

Genetic Counseling and Education

AB022. External quality assessment of clinical genetics: from pilot assessment to full external quality assurance scheme

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Background: Quality assessment has long been associated with laboratory, but not clinical services.

Methods: To address this obvious gap, CEQAS provided an external quality assurance (EQA) for clinical genetics and genetic counselling services. A working group prepared four case scenarios in the fields of cardiogenetics, oncogenetics, monogenic disorders and dysmorphology. Each scenario started with a referral letter and consisted of multiple stages, to reflect an episode of clinical care. At each stage further clinical and genetic test information was given and a number of questions presented. For each EQA, consensus answers were provided by at least two clinical

geneticists and a patient organization was also consulted. Twelve pilot EQAs were distributed over the last three years and in 2016, to 57 genetic centres across 25 countries. All EQA submissions were reviewed by two assessors per case. The EQA for genetic counselling was feasible and highly educational. Learning objectives were included the summary letter.

Results: The EQA identified that some centres omitted to include the need for the following: (I) for consent of the index patient; (II) for a three generation family history to be taken; (III) to discuss the implications of a recessive disorder; (IV) to give comprehensive reproductive options; (V) for psychosocial support for the patient and family. The EQA for clinical genetics proved to be highly educational. The answers provided by the centres highlighted differences in clinical genetics practice across Europe. In addition, the assessors identified that some case scenarios tried to cover too many aspects and needed to be streamlined so that only 3–4 learning points were covered. Data from the pilot EQAs and some of the genetic counselling issues identified will be discussed.

Conclusions: This EQA is currently run by CEQAS as a standard EQA, but the working group will further improve the process whenever necessary.

Keywords: Quality assessment; clinical genetics; genetic counselling

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