Clinical genetics and genetic counselling external quality assessment (EQA) provision

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GenQA Genomics Quality Assessment
Member of UK NEQAS consortium

Introduction

Clinical Genetics educational case scenarios were introduced by **GenQA** in 2014 and complete the exclusive provision of **External Quality Assessment (EQA)/Proficiency testing (PT)** across the entire clinical genomics service from patient counselling, sample preparation, testing processes, result interpretation and reporting.

Annual EQA programs for all Clinical Genetics disciplines are available and easily accessible on a bespoke website. Pilot EQAs for Inherited Metabolic Disorders and Genomic multi-disciplinary team (MDT) working are offered for the first time in 2023. Clinicians from across the world can therefore participate in a customised EQA program and review their clinical practice, learn from peers and demonstrate continuing professional development. Satisfactory participation evidences training to a recognised level and demonstrates ongoing professional competency, building public confidence and ultimately improving patient care.

Methods

Annual online multi-stage case scenarios which follow the patient pathway and **reflect real clinical cases** are provided for each EQA (*Table 1*).

Clinical genetics EQAs: These programs focus on the correct selection of genetic testing based on the clinical presentation and family history and then the correct interpretation of test results to provide a clinical diagnosis.

Genetic counselling EQA: This program was introduced in 2021 and has an emphasis on provision of relevant genetic counselling.

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Anonymised submissions are marked according to professional standards by an expert panel of international clinicians and a tailored score report with feedback comments is then provided to each participating centre. A participation certificate is awarded to each centre showing the performance status (where applicable) for each program.

The detailed EQA Summary reports provide the expected learning outcomes and recommendations for the clinical scenario and give an anonymised overview of all submissions. These reports may be used by participating centres for educational purposes, internal review and quality improvement. GenQA offers support to all centres with sub-optimal performance.

Why is EQA important for clinical teams?

What are the benefits?

- Patient safety: If a clinical geneticist does not recognise the clinical features of a disorder, they may request the wrong tests and the disorder/disease is not diagnosed;
- Do the laboratory results adequately explain the clinical presentation? Professional judgement is required to correctly interpret results;
- Patients receiving an incorrect diagnosis or prognosis may receive compromised treatment and therefore an impact on mental wellbeing;

- Harmonise and benchmark the quality of clinical services;
- Give an overview of/refine best practice (surveillance role);
- Promotes quality across national borders;
- Ensure that families are counselled appropriately and in the same way;
- Demonstration of excellence, builds public confidence: external verification of clinical services;
- Identifies imperfect practice and gaps in internal quality management systems;
- Improves quality of care;
- Training and maintenance of competency CPD/CME.

Results

Participation has steadily increased over the years and a total of 105 submissions were received from 27 countries for the 2022 EQAs (*Figure 1*). An **EQA participation fee** was introduced **in 2017**.

A total of six **critical errors** have been identified since performance criteria were introduced in 2021: five critical clinical diagnostic errors for missed diagnoses due to incorrect genomic test requests and one critical counselling error where an incorrect recurrence risk for an X-linked disorder was described.

Figure 1 – Chart of participant numbers since 2014 by EQA

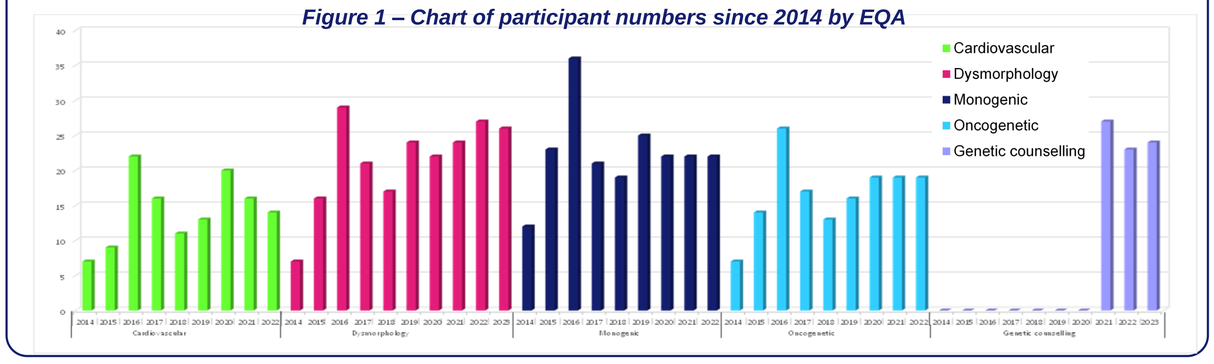


Table 1 – Summary of EQA case scenarios and critical errors

Year	EQA				
	Cardiovascular Disorders	Dysmorphology	Monogenic Disorders	Oncogenetic Disorders	Genetic Counselling (Pilot)
2023	*Opens on 6th November*	Noonan-like syndrome (1 critical error*)	*Opens on 6th November*	*Opens on 6th November*	Turner syndrome
2022	Noonan syndrome	Kabuki syndrome	X-linked female-limited myopia-26 (1 critical error)	HBOC (*PALB2*)	Cystic fibrosis
2021	Brugada syndrome	Angelman syndrome	Hypomyelinating leukodystrophy (4 critical errors)	MEN1 syndrome	HBOC (*BRCA2*)
2020	Loeys-Dietz syndrome	Cornelia De Lange syndrome	Pompe disease	PTEN hamartoma tumour syndrome	
2019	Hypertrophic cardiomyopathy (*MYBPC3*)	Mowat-Wilson syndrome	HHT with juvenile polyposis	Li Fraumeni syndrome	
2018	Holt-Oram syndrome	Williams syndrome	Stickler syndrome	Hereditary melanoma	
2017	*TSC2/PKD1* contiguous gene syndrome	Turner syndrome	Autosomal recessive polycystic kidney disease	Von Hippel-Lindau syndrome	
2016	Barth syndrome	CHARGE syndrome	MUTYH polyposis	Neurofibromatosis	
2015	Duchenne muscular dystrophy	Fragile X	Deafness	HBOC	
2014	Marfan syndrome	Wolf-Hirschhorn syndrome	Cystic fibrosis	Lynch syndrome	

Performance criteria were introduced in 2021 (not applied to Pilot EQAs)

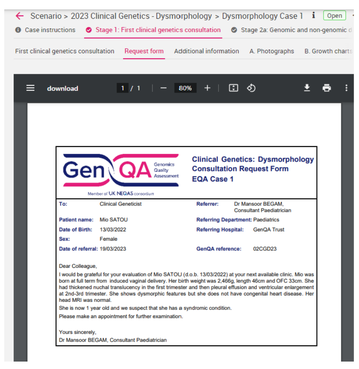
HBOC: Hereditary breast and ovarian cancer syndrome

**Pending ratification*

Example EQA case scenario – Dysmorphology 2023

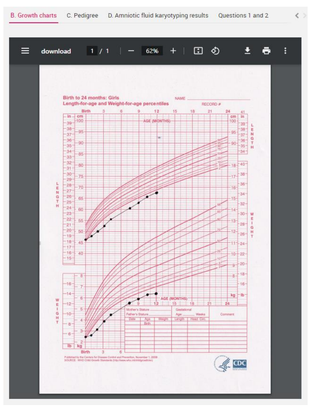
Stage 1: First clinical genetics consultation

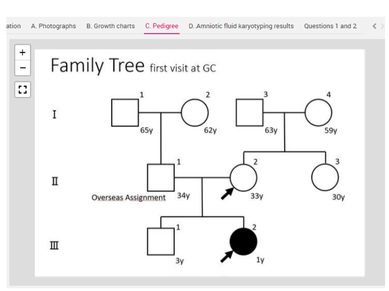
Information regarding the clinical presentation of the proband is provided on the website:



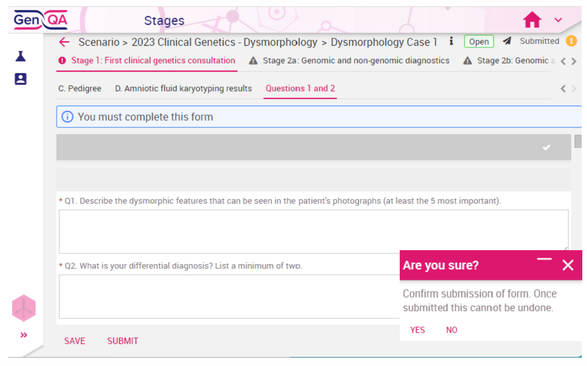
- Request form
- Additional clinical details are also provided

- Clinical information provided is specific to the case scenario





- A family pedigree may be given or a question asked about how a family/medical history should be taken



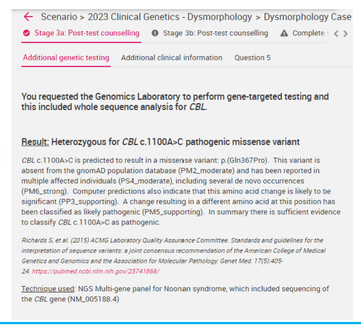
- Participants are asked to describe for example dysmorphic features, and/or suggest candidate genetic disorders or give differential diagnoses

Subsequent stages are blocked until answers are submitted

Stage 2: Genomic and non-genomic diagnostics

- Further clinical information is provided;
- Questions asked include genomic and non-genomic testing recommendations.

- Test results can then be accessed on the website;
- There may be further questions regarding follow-up testing.



Stage 3: Post-test counselling

- Questions asked cover the discussion topics concerning the consequences of the genetic diagnosis (*i.e.,* prognosis, co-morbidity, treatment and surveillance, if relevant);
- Participants are asked what information should be given to the proband/family *e.g.,* mode of inheritance, recurrence risk, reproductive options available.

The scenario ends with an option to submit any comments for the assessors *e.g.,* regarding local/national reporting policies.

Patient consent: Consent is obtained from patients for their clinical photographs and information to be used for EQA. Full clinical photographs are available to EQA participants only. Patient data is fully anonymized.

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

Conclusion

Optimal patient care and **promotion of best practice** is facilitated by these EQAs, which offer a **valuable educational opportunity** for Clinical Geneticists and Genetic Counsellors. Regular EQA participation has resulted in improvement in the accuracy of answers and the overall scores obtained. These programs also provide a mechanism for the **continuous assessment** of trainees against **agreed quality standards.**

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