

QUALITY CONTROL

EQA: a perspective from a participating laboratory

Marta Rodríguez de Alba



No Medical Genetics Specialty

Recent legislation (at a regional level):

- * Programme in Genetic Counselling (Andalucia)
- * Regulation of the requirements to be Head of a Genetic Service/Lab (Madrid)

At a national level:

- * June 2007 _ LAW FOR BIOMEDICAL RESEARCH *'Genetic analysis will be done with appropriateness, **quality**, equity, and accessibility'*
- * Near future_ Committee that will settle the 'National strategy for rare diseases in the National Health System' (Quality Agency of the Ministry of Health)

No specific regulation for Genetic Services concerning the recommendation/obligation to take part in EQAs

Two existing Spanish EQA on Medical Genetics

- For haematological cytogenetics under the auspices of the Spanish Association for Haemathology
- For constitutional cytogenetics under the auspices of the Spanish Association for Prenatal Diagnosis
- Both have guidelines
- Both are voluntary



Three spanish members from both assesment bodies are assessors for CEQA
(Eurogentest)

Our lab has been doing the Spanish Constitutional Cytogenetics EQA since the first running



➤ CEQA: Pilot since 2006 (2 assesments to date)

➤ CF Network

➤ EMQN

Charcot Marie Tooth disease

Duchenne Muscular Dystrophy

Fragile-X syndrome

Friedreich ataxia

Hungtinton disease

Haemochromatosis

Prader Willi/Angelman Syndrome

Spinocerebellar ataxia's

Y-chromosome microdeletion

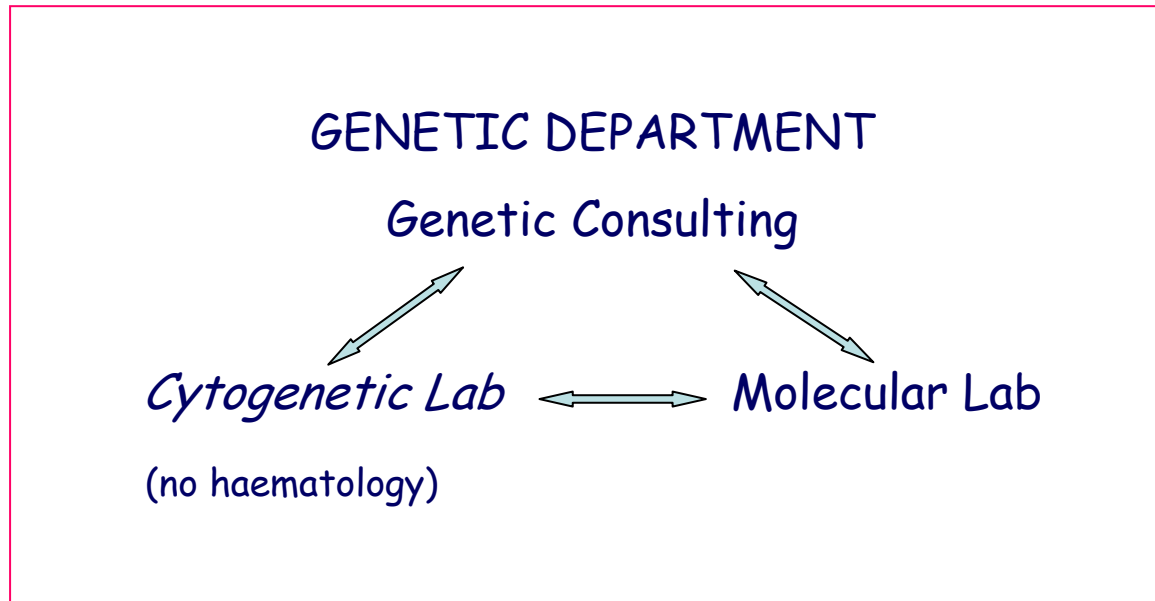
DNA Sequencing

Myotonic Dystrophy_ in the near future (now pilot and restricted)

Since 2000

Why are we involved in European schemes?

- Not mandatory, optional
- Lack of Spanish schemes



To improve quality

What aspects of European EQA are important to us?

- Existence of *guidelines*
 - Helps to improve National EQAs
 - The possibility to exchange information with a committee of experts
-
- We have increased the quality of the analysis
 - Harmonization of reports

What we have we
learned from EQA

Improvement in quality of the analysis

- Permanent evaluation and adjustment of the procedures to the quality guidelines: cytogenetics and molecular genetics
- Incorporation of the new diagnostic strategies to improve the analytical validity
- PD is performed at least by two different methods: direct + direct // direct + indirect
- The use of the indirect method also tests the purity of the CVS

Harmonization of reports

Correct and complete interpretation

RESULT:

- * correct use of the ISCN nomenclature

CONCLUSION:

- * result consistent with certain syndrome and referral indication

CLINICAL VALIDITY

- * should say 'normal alleles' mean 'patient will not develop the disease'
- * study for a recessive disease for a patient whose partner is already known to be a carrier, add the reference of the known carrier to the report
- * specify what is excluded in the analysis 'both, paternal and maternal alleles show normal methylation pattern'_ excludes all mechanisms for PWS

EuroGenetest



The European Molecular Genetics Quality Network



European
HUMAN
GENETICS
Conference 2008