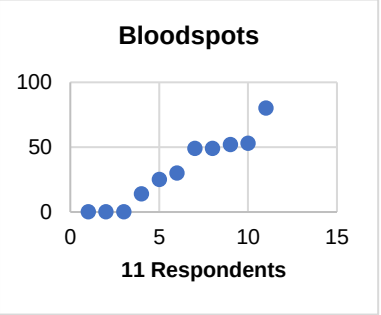


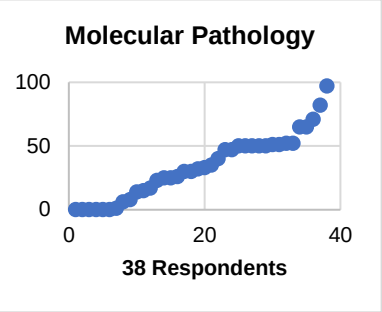
Participation ranged from 8% (bloodspots) to 58% (Core Molecular Genetics). This is not a reflection of participant numbers but the percentage of those completing the survey.

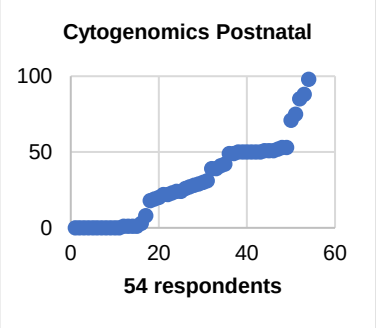
D. Sample Quality

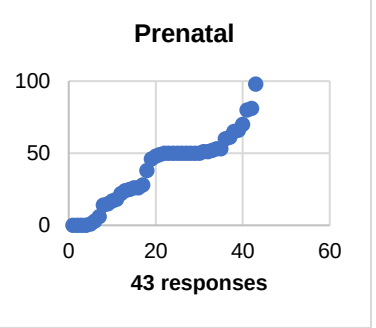
How do you rate the quality of samples in these EQAs:

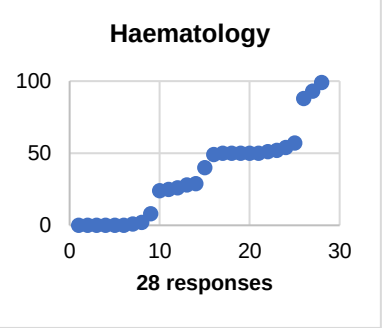


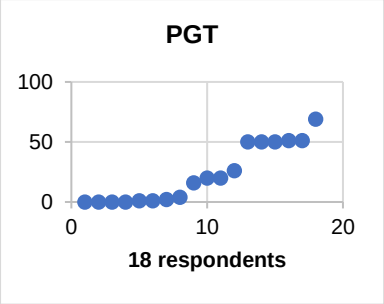


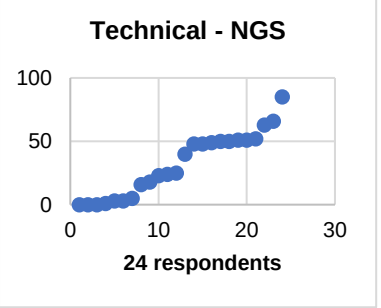


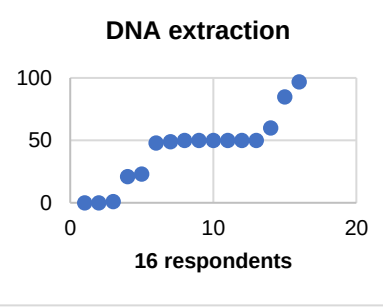












GenQA received repeat sample requests for 261 (~2.5%) samples from a total of 9,807 EQA samples distributed. These requests came from 160 different laboratories:

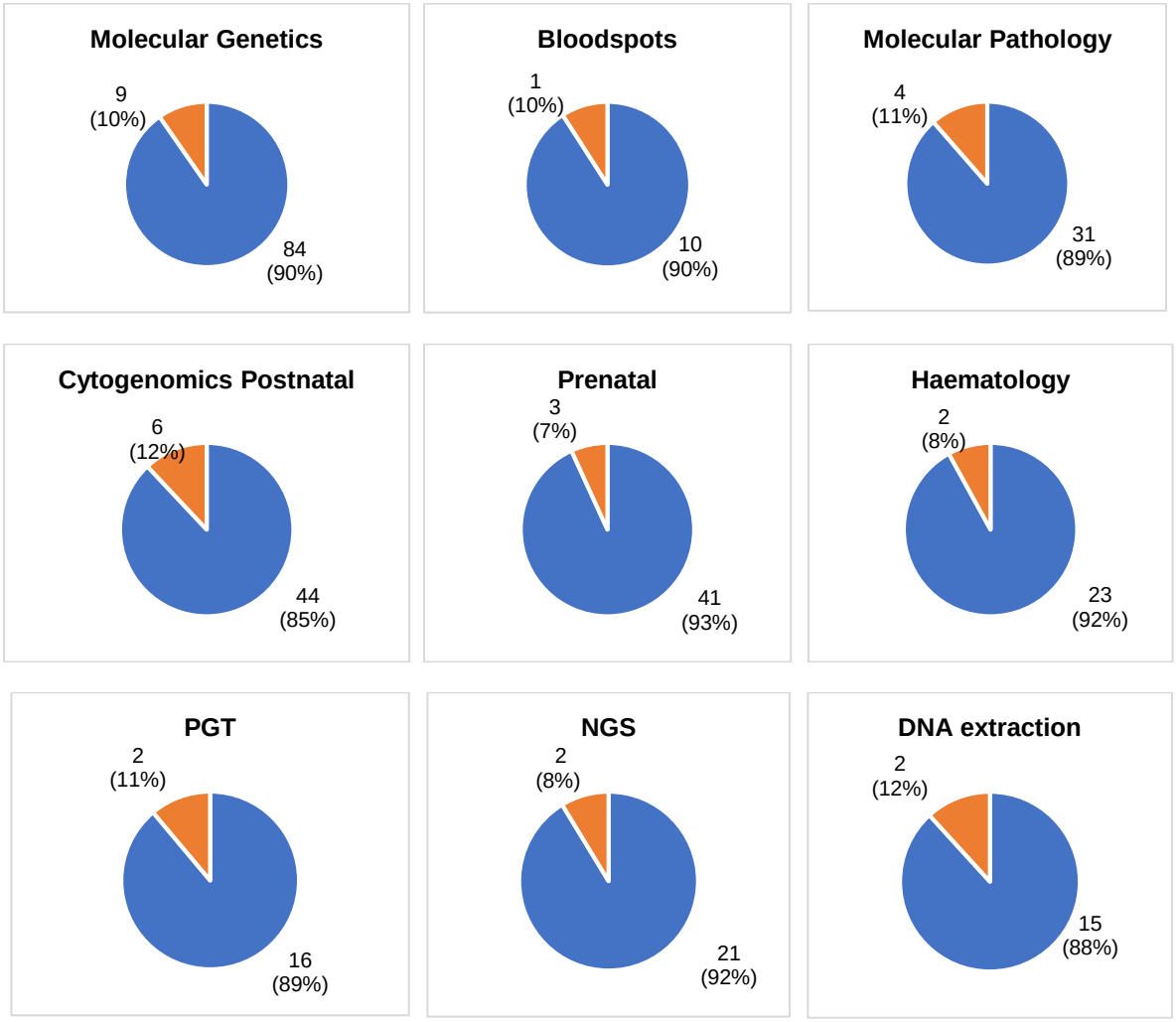
1 laboratory requested 8 repeat samples.
1 laboratory requested 6 repeat samples.
5 laboratories requested 5 repeat samples.
3 laboratories requested 4 repeat samples
3 laboratories requested 3 repeat samples.
106 laboratories requested 1 or 2 samples.

The laboratories requesting 6 and 8 repeats samples had participated in 32 and 48 EQAs respectively.

The comments regarding quality were focussed on the following areas:

	Participant comments regarding sample quality	GenQA response
1	Low volume/concentration of DNA supplied, which amplifies less well than usual samples.	Where possible GenQA prefer to provide patient samples e.g. DNA and for some EQAs, particularly those where a large amount of DNA is required. GenQA can only provide the minimum amount necessary for the test due to limited sample availability. We also use commercial samples from Horizon Diagnostics and Coriell which may also amplify less well due to the nature of their creation (e.g. by cell-line). However, the validation processes enable GenQA to ensure reportable results can be obtained from the sample provided.
2	Request for more FFPE slices	Where possible GenQA provides patient tissue samples e.g. FFPE and for some EQAs, can only provide the minimum amount necessary for the test due to limited sample availability. This is a limitation of using patient material for molecular pathology testing, but we believe that the benefit of using real material to address issues such as heterogeneity, low tumour content, necrosis etc outweighs the use of large scale manufactured samples. The validation processes enable GenQA to ensure reportable results can be obtained from the sample provided.
3	The rolled sections are stored together in one tube. Because they are not really 'rolled it is difficult to separate the two sections from each other	GenQA provides two sections in the same tube where it is expected that DNA extraction is performed on both sections. A separate option for Idylla testing in specific EQAs is available where the samples are split into two tubes.
4	Mutation level in some samples is low.	GenQA try to provide a mixture of samples with different levels of mutation for molecular pathology and molecular genetic disorders. This is to ensure that laboratories are aware of the limitations of their test and to reflect routine testing of patient samples.
5	Request for blood samples/slides for cytogenetic analysis	Where possible GenQA provides patient samples. This is a large EQA with >170 participants and many of the cases are from young children/adults. It is currently not feasible to obtain sufficient blood from the patients for an EQA program. GenQA is always reviewing its processes and this will be discussed at the next Postnatal SAG meeting. Both slides and slide exchanges were sent many years ago when there were fewer participants. However, participant feedback was that an online analysis of metaphase images improved the both the quality and consistency compared to the slides.
6	Poor quality of fixed samples	Several EQAs involve fixed cell suspension. If this is referring to the Mature B & T cell FISH EQA, then the fixed cells do not have a hypotonic pre-treatment and the interphase cells may appear different. Please can you contact the GenQA office directly on info@genqa.org so that staff can address your concerns.
7	The quality of Haematology samples has improved.	Thank you for your positive feedback.
8	PGT-M samples not good enough for STR amplification.	In general, we do not receive sample requests for repeat samples for the PGT-M EQA. Please contact GenQA on infor@genqa.org so staff can address your concerns.

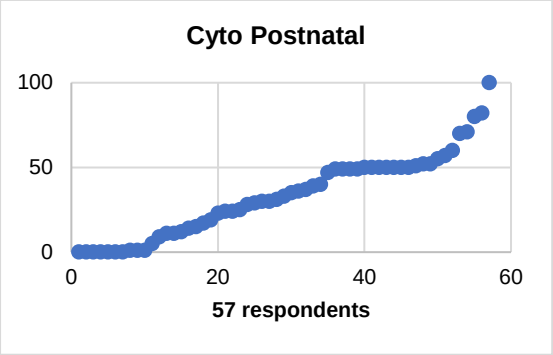
Do the samples provided represent the clinical material routinely tested? (Yes: ■ No: ■)

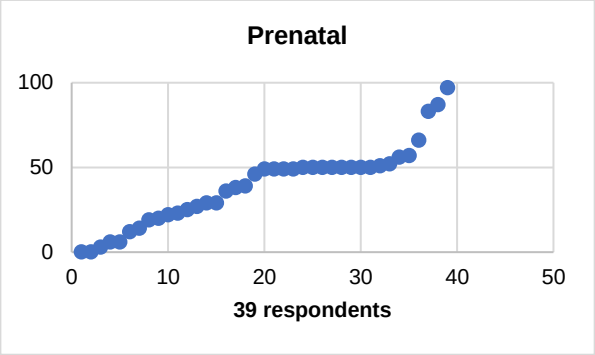


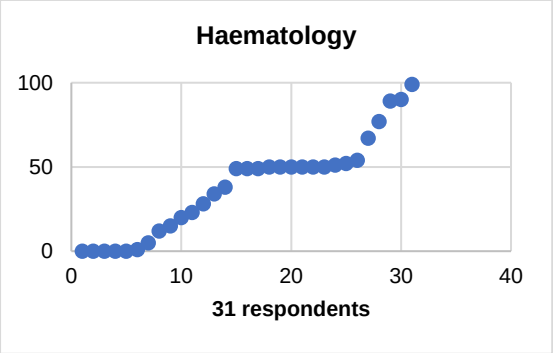
At least 88% of participants for each specialty considered that the EQA samples provided represented the clinical material received by the laboratory.

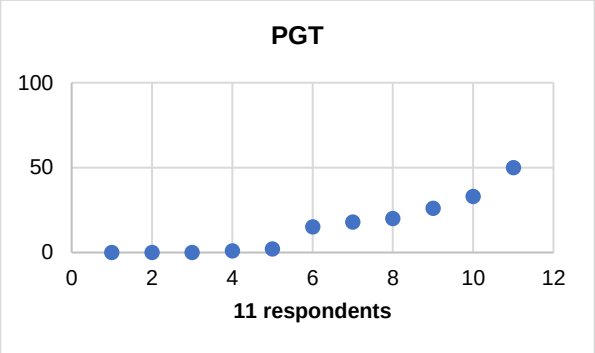
	Participant comments regarding relevance of clinical material	GenQA response
1	Comments regarding the samples that laboratories standardly receive and whether these are the same as the EQA sample types.	GenQA try to include a broad range of samples to satisfy the testing and requirements of different laboratories. This may include differing material, material provided in a different format or a range of the amount of DNA supplied.
2	EQA samples are regularly more complex, which generally represent a small percentage of a laboratory's workload.	We understand that EQA cases may represent more complex and uncommon cases, but as the laboratories receive these rarely it is important that laboratories are able to deal effectively with these scenarios. This also provides an educational element for participants. However, we understand the request for more 'routine' EQA cases and will bear this in mind for future EQA cases.
3	Not always representative of 'heel prick' blood spots.	The EQA samples are created from donated patient blood samples which are then spotted onto newborn screening cards for analysis. GenQA includes a variety of genotypes which represent the possible scenarios that laboratories might encounter.
4	It would be useful to receive slides or blood for postnatal analysis (rather than just images).	Where possible GenQA provides patient samples. This is a large EQA with >170 participants and many of the cases are from young children/adults. It is currently not feasible to obtain sufficient blood from the patients for an EQA program. GenQA is always reviewing its processes and this will be discussed at the next Postnatal SAG meeting. Both slides and slide exchanges were sent many years ago when there were fewer participants. However, participant feedback was that an online analysis of metaphase images improved the both the quality and consistency compared to the slides.
5	Outdated system – it should be about testing for a referral reason/pathway, rather than sample type (e.g. amnio or CVS)	GenQA is introducing more patient pathways EQAs. However, some genetic centres still require sample based EQAs so these will continue to be offered. Please also note that ISO15189 requires testing of the technology used diagnostically so some EQAs will also cover this aspect of diagnostic testing.

How do you rate the quality of images in these EQAs?

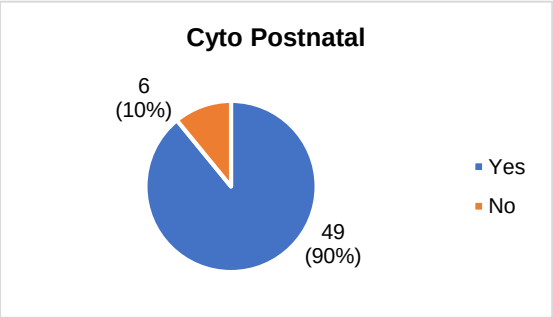


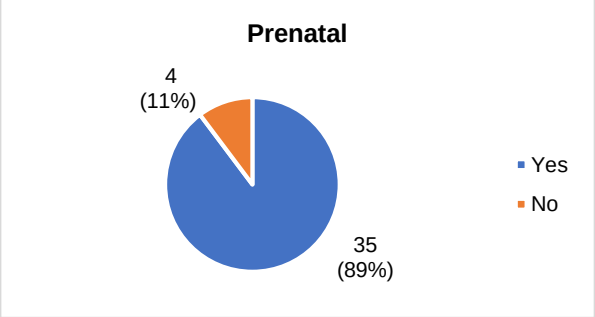


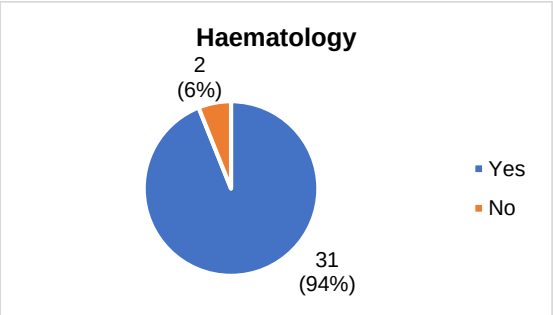


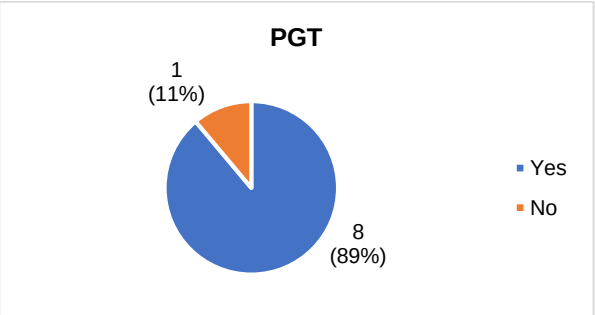


Do the images provided represent the clinical material routinely tested?







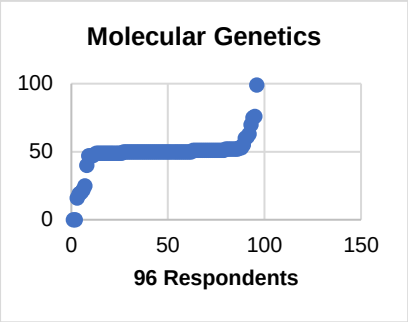


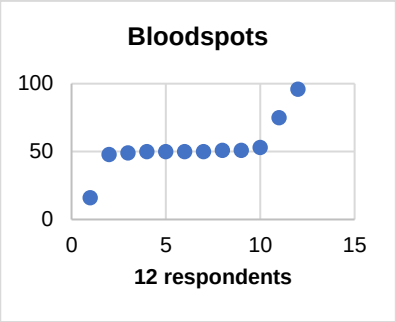
At least 89% of participants for each specialty considered that the EQA images received were representative of clinical material.

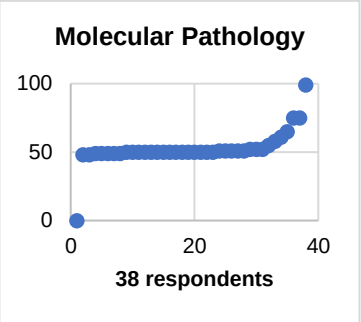
	Participant comments regarding relevance of images	GenQA response
1	Poor quality of images with inconsistent size and number – unclear how many images are available.	GenQA is dependent on the assessors providing suitable images for EQA which are then analysed and validated 'blind' to ensure that there are sufficient images to obtain a result. Sometimes mosaicism may need to be excluded and therefore additional images are supplied. The website shows all the images available so when you open the EQA, you can see how many images there are either by counting or looking at the numbering system. As there are multiple image analysis systems available and each is customised to the user the EQA images provided may on rare occasions need re-sizing. You can resize the images if they are too small/large and your image analysis provider will be able to assist you.
2	Limited number of cells, sometimes with poor banding/definition.	GenQA is dependent on the assessors providing suitable images for EQA which are then analysed and validated 'blind' to ensure that there are both sufficient images and the metaphases are of a suitable quality to obtain a result. Sometimes mosaicism may need to be excluded and therefore additional images are supplied.

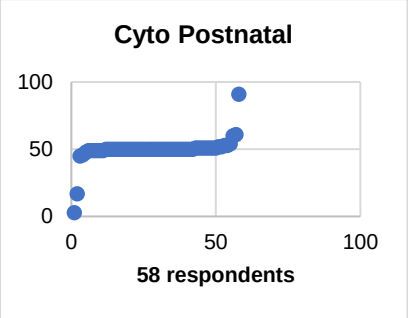
E. EQA Distributions

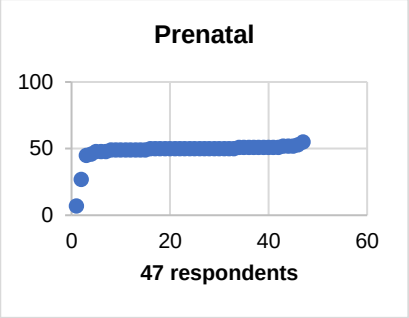
How do you rate the number of EQA samples provided per EQA distribution?

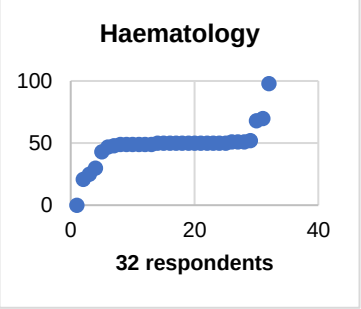


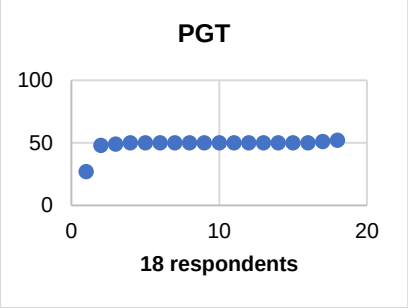


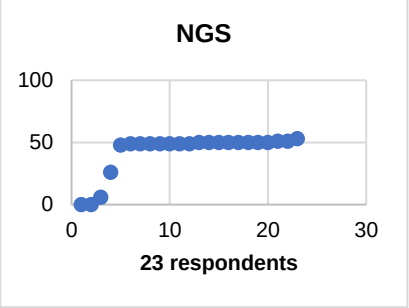


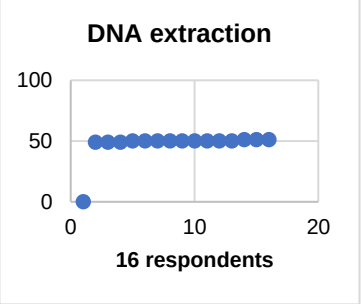






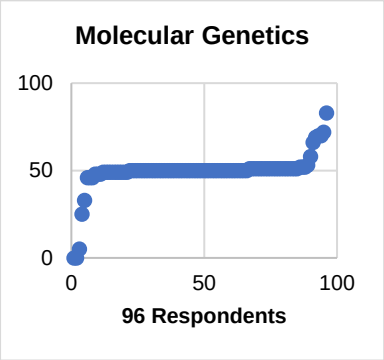


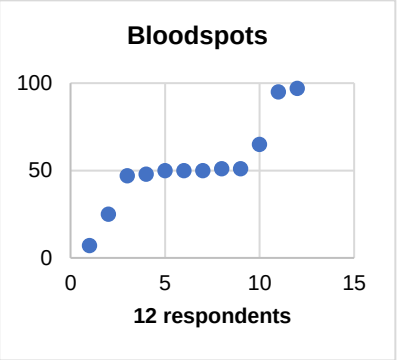


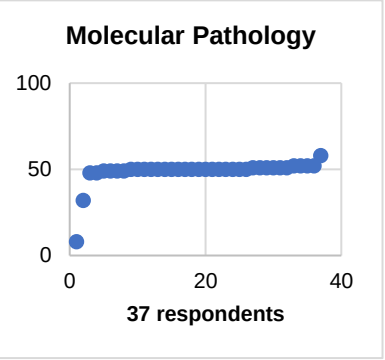


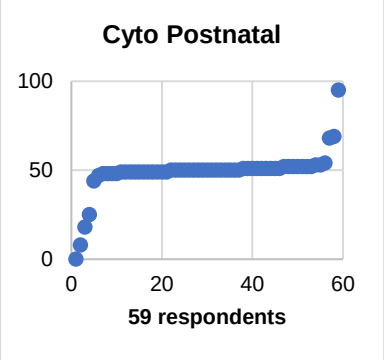
In general respondents are happy with the number of EQA samples provided per distribution.

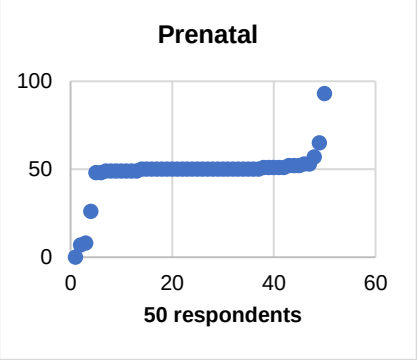
How do you rate the frequency of EQA runs?

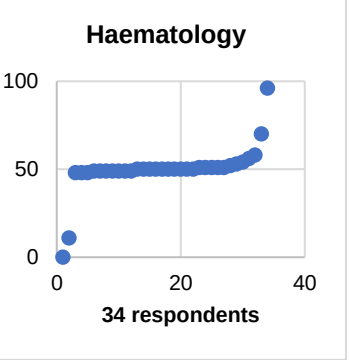


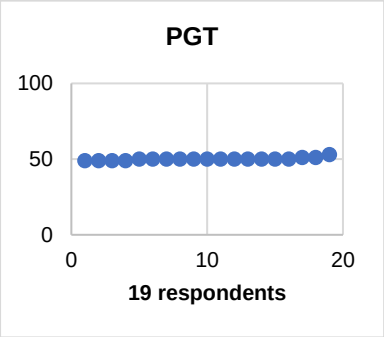


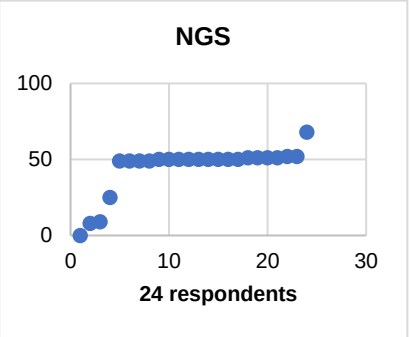


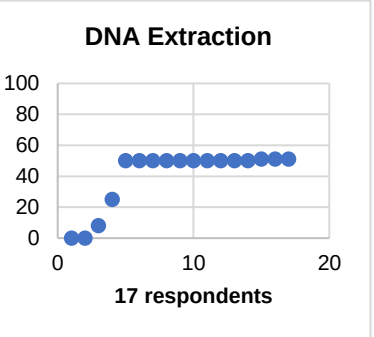






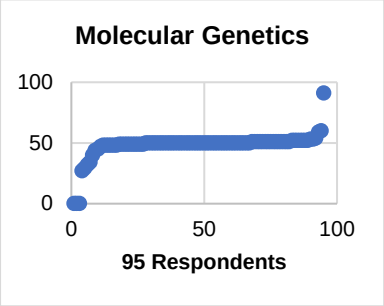


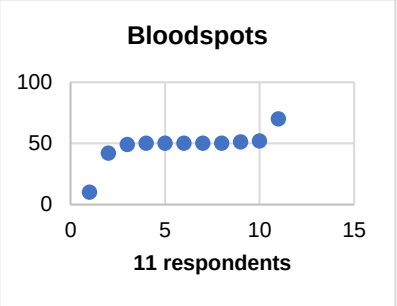


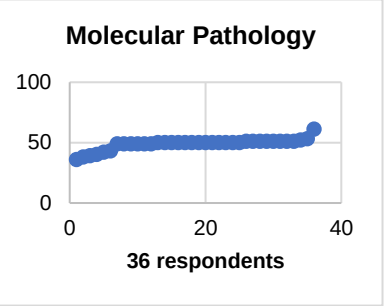


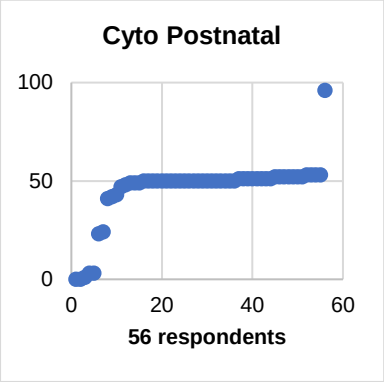
In general respondents were happy with the frequency of EQA distributions.

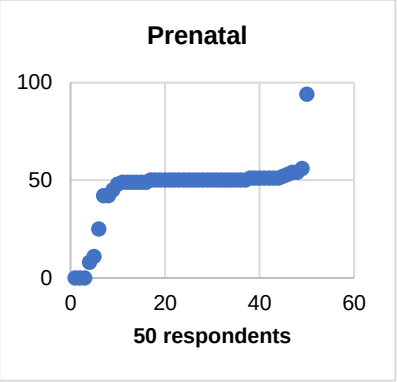
How do you class the information in the EQA distribution letter?

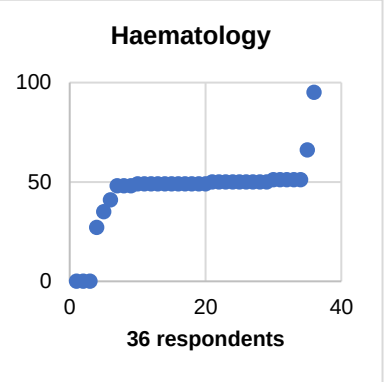


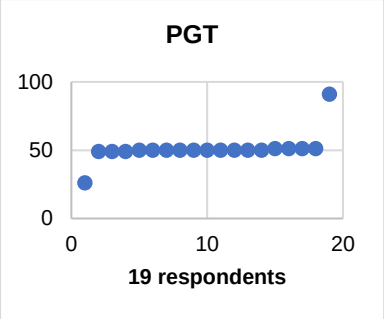


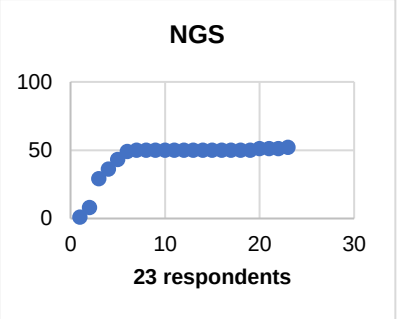


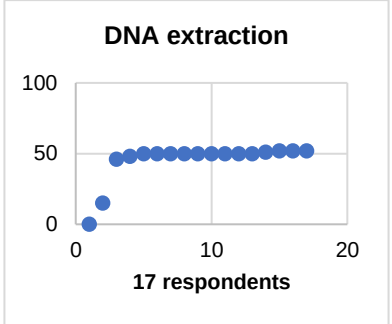








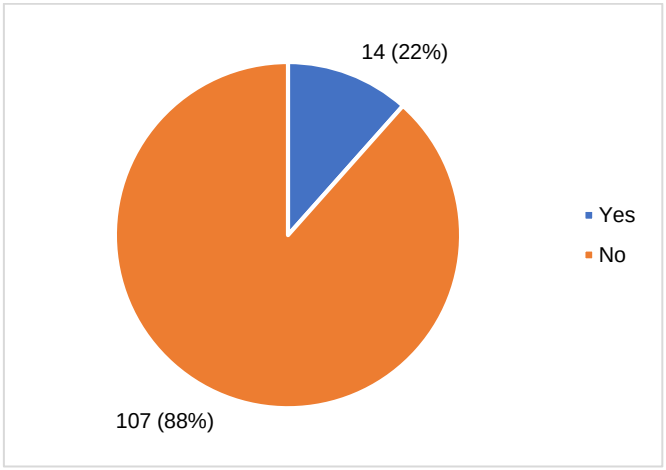




In general, respondents were satisfied with the information provided in the distribution letter.

G. Submission

Did your laboratory experience any issues when submitting the EQA returns (reports, result proformas, additional/supporting information)? (n=121)

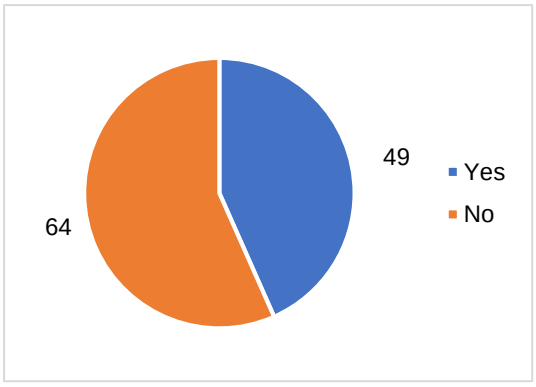


No issues were experienced by 88% of respondents when submitting EQA returns.

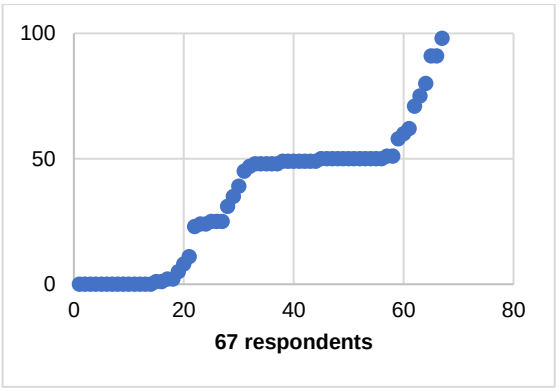
	Participant comments regarding submitting EQA returns	GenQA response
1	Difficulties submitting on a MAC platform.	GenQA is not aware of issues when using a MAC system and encourages this laboratory to contact the Scheme so we can investigate this issue.
2	No confirmation that documents had uploaded correctly.	When the submission is submitted the complete status changes from a yellow circle to a green tick. You may need to refresh your browser to see this. GenQA do not want to send multiple emails when reports are uploaded as many laboratories participate in lots of EQAs.
3	Difficulty in using panel test forms, cannot amend them as they are online.	NGS gene panels test forms are a relatively new development and GenQA is not aware of any participant issues. Please contact the Scheme so we can improve the user experience.
4	Process for uploading results is complicated and dispersed in numerous documents.	GenQA is not aware of major participant issues and encourages this laboratory to contact the Scheme so we can improve the user experience.
5	GenQA website unavailable due to IT error for several hours	We apologies for this inconvenience and are working with our website provider to minimise any downtime, particularly when EQAs are active.
6	Layout of website make user experience poor	Please contact the Scheme as we would like to understand how the website could be improved.

H. Appeals

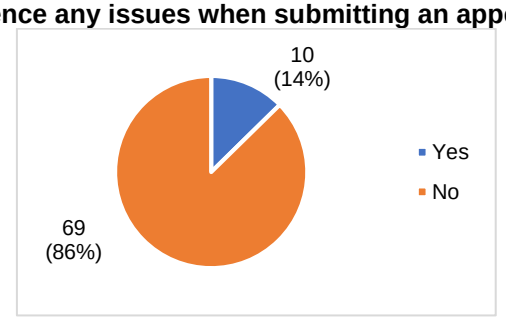
Did your laboratory submit an appeal using the online process? (n=113)



How would you rate the ease of submitting an appeal?



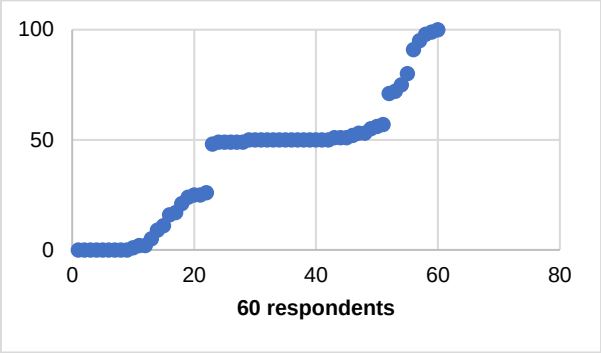
Did your laboratory experience any issues when submitting an appeal? (n=80)



Ten participants (12%) experienced an issue when submitting an appeal.

	Participant comments regarding submitting an appeal	GenQA response
1	Some website technical issues	We hope the technical issues were resolved quickly and your appeal submission was not affected.
2	Prefer to submit appeal in own language, rather than English.	GenQA are investigating the possibility of laboratories being able to submit appeals in their own language, as we appreciate that submitting in English may result in loss of context of an appeal. Some of our EQAs do allow multiple language submission (see specific EQA distribution letter)
3	No facility to include images or supporting information.	Thank you for this feedback. The website has been improved and participants are now able to attach images and supporting information to their submission.
4	Word limit was too restrictive.	Thank you for this feedback. The word limit has been extended, although we would still encourage laboratories to submit a concise and succinct appeal.

How would you rate the feedback received regarding the outcome of the appeal?

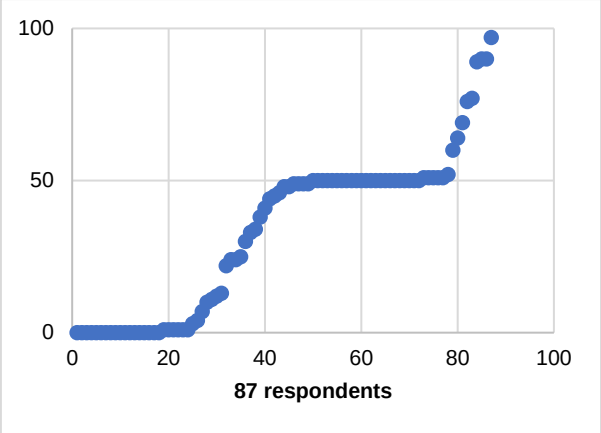


How can the feedback regarding the appeals be improved?

	Participant comments regarding appeals feedback	GenQA response
1	Request for a more in-depth explanation as to why an appeal was rejected	GenQA accept that it is extremely important that participants understand the rationale for rejecting an appeal and will endeavour to provide as comprehensive an explanation as possible.
2	Request for more dialogue between the laboratory submitting the appeal and GenQA.	GenQA has to provide a fair and equitable appeals process for all laboratories. GenQA are happy to answer any further questions that a participant may have regarding an incorrect result, particularly if this has implications for other patients and the testing process within a laboratory. However, GenQA are unable to enter into complex dialogue regarding a specific appeal during the appeal process. Laboratories are contacted by email once an appeal response is available on the website.

H. ILRs and Summary Reports

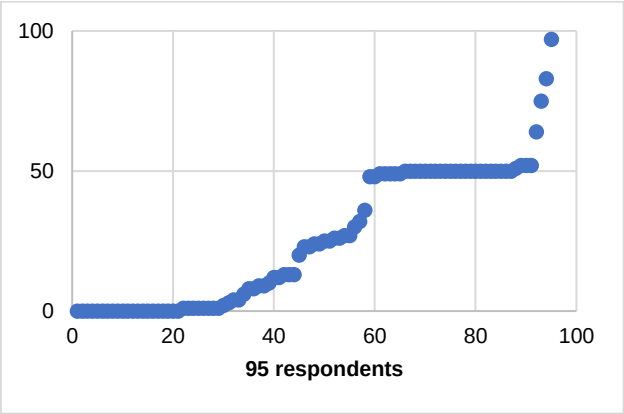
How would you rate the feedback received on the Individual Laboratory Reports (ILRs)?



How can the feedback on the ILRs be improved?

	Participant comments regarding ILRs	GenQA response
1	Provide examples of reports	GenQA will look into providing a selection of 'good' reports for an EQA. General guidance on reporting for genetic laboratories was published by the ACGS in 2015 (in collaboration with GenQA/UKNEQAS) and by ESHG Quality Committee in 2014 in collaboration with GenQA.
2	Include more detail in the ILR	Thank you for this feedback. The ILR comments are provided as succinct as possible to prevent misunderstanding. All recurring issues are discussed more fully in the EQA Summary Report however we shall take this feedback into account for future EQAs.
3	More reference to documents used in decision making	GenQA will work towards ensuring that references used in the decision making process for scoring are included in the summary report.
4	It takes a long time to receive feedback	GenQA understands this issue and for the 2020 EQAs, the analytical/ genotyping results will be released to the laboratories as soon as the assessment has taken place. This will help laboratories to identify any critical errors and can take action if required. Unfortunately, the collation of the scores, feedback comments and preparation of summary reports is a lengthy process, with input for many experts and therefore takes some time.
5	Inconsistent approach to EQA marking, personal opinion of assessors rather than best practice.	Very occasionally there is an inconsistent approach to marking that has not been identified by assessors or the SAGs. Sometimes the marking may appear inconsistent because new guidelines have been introduced. The assessors' marking is based on professional guidelines, international standards and expertise/experience. Please contact info@genqa.org or use the appeals process so we can address any issues.

How would you rate the content of the EQA Summary Reports?



How can the content of the EQA Summary Reports be improved?

	Participant comments regarding EQA Summary Reports	GenQA response
1	Improve lay-out/structure to make it easier to find the main / important points.	Thank you for the feedback. GenQA has reviewed the format and content of the EQA Summary Report and is trialling it for a subset of EQAs early in 2020. We hope this will make finding the important items easier.
2	More focus on highlight methods/techniques that may be underperforming.	This is part of the assessment process and the Scheme reports back any issues identified to the laboratories and the manufacturer (if appropriate).
3	More examples of good reports.	GenQA will look into providing a selection of 'good' reports for an EQA. General guidance on reporting for genetic laboratories was published by the ACGS in 2015 (in collaboration with GenQA/UKNEQAS) and by ESHG Quality Committee in 2014 in collaboration with GenQA.
4	Ensure evidence based and not personal opinion.	The assessors' marking is based on professional guidelines, international standards and expertise/experience. Please contact info@genqa.org or use the appeals process so we can address any issues.

F. Additional EQAs requested

Are there any EQAs you would like GenQA to provide?

	Participant comments regarding additional EQAs	GenQA response
1	Include all tests in the NHS test directory	GenQA are working towards this.
2	BRCA1/2 predictive testing	Included as part of the Hereditary Breast and Ovarian (HBOC) EQA.
3	SMA exon 7/8 deletion	Included in Hypotonic Infant EQA.
4	MLPA analysis	Any EQA where dosage testing is required.
5	PMS2	Included in the Familial Colorectal Cancer and Polyposis EQA.
6	UPD for chromosomes 7 and 14	Included in Imprinting EQA.
7	ALK	Included in Lung Cancer EQA.
8	MSI	Included in MSI and Colorectal Cancer EQA.
9	EGFR	Included in Lung Cancer EQA.
10	AML	Included in Myeloid Disorders and Technical FISH EQAs.
11	CML	Included in Myeloid Disorders and Technical FISH EQA.
12	IGHV mutation screening	Available as a pilot EQA in 2019.
13	Y microdeletions by MLPA	Included in the online Infertility EQA.
14	Quantification of validated concentration samples	Available as DNA quantification EQA.
15	ApoE genotyping	Not provided as this is a risk factor only but GenQA will review this.
16	Pathogen identification CMV	This is offered by a different UK NEQAS provider. Please contact office@ukneqas.org.uk
17	cfDNA/ctDNA	Available as the cfDNA testing in lung cancer.
18	ELISA	This is offered by a different UK NEQAS provider. Please contact office@ukneqas.org.uk
19	Variant interpretation and reporting for cancer panels	Included in the Molecular Pathology EQAs.
20	Array CGH	Included in the following EQAs: Prenatal CNV detection, Postnatal CNV detection, Pregnancy loss molecular or Acquired array.
21	Array on chorionic villus	Included in Prenatal CNV detection.
22	RPGR ORF15	Thank you for your suggestion which GenQA will review.
23	Segmental Overgrowth (PTEN, PIK3CA)	Thank you for your suggestion which GenQA will review.
24	*MPL1, CALR, MYD88*	MYD88 is offered by a different UK NEQAS provider. Please contact office@ukneqas.org.uk
25	De novo variant detection - PGT	Thank you for your suggestion which GenQA will review.
26	FISH5 probe for PGT-A	Thank you for your suggestion which GenQA will review.

	Participant comments regarding additional EQAs	GenQA response
1	The range of tests performed in each saliva run has changed from run to run, and some of the more appropriate tests have been dropped in favour of broader criteria.	The Sample Handling SAG is currently reviewing the quality metric measurements for all of the DNA extraction EQAs to standardise the approach.
2	More disease based with choice of techniques.	GenQA is offering more EQAs which follow clinical indication/patient pathways. More such EQAs will be introduced. Some centres still require a sample based EQA so these will continue.

J. Improvements

GenQA is constantly improving the standard of EQA provision and would welcome any comments or suggestions on how GenQA could improve the service in the box below.

	Participant comments regarding improvements	GenQA response
1	Staggering of EQA sample distribution due to difficulty managing the usual workload, urgent samples and processing a number of EQA schemes with the same deadline, especially if they are all processed by the same team.	GenQA frequently reviews the EQA timetable and it is difficult to balance the number of EQAs with keeping the costs as low as possible. To spread out the distributions for similar EQAs would increase the costs of providing the EQA considerably and would impact the laboratories. If there are particular EQAs which you have issues with then please contact the Scheme so we can review the timetable.
2	More clinical information especially in prenatal e.g. ultrasound scan, parents karyotype images	The EQA cases are based on real patient cases and the information given reflects what the genetic centre received. GenQA is aware that different countries/hospitals may provide additional information. Parental karyotypes are not done at the same time as a prenatal invasive technique in many countries
3	Less errors in your documentation that are clearly copy / paste	GenQA are reviewing the format of their distribution documentation to reduce the number of copy/paste errors.
4	Deductions for not including specific markers (QF-PCR) when we state in our report that we do not usually specify it, but that information can be provided to referrer if requested.	The MRA EQA follows the guidelines which require this information in the report.
5	Please seal the slide containers as sometimes they arrive open in the post.	Thank you for this feedback. GenQA will review and audit the process.
6	Images of marked H&E slides would be better than having to send slides to our histopathologists for staining and marking.	Thank you for your suggestion. GenQA will review this option.
7	Provision of a certificate of successful participation, without Score and Comments.	At the end of the EQA year, a certificate of performance is provided. For accreditation purposes, a certificate of participation is no longer available as this gives no indication on whether a laboratory obtained a satisfactory or a poor performance.
8	Faster reporting back.	Thank you for this feedback. GenQA is trying to reduce the 'reporting back' time interval.
9	Zoom in of markers in PCR trace images would be helpful.	Thank you for your suggestion. GenQA will review this option.
10	Website needs improvement as it is non-user friendly.	Please contact the Scheme as we would like to understand how the website could be improved.

11	General principles of EQA schemes are in my opinion 1. Frequent distributions 2. Rapid feedback. 3. Cumulative scoring system. 4. Informative report 5. Samples and scenarios that resemble real clinical cases. 6. Reliable standard. GenQA doesn't meet the first 3. Persistent poor performance will take 3 years to identify in most GenQA schemes which is unacceptable.	Thank you for this feedback. We will review this internally.
12	Format is outdated – testing by sample type rather than clinical indication.	GenQA is offering more EQAs which follow clinical indication/patient pathways. More such EQAs will be introduced. Some centres still require a sample based EQA so these will continue.
13	We participated in the variant interpretation exercise. The variation of variants was good.	Thank you for this positive feedback.
14	No suggestion, thanks for a great service, keep up the good work.	Thank you for this positive feedback.

Conclusion

GenQA would like to thank all the participants who took the time to complete the survey. If you would like specific feedback to individual comments or have further comment, then please contact us at info@genqa.org.