

The cystic fibrosis external quality assessment scheme, monitoring the quality of laboratory performance

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1. Introduction – Aim of the study

Given the potential serious health consequences of genetic test results, **mechanisms** should be in place **to assure the quality of the tests and the interpretation of the data.**

In this regard, the International Organization for Standardization Standard **ISO 15189** and the Organisation for Economic Co-operation and Development (**OECD**) **Guidelines for quality assurance** in molecular genetic testing, contain **requirements and recommendations** for laboratories **to participate in external quality assessment schemes (EQA).**

The **European Cystic Fibrosis Network** has been providing EQA schemes since 1996. This study focuses on the **comparison of the CF EQA scheme results of all participating laboratories over the previous three years:** 2005 (191 labs), 2006 (208 labs), 2007 (212 labs), including a group of 152 laboratories **that participated in the CF EQA scheme for each of the previous three years**

Does participation in **EQA monitor and/or improve quality** in genetic testing laboratories?

2. Results

ISO 15189 clause 5.6.4 requires that a laboratory:

- participates in **inter-laboratory comparisons** such as those organized by EQA schemes
- monitors the results of EQA**
- participates in the **implementation of corrective actions** when control criteria are not fulfilled

- Only laboratories that provided **genotype results and interpretation** are included in the study.
- Participation in the CF EQA scheme **has increased** year after year (**Figure 1**): 191 labs in 2005, 208 in 2006 and 212 in 2007.
- Participating laboratories are situated in **35 countries** in Europe and even a small number in the United States (6 labs in 2007) and Australia (11 labs in 2007).
- About 50% of the laboratories that participate in the CF EQA scheme **analyse 10-250 samples per year** (**Figure 2**).
- The number of laboratories participating in the CF EQA scheme and have achieved **accreditation or certification has increased slightly year after year** (**Figure 3**).

Figure 1: Number of laboratories that submitted genotypes and interpretation results for the CF EQA scheme in 2005, 2006 and 2007

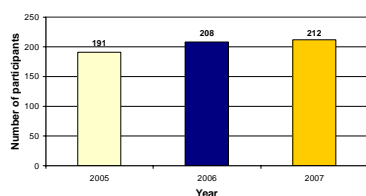


Figure 2: Percentage of samples analysed per year by laboratories that participate in the CF EQA scheme in 2005, 2006 and 2007

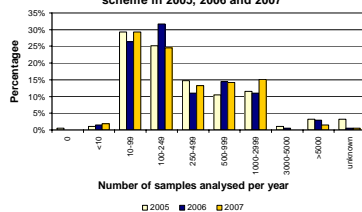
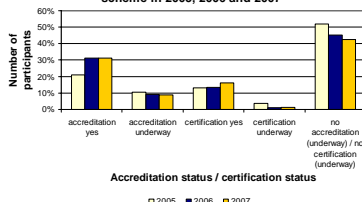


Figure 3: Accreditation status / certification status of laboratories that participated in the CF EQA scheme in 2005, 2006 and 2007



ISO 15189 clause 5.8.3 requires that reports include, but not be limited to, the following:

- unique identifier of the **requester**
- unique identification and location of the **patient**
- date and time of **primary sample collection**
- primary sample type**
- date and time of **sample receipt** by the laboratory
- interpretation** of the results
- date and time of the release of the **report**
- signature** of the person checking or releasing the report



Figure 5: Percentage of 'elements required by the ISO 15189 standard' included in the reports of laboratories that participated in the CF EQA scheme for each of the three previous years (152 labs)

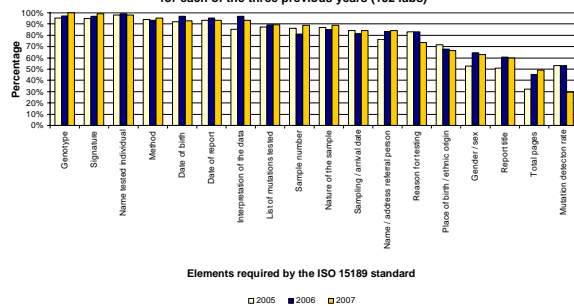
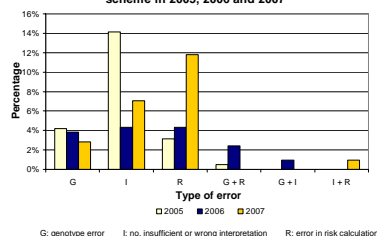


Figure 4: Percentage of error types made by laboratories that participated in the CF EQA scheme in 2005, 2006 and 2007



- Genotyping errors have decreased** over the years (**Figure 4**).
- On the other hand, errors in **interpretation** and **risk calculation have increased**.
- Errors made for interpretation and risk calculation are **related to the complexity of the EQA cases**.
- Moreover, the **overall assessment criteria** for CF EQA scheme **has become more stringent over the years**.
- In the **beginning** of the scheme **focus** was mainly on **genotyping**, while reporting and interpretation is now assessed as stringently as genotyping.
- The trend for interpretation and risk calculation errors should be monitored carefully and the task of the assessors is to **inform and educate laboratories in order to decrease these type of errors**.

- The elements required by the ISO 15189 standard were assessed only for **laboratories (152) that participated in the CF EQA scheme three consecutive years (Figure 5)**.
- When an element was assessed as **wrong or not clearly defined** in the laboratory report this element was not counted as present in the report and thus **not counted for this graph**.
- '**Mentioning the mutation detection rate**' decreased in 2007. This is due to the fact that a mutation detection rate for a **population other than the one of the patient** has been assessed as **not correct** since 2007.
- Mentioning the **genotype**, including a **signature** and **name of the referral person** has continued to **increase** over the years.
- Reason for testing, gender and ethnic origin of the patient is not included in about 30% of the genetic reports.**

3. Conclusions

- The **increase of participation in EQA** and the **increase of accreditation / certification status** indicates that genetic testing laboratories are working on the improvement of their quality systems.
- Participation in EQA schemes is a **tool to monitor quality improvement** and to **educate laboratories** by assessors, aided by making the assessment criteria more stringent.

- When the presence of certain elements decreases, this is mainly due to a **more stringent assessment**.
- The overall tendency of small increases or 'no changes', in the presence of elements required by ISO 15189, indicates that **laboratories have improved their reporting year by year**.