Belgian guidelines for the minimal frequency of participation to EQAs in the context of hereditary rare diseases

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Introduction: Rare diseases in numbers

distinct rare diseases identified

Each one affects

< 1 in 2,000

people

30 million people

affected by a rare disease in Europe (as patient or family member)

80 % with a genetic origin

No Cure for the vast majority

50 % affect **children**

Commission of the European Communities, Communication from the Commission to the European Parliament, the Council, the European Economic and Social Committee and the Committee of the Regions on Rare Diseases: Europe's challenges, 2008

Introduction: Europe and rare diseases

Actions of the European Union:

- ☑ Improving access to diagnosis, information and care for affected patients
- ☑ Ensuring adequate traceability and coding of rare diseases in health information systems
- ☑ Supporting national plans for rare diseases in EU countries
- ☑ Creating European Reference Networks linking patient and professionals in different countries to share expertise

Introduction: Belgian Plan for rare diseases

Supporting national plans for rare diseases in EU countries

❖ Development of a Belgian Plan for rare diseases

- Improvement of the access to diagnosis, information and knowledge for patients

⇒ **strengthening the role of Centers of Human Genetic in the areas of diagnosis, genetic counselling and quality management**

- Care optimisation

- Monitoring the implementation and durability of the measures

L. Onkelinx, "Belgian Plan for rare diseases," Ministère fédéral des Affaires sociales, de la Santé publique et de l'Environnement., 2013

Method: based on "RAND/UCLA appropriateness method"

Preliminary phase

- Rare diseases related EQAs inventory
- Survey about the BCHGs participation to the inventoried EQAs

February - June 2018

Development phase

- Creation of a working group of experts from the BCHGs
- Development of guidelines

July 2019 – April 2019

Validation phase

- Guidelines revision (Belgian College of human genetics and rare diseases; Belgian accreditation body)

April – July 2019

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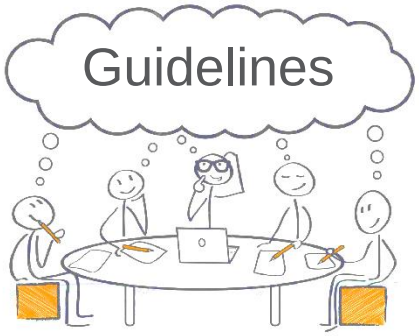
K. Fitch, Ed., "The Rand/UCLA appropriateness method user's manual". Santa Monica: Rand, 2001

Objective: Harmonization

Development of national guidelines about the minimal frequency of participation to EQA schemes focused on hereditary rare diseases

- (i) instructions on the required frequency of participation to EQAs schemes focused on hereditary rare diseases
- (ii) harmonized European framework for the frequency of quality assessment

Scope of the guidelines



➔ 75 EQA schemes for hereditary rare diseases related to

I. Analytical techniques = method-based EQA

II. Non-invasive prenatal testing schemes and preimplantation testing schemes = NIPT and PGT EQA

III. Correct identification of a rare disease including germline predisposition to cancers and pharmacogenomics (drug sensitivity diseases) = disease-based EQA

Scope of the guidelines (2)

Guidelines cover 4 aspects:

- General recommendations
- Particular case
- How to address poor performances ?
- Quality follow-up and surveillance

General recommendations

Technique assessment

1x / year for all techniques, PGT and NIPT

If no method-based EQA available

Disease based EQA involving the targeted technique and covering genotyping and interpretation

1 x / year if
- low annual volume of tests
- performed ≤ 5 years
- for diseases with variants heterogeneity (e.g mitochondrial diseases)

ex: if less than
- sufficient annual volume of tests
- performed ≥ 5 years
- use of a commercial CE labelled kit

Best scenario

\+

Annual participation to EQAs assessing both genotyping and clinical interpretation, even if based on virtual images

Disease assessment

1 x / 3 years for:

➔germline mutations

➔predispositions to cancers

➔pharmacogenomics

Particular case :

- For diseases-based schemes involving interpretation based on virtual images : triennial participation

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Poor performances' management and quality follow-up

Management of poor performances

Analytical or clerical errors:

➔internal investigation

➔correction

➔documentation

Genotyping or critical interpretation errors:

➔participation to an EQA the following year

➔documentation

Quality follow-up and surveillance

- regular adaptions based on:
  I. risk assessment
  II. changes of activities or infrastructure
  III. new schemes availability
- annual review and update of the guidelines

Strengths and limitations of the guidelines

- ⊕ Based on ISO norms 15189 (2012); 17043 and on ILAC* policy for participation in PT activities (2014)
- ⊕ Large scope of method and diseases-based EQAs focused on genetic testing related to hereditary rare diseases
- ⊕ Developed according to the opinions of all centers' experts involved in the performance of the analyses and quality management
- ⊕ Developed in interaction with Belgian healthcare authorities, BELAC and Belgian College of Human Genetics and Rare Diseases
- ⊖ *Ring tests* not included
- ⊖ *Specific for hereditary rare diseases*
- ⊖ *Specific for Belgium*

** ILAC: International Laboratory Accreditation Cooperation*

Conclusion

Help the BCHGs to structure their quality management system by providing recommendations on

- the frequency of participation to EQAs
- How to deal with poor performance and change management

Provide harmonization at a Belgian level

➔ May serve as a starting point for discussion at a broader level

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Thank you for your attention

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