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.

These EQAs were developed at the request of the **ESHG Genetics Services Quality Committee** to address a need for a European Quality Assessment scheme for Clinical Genetics and counselling. Satisfactory participation in these EQAs evidences training to a recognised level and demonstrates ongoing professional competency.

Clinicians across the world can now review their clinical practice, learn from peers and demonstrate continuing professional development.

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: 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: More emphasis on provision of relevant genetic counselling.

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An expert panel of international clinicians, including **ESHG and EBMG board and committee members** mark the anonymised submissions based on professional guidelines and a tailored score report with feedback comments is provided to each participating centre. A participation certificate is awarded to each centre showing the performance status (where applicable).

The EQA Summary report provides an overview of the submissions and details the expected learning outcomes and recommendations for the clinical scenario. This may be used by participating centres for internal review and quality improvement. Centres with sub-optimal performance are offered support by GenQA.

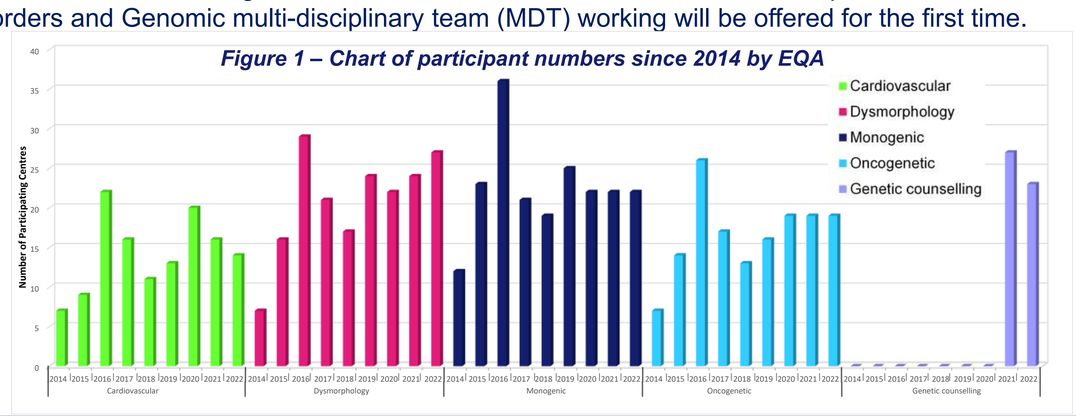
Results

Participation

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**.

The Genetic Counselling Pilot EQA was introduced in 2021. In 2023, pilot EQAs for Metabolic disorders and Genomic multi-disciplinary team (MDT) working will be offered for the first time.

Figure 1 – Chart of participant numbers since 2014 by EQA



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.

Table 1 – Summary of EQA case scenarios and critical errors

Year	EQA				
	Cardiovascular Disorders	Dysmorphology	Monogenic Disorders	Oncogenetic Disorders	Genetic Counselling (Pilot)
2022	Noonan syndrome	Kabuki syndrome	X-linked female-limited myopia-26	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	

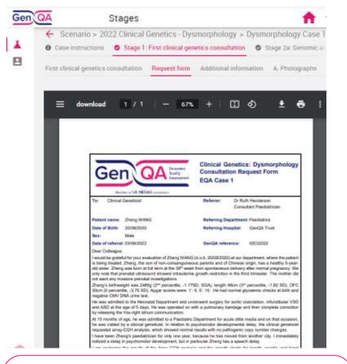
Performance criteria were introduced in 2021 (not applied to Pilot EQAs)

HBOC: Hereditary breast and ovarian cancer syndrome

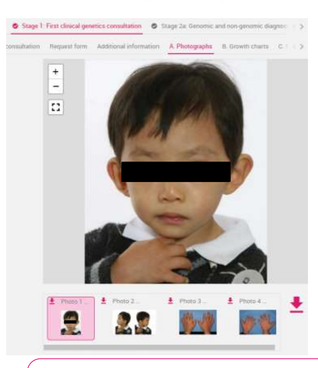
Example EQA case scenario – Dysmorphology 2022

Stage 1: First clinical genetics consultation

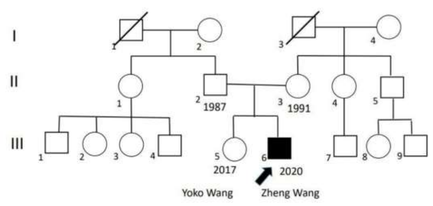
Information regarding the clinical presentation of the proband is provided on the website:



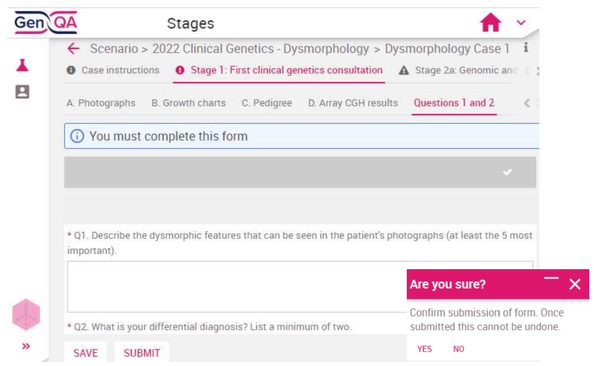
❖ Request form
❖ Additional clinical details are also provided



❖ Clinical information provided is specific to the case scenario



❖ Family pedigrees may be provided or questions are asked about how a family/medical history should be taken



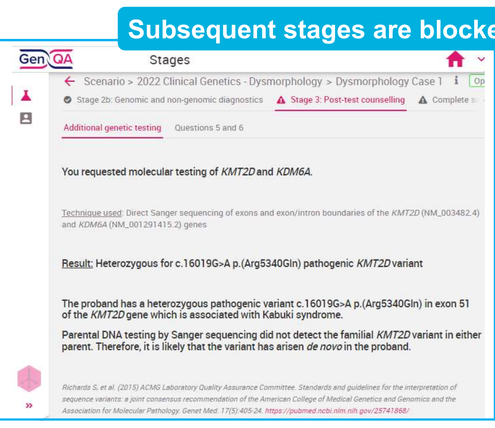
❖ Participants are asked to describe *e.g.,* dysmorphic features, and/or suggest candidate categories of genetic disorders or differential diagnoses

Subsequent stages are blocked until answers are submitted

Stage 2: Genomic and non-genomic diagnostics

❖ Further clinical information is provided;
❖ Questions asked cover 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 anonymised.

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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.
These EQAs also provide a mechanism for the **continuous assessment** of trainees against **agreed quality standards** (a **requirement** of the **European Union of Medical Specialists**).

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