

Eleven years experience of being under external quality assessment for the molecular genetic diagnosis of hereditary haemochromatosis HFE-associated and accreditation under ISO 15189

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Laboratory quality is continuously present in our daily practice of molecular diagnosis in human genetics. Our participation in external quality assessment (EQA), specially with EMQN, gave us the opportunity to improve the reports of our genetic tests, to compare our performance/scores with other European laboratories, and to be permanently updated with the most recent recommendations for best practice in human molecular diagnosis.

Since 2005, our lab has been participating in the EMQN external quality assessment program for the molecular diagnosis of hereditary haemochromatosis HFE-associated (HH-HFE). Since then, our score regarding the genotyping category has achieved every year the highest marks (2.0), while for the interpretation category, in two years, the mean score was 1.75. These scores lead us to improve the “interpretation section” of our reports in order to give the best result to the physicians and patients. In addition of a correct genotyping, the report “interpretation section” is particularly important, it should include the main suggestions for the best patient’s clinical management: i) immediate impact of the result for the patient, ii) patient’s clinical follow-up and other diagnostic options, iii) long term-impact (specially in predictive tests), iv) relevance of the result for relatives and, v) recommendations of genetic counselling.

In accordance with OCDE disease specific guidelines for quality assurance in molecular genetic testing, and with the requirements of ISO 15189, in 2014 we were the first Portuguese laboratory accredited by IPAC, for HH-HFE - variants p.H63D and p.C282Y, and other genetic tests (http://www.ipac.pt/pesquisa/ficha_15189.asp?id=E0015). Accreditation under the International Standard ISO 15189 is challenging but contributes to introduce improvements in our current practice because it comprises “management requirements” (e.g. quality management system, external services and supplies, preventive and corrective actions, control of records) and “technical requirements” (e.g. accommodation and environmental conditions, laboratory equipment, reagents and consumables, training and qualifications of personnel, examination processes, results reporting). Accreditation enhances laboratory quality at different levels, gives credibility, competency and confidence, but primarily contributes to a better patient’s clinical diagnosis reducing the turnaround time, patient management and treatment, and genetic counselling.