Introducing GENie, a genomics online individual education platform

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Introduction

Clinical genomic testing relies on an accurate classification of the pathogenicity of genomic variants, and the assessment of their clinical impact for each patient. Although more guidance is being generated to assist healthcare professionals with this, there is a challenge to standardise approaches across variant types and in different clinical settings.

GenQA, the only end-to-end External Quality Assessment (EQA) provider in genomics, has globally delivered laboratory-based EQAs for variant interpretation since 2013. The ISO15189[1] standard requires demonstration of the competency of individuals to perform specific tasks. Therefore, GenQA has adapted the EQA approach to provide a mechanism for **individuals to demonstrate their competency**, and to support the education of the scientific and clinical workforce through the **GenQA Genomic Individual Education (GENie) platform.**

Methods

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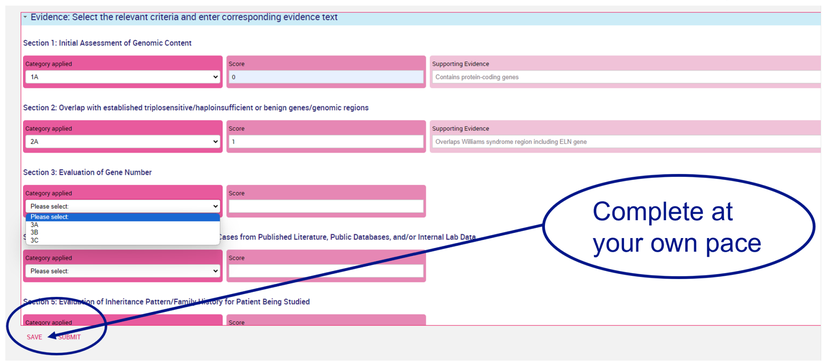
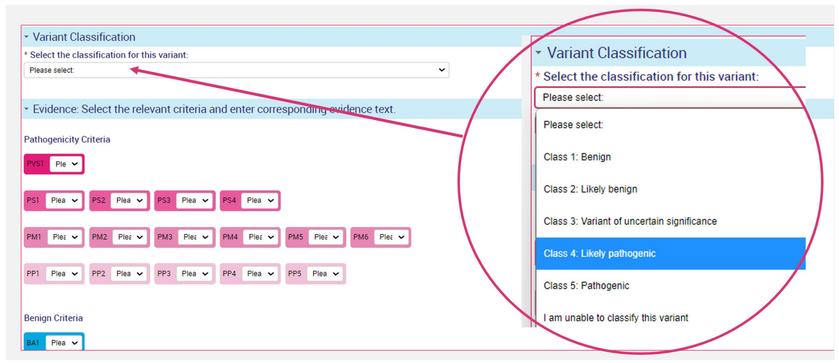
- The Genomics Individual Education (GENie) platform was launched as a pilot trial in June 2023 and as a full module in August 2023, offering online modules for the classification of single nucleotide variants (SNVs) and copy number variants (CNVs).
- The vast bank of variants includes prenatal, postnatal, diagnostic, and predictive clinical scenarios. Panels of expert advisors classified the SNVs according to the ACMG[2] and ACGS[3] guidelines and classified the CNVs according to the ACMG/ClinGen guidelines[4].
- Details of a randomised set of six variants and their clinical settings are provided per assessment. Individuals are expected to classify the variants along with documenting the evidence used to support the classifications.
- Real-time assessment provides performance feedback along with details of the expected variant classifications and associated evidence. Individuals can re-take the assessments as required.
- GenQA and EMQN have also provided global annual or biannual online individual competency assessments for the classification of *BRCA1*, *BRCA2* and other HRR (Homologous Recombination Repair) gene variants since 2017. These modules are now available on GENie.

Results: Variant classification modules

Figure 1 – Screenshots of the GENie variant classification modules and Participation Certificate available on completion of each assessment. Performance feedback is also provided.





Trial Participation

Over six weeks, 295 individuals completed the SNV module and 334 individuals completed the CNV module. Figure 2 summarises the number of times participants completed a set of variant classifications. Figure 3 shows the number of sets of variants participants completed. The majority completed one set for both the SNV and CNV modules.

Figure 2 – Number of times participants completed a set of variants

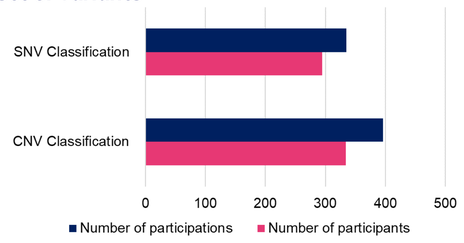
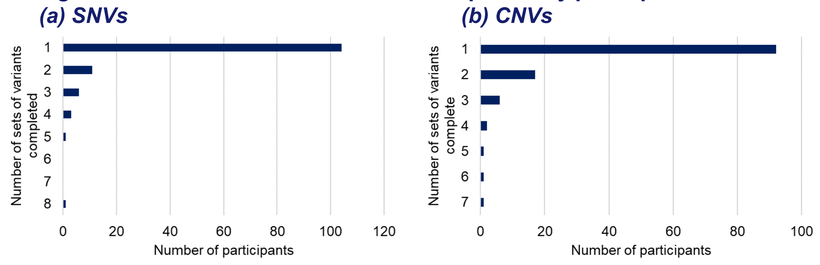


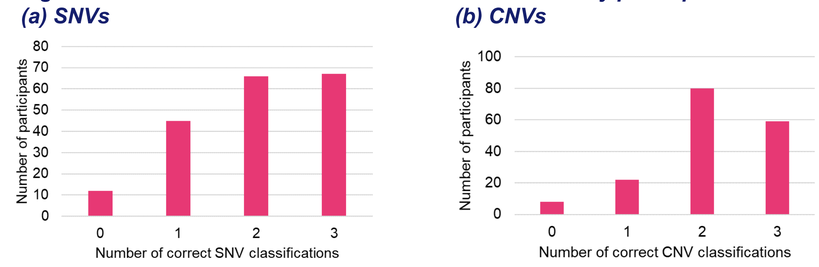
Figure 3 – Number of sets of variants completed by participants
(a) SNVs (b) CNVs



Performance

The majority of individuals correctly classified 2 or 3 out of the 3 variants in the SNV module. The majority of participants classified only 2 out of the 3 CNVs (Figure 4).

Figure 4 – Number of correct classifications submitted by participants
(a) SNVs (b) CNVs



Results: BRCA and HRR

Participation

Annual or biannual online individual competency assessments for the classification of *BRCA1*, *BRCA2* and other HRR (Homologous Recombination Repair) gene variants are available on the GENie platform. To date, **~1600 individuals** have participated from **over 60 countries** worldwide, including medics, pathologists, scientists and students (Figures 5, 6 and 7).

Figure 5 – Participants come from a range of specialties/ departments

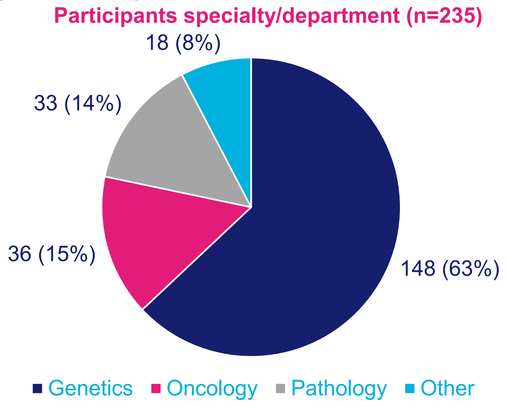


Figure 6 – Participants have a broad range of expertise

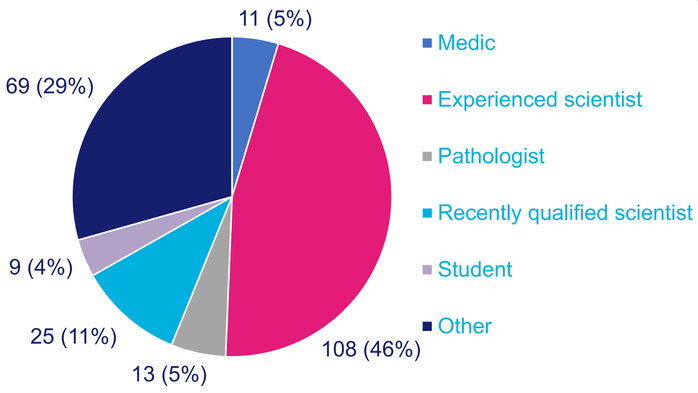
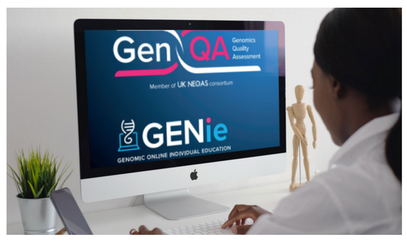


Figure 7 – Participants in Run 11 have come from a wide range of countries, with those more frequently represented shown here in larger font



Performance

Table 1 lists the genes assessed and the total number of individual submissions in the most recent of the 11 Runs to date. The proportion of individuals who attempted all variants in a Run and correctly submitted all of the expected classifications is also shown. Less than 5% of participants attained the expected classification for all variants within several assessment runs. This data demonstrates that there is a continued need for a tool to educate individuals and promote standardisation of variant classification.

Guidelines

There is variability in the guidelines routinely used by participants for the classification of these variants (Figure 8). There is also variability in the application of the same guidelines to the same variant, resulting in inconsistent classifications across participants.

Figure 8 – Various guidelines are used by participants for classifying these variants

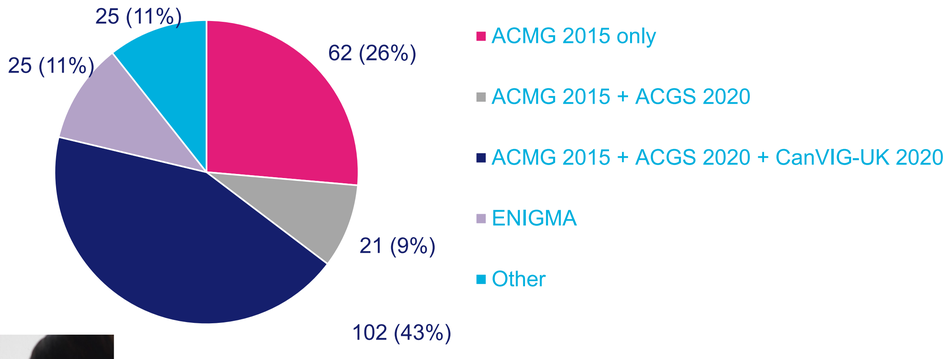


Table 1 – Summary of participant numbers in recent BRCA-HRR assessments

Year	Run	Assessment	Total number of participants	Participants successfully classifying all six variants
2023	**11**	BRCA and HRR	322	17% (42/235)
2023	**10**	BRCA and HRR	463	0 (0/374)
2022	**9**	BRCA and HRR	369	1% (5/316)
2022	**8**	BRCA and HRR	400	13% (53/354)
2021	**7**	HRR only	289	4% (11/289)
2021	**6**	*BRCA1/2* only	440	1% (5/440)
2020	**5**	BRCA and HRR	240	2% (6/240)

References: 1. ISO 15189:2012. Medical laboratories – requirements for quality and competence.
2. Richards *et al.*, 2015 PMID: 25741868
3. Ellard *et al.*, 2020 (https://www.acgs.uk.com/quality/best-practice-guidelines/)
4. Riggs *et al.*, 2020. PMID: 31690835

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Conflicts of interest: The authors have no conflicts of interest to declare.

Conclusion

There continues to be variability in the approach and classification of genomic variants. Educational modules such as GENie can deliver assessment for individuals to promote good practice and standardisation.

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