Belgian Plan for rare diseases

By Elfriede Swinnen

Scientific Institute of Public Health
Health Services Research
Team Rare Diseases

21/11/2015
## Development of a Belgian plan rare diseases .... takes time

<table>
<thead>
<tr>
<th>Year</th>
<th>Event</th>
</tr>
</thead>
<tbody>
<tr>
<td>1983</td>
<td>Orphan Drug Act</td>
</tr>
<tr>
<td>1995</td>
<td>1st EU meeting on RD</td>
</tr>
<tr>
<td>1997</td>
<td>° Orphanet, ...</td>
</tr>
<tr>
<td>2000</td>
<td>Orphan Drug Regulation</td>
</tr>
<tr>
<td>2002</td>
<td>Rare Diseases Act</td>
</tr>
<tr>
<td>2007</td>
<td>1st Proposal for a Belgian Plan by Y. Avontroodt</td>
</tr>
<tr>
<td>2008</td>
<td>° Fund Rare Diseases</td>
</tr>
<tr>
<td></td>
<td>King Baudouin Foundation</td>
</tr>
<tr>
<td>2009</td>
<td>Council Recommendation COM 2009/C 151/02</td>
</tr>
<tr>
<td>2009</td>
<td>Unanimous approval Resolution by the Parliament</td>
</tr>
<tr>
<td>2010</td>
<td>Fund’s Recommendations for a Belgian Plan (First Report)</td>
</tr>
<tr>
<td>2011</td>
<td>Fund’s Recommendations for a Belgian Plan (Final Report)</td>
</tr>
<tr>
<td></td>
<td>Registry Rare Diseases Orphanet Belgium</td>
</tr>
<tr>
<td>2012</td>
<td>Belgian Plan Rare Diseases</td>
</tr>
<tr>
<td>2013</td>
<td>(see further)</td>
</tr>
</tbody>
</table>

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6th European Symposium on Rare Anaemias - 1st Dutch-Belgian meeting for patients and health professionals
Implementation Belgian plan rare diseases: main actors

Federal Level:

- National Institute Health & Disability Insurance
  
- Federal Public Service Health
  \((also: representing Belgium in EUCERD, ERN Board of Member states...\))
  
- Scientific Institute of Public Health
  
But certain actions responsibility of the communities
Belgian Plan Rare Diseases: 20 actions in 4 domains

Actions were selected taking into account:
- Existing relevant measures of the Belgian Cancer Plan and Plan Chronic Diseases
- Recommendations by the Fund Rare Diseases
- 59 process & outcome indicators by EUROPLAN
- 21 key recommendations by EUCERD

Domain1: “Diagnosis and Patient Information”

A1: Non-DNA analyses
A2: Quality Assurance in genetic centres
A3: Genetic counseling in expert centres
A4: Multidisciplinary consultations
A5: Patient oriented communication
A6: Europlan
Belgian Plan Rare Diseases: 20 actions in 4 domains

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Domain 2: “Optimisation of care”

A7: Concentration of expertise
A8: Expert centres for hemophilia
A9: “Functie Rare Diseases”
A10: Networks Rare Diseases
A11: New expert centres
A12: Medical nutrition
A13: Multidisciplinary electronic health records
A14: Unmet medical needs
A15: Inventory unmet needs
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Domain 3: “Knowledge management”

A16: Central Registry Rare Diseases  
A17: Orphanet Belgium  
A18: Education of care providers  
A19: Codification and terminology
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- 21 key recommendations by EUCERD

### Domain 1: “Diagnosis and Patient Information”

- **A1**: Non-DNA analyses
- **A2**: Quality Assurance in genetic centres
- **A3**: Genetic counseling in expert centres
- **A4**: Multidisciplinary consultations
- **A5**: Patient oriented communication
- **A6**: Europlan

### Domain 2: “Optimisation of care”

- **A7**: Concentration of expertise
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### Domain 3: “Knowledge management”

- **A16**: Central Registry Rare Diseases
- **A17**: Orphanet Belgium
- **A18**: Education of care providers
- **A19**: Codification and terminology

### Domain 4: “Governance en sustainability”

- **A20**: Evaluation and monitoring of the Plan

6th European Symposium on Rare Anaemias - 1st Dutch-Belgian meeting for patients and health professionals
Domain 1: “Diagnosis and Patient Information”

A1: Focus on Non-DNA analyses
   Overall objective: Improve diagnosis and follow-up of rare diseases
   Finance currently non-reimbursed (expensive) non-DNA tests
   Improve quality of care by appointing reference laboratories
   ➔ WIV-ISP is financed to perform a feasibility study and elaborate a project proposal

A2: Quality Assurance in genetic centres
   Improve quality of care by evaluation of indicators
   Improve quality of services by implementation of quality systems
   Assess demand of care
   Epidemiology
   ➔ WIV-ISP is financed to perform a feasibility study, elaborate a project proposal
**Action 1 – focus on non-DNA analyses for rare diseases**

**Current status Action 1:** Feasibility study comprising of:

- A complete list of non-DNA analyses of clinical biology prescribed by Belgian clinicians for the diagnosis and/or follow-up of rare diseases.

- A “Short-list” of non-DNA analyses eligible for reimbursement based on their clinical relevance and analytical efficiency.

- Proposal Selection criteria and procedure for future Reference Laboratories of Clinical Biology, recognized to perform the selected non-DNA analyses.

- Financing proposition for selected non-DNA analyses and future Reference Laboratories.

(*) Identification of gene mutations, measurement of the gene expression, ... are excluded.
**Objectives:**  Improve quality of care, Estimate demand of care, Epidemiology

**Current status:** Feasibility study in collaboration with the 8 genetic centres – 4 main parts

- A) Quality assurance of their laboratory activities
- B) Quality assurance of their clinical activities
- C) Database of performed genetic tests (nationally, crossborder)
- D) Registry of genetic test results (patient level)
Belgian Plan Rare Diseases: 20 actions in 4 domains

Domain 1: “Diagnosis and Patient Information”

A3: Genetic counseling in expert centres
- Better collaboration between genetic centres and (future) expert centres
- Less diagnostic delays, less mistakes
- Improve access to genetic counselling
  ➔ additional financing of genetic centres to perform the consultations

A4: Multidisciplinary consultations for diagnosis and follow-up
- Less diagnostic delays, less mistakes
- Improve access to high quality care
  ➔ conditions specified by Law!
  ➔ to be implemented by the communities
Belgian Plan Rare Diseases: 20 actions in 4 domains

Domain 1: “Diagnosis and Patient Information”

A5: Patient oriented communication
   Improve communication between patients and care givers
   Stimulate patient empowerment
   ➞ requirement 24/24 call center by expert centres

A6: Europlan
   Harmonisation of national initiatives with European strategies
   ➞ national Europlan conference in 2014
Domain 2 Optimalisation of Care

Domain 2: “Optimisation of care”

A7: Concentration of expertise

- improve coordination of care
  - introduction of care coordinator in expert centres
  - introduction of consultations specific for transition from pediatric to adult services
  - introduction genetic counselling in expert centres (Action3)

- assurance of quality of care, maintain level of expertise
  - concentration of expertise in expert centres (defined by Law!)
  - existing Belgian “reference centres” (old system) should also evolve towards expert centres

existing reference centres are

- cystic fibrosis
- hereditary metabolic diseases
- neuromuscular diseases
- pediatric nephrology
- spina bifida
- refractory epilepsy
Domain 2: “Optimisation of care”

A8: Reference centres for hemophilia

improve quality of care for patients with serious forms of hemophilia A or B, Von Willebrand or other coagulation disorders

⇒ 1 coordinating centre and 2 additional treatment centres were selected
⇒ centres sign ‘contract’ (convention) detailing conditions
Domain 2 Optimisation of Care

Domain 2: “Optimisation of care”

A9: “Functie Rare Diseases” conditions defined by Law
framework to implement multidisciplinary approach for diagnosis and follow-up
framework within which expert centres may be recognized
⇒ conditions to be fulfilled by the hospital specified by Law
  (e.g. necessary medical and paramedical personnel, available diagnostic
facilities, psychosocial services, data-management and registration, ....)

A10: Networks Rare Diseases conditions defined by Law
sharing expertise and improvement of (back) referral through
  local networks > “shared care”
  national networks of expert centres
  international/European networks of expert centres
⇒ financing of “network coordinators”

A11: New expert centres conditions defined by Law
Less diagnostic delays, less mistakes
Improve access to high quality care
Improve quality of life of rare disease patients
Identification and concentration of expertise
Domain 2 Optimisation of Care

Domain 2: “Optimisation of care”

A12: Medical nutrition
  ➔ yearly expansion of available budget for reimbursement of special medical nutrition

A13: Multidisciplinary electronic health records
  Improve communication between care givers
  Diminish mistakes by lacking information
  improve care continuity
  ➔ expert centres obliged to maintain multidisciplinary EHR for rare disease patients

A14: Unmet medical needs

A15: Inventory unmet needs
  Define priorities in this domain
  ➔ working group has been established to identify all unmet needs and elaborate evidence-based recommendations
Domain 3 Knowledge management

Domain 3: “Knowledge management”

A16: Central Registry Rare Diseases (longitudinal, prospective)
  generate (epidemiological) data on rare disease patient population for all stakeholders
  monitor activities of genetic/expert centres (consultations)
  exchange data at European level (future) and facilitate clinical trials
  ➔ WIV-ISP is financed to gradually implement the Central Registry
  current situation: proof of concept phase finished
  genetic centres collecting patient data
  inclusion of hemophilia centres planned for 2016

A17: Orphanet Belgium
  improve visibility of Belgian specialised services and research activities at European level
  provide information on rare diseases in Dutch and French for the general public
  ➔ WIV-ISP is financed to maintain the Orphanet database
  to translate and validate Orphanet abstracts and medical terms
  Current situation: 40,000 terms, 880 scientific abstracts and framework of the Orphanet portal translated to Dutch

Contact persons: elfriede.swinnen@wiv-isp.be; annelies.mallezie@wiv-isp.be
Domain 3 Knowledge management

Domain 3: “Knowledge management”

A19: Codification and terminology

- improve visibility of rare disease patients in health information systems
- enable studies based on ORPHA codes

Example: for rare anemias there are
- 48 ICD9 codes,
- 49 ICD10 codes
- but 180 ORPHA codes

⇒ mapping of the detailed ORPHA rare disease codes to ICD9 will enable studies on administrative hospital data (currently coded in ICD9)

Future:
⇒ Pilot projects using Snomed-CT are running
- Translation of terms ongoing
- Use by nurses (procedures) in primary hospital systems
⇒ Future: also for secondary use such as automatic extraction of data of rare disease patients for the Central Registry

Contact person: Paz.urbina@wiv-isp.be
6th European Symposium on Rare Anaemias - 1st Dutch-Belgian meeting for patients and health professionals
Identificatie van patiënten met zeldzame ziekten in de belgische gezondheidssystemen

• Niet mogelijk met de huidige registratiesystemen
• Vb: Minimale ziekenhuisgegevens (MZG) van elke opname moet doorgestuurd worden naar FOD. De diagnose wordt gecodeerd in ICD-10-CM/PCM (ICD-9-CM/PCS tot 31/12/2014) → groot verlies aan granulariteit/detail want algemenere codes en meer gegroepeerd
• Vb voor zeldzame anemieën:
  - ORPHA  180 ziekten/codes
  - ICD-10  49 codes
  - ICD-9   48 codes
Waarom SNOMED CT gebruiken

Verbetering van de communicatie tussen

• zorgverstrekkers

• zorgverstrekker en patiënt

• Voor secundair gebruik = registraties voor financiering, wetenschappelijk onderzoek, kwaliteitscontrole, enz.

• Administratief
SNOMED CT terminologie

• Beheerd door IHTSDO, gebaseerd op “concepten” met definities, relaties, hiërarchieën en unieke codes

• Mapping SNOMED CT ↔ ORPHA-classificatie = samenwerking IHTSDO en INSERM - ongoing

• BE: Vertaling en validatie naar Frans en Nederlands - ongoing

• BE: POC fase voor algemene heelkunde, vasculaire heelkunde, gastro-enterologie en procedures (gedeeltelijk) – ongoing

• BE: vertaling en validatie van de ORPHA termen naar het Nederlands door WIV-ISP
POC voor secundair gebruik

Samenwerking CRRD-WIV en FOD Volksgezondheid:

• Integratie van CRRD variabelen in EHR

• Mapping van ORPHA codes en SNOMED CT codes (INSERM-IHTSDO)

• Mapping van resultaatcodes naar SNOMED CT en validatie door experten

• Automatische extractie van gegevens uit EHR voor CRRD gebaseerd op SNOMED CT codes voor zeldzame ziektes en vergelijking met data in CRRD ter validatie van de methode
POC voor secundair gebruik

• Doel:
• Automatische extractie van de gegevens voor CRRD uit het HER.
• In verre toekomst: terminologieserver met geïntegreerd alarm indien de diagnose mogelijks een zeldzame ziekte betreft
• Vb: Alpha-thalassemia
## ORPHA classificatie

Alpha-thalassemia and related diseases

<table>
<thead>
<tr>
<th>Code</th>
<th>Description</th>
</tr>
</thead>
<tbody>
<tr>
<td>36467003-68913001</td>
<td>Alpha-thalassemia</td>
</tr>
<tr>
<td>48553001</td>
<td>Hemoglobin H disease</td>
</tr>
<tr>
<td></td>
<td>Hb Bart's hydrops fetalis</td>
</tr>
<tr>
<td>277918006</td>
<td>Alpha-thalassemia - X-linked intellectual deficit syndrome</td>
</tr>
<tr>
<td>277918006</td>
<td>Alpha-thalassemia - intellectual deficit syndrome linked to chromosome 16</td>
</tr>
<tr>
<td></td>
<td>Alpha-thalassemia - myelodysplastic syndrome</td>
</tr>
</tbody>
</table>

## SNOMED CT terminologie

<table>
<thead>
<tr>
<th>Code</th>
<th>Description</th>
</tr>
</thead>
<tbody>
<tr>
<td>68913001</td>
<td>Alpha-thalassemia</td>
</tr>
<tr>
<td>234385007</td>
<td>Alpha thalassemia-2 trait (disorder)</td>
</tr>
<tr>
<td>191187006</td>
<td>Alpha trait thalassemia (disorder)</td>
</tr>
<tr>
<td>36467003</td>
<td>alpha^+^ Thalassemia (disorder)</td>
</tr>
<tr>
<td>86242003</td>
<td>alpha^+^ Thalassemia, deletion type (disorder)</td>
</tr>
<tr>
<td>85422000</td>
<td>alpha^+^ Thalassemia, nondeletion type (disorder)</td>
</tr>
<tr>
<td>66055002</td>
<td>alpha^0^ Thalassemia (disorder)</td>
</tr>
<tr>
<td>5300004</td>
<td>Hemoglobin Bart's hydrops syndrome (disorder)</td>
</tr>
<tr>
<td>234386008</td>
<td>Hemoglobin Constant Spring trait (disorder)</td>
</tr>
<tr>
<td>447117006</td>
<td>Hemoglobin H constant spring thalassemia (disorder)</td>
</tr>
<tr>
<td>48553001</td>
<td>Hemoglobin H disease (disorder)</td>
</tr>
<tr>
<td>234383000</td>
<td>Homozygous alpha thalassemia (disorder)</td>
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<tr>
<td>127045008</td>
<td>Sickle cell anemia with coexistent alpha-thalassemia (disorder)</td>
</tr>
<tr>
<td>127046009</td>
<td>Sickle cell trait with coexistent alpha-thalassemia (disorder)</td>
</tr>
<tr>
<td>277918006</td>
<td>Alpha thalassemia-mental retardation syndrome (disorder)</td>
</tr>
<tr>
<td>ICD-10</td>
<td>ICD-9</td>
</tr>
<tr>
<td>---------------</td>
<td>----------------------------</td>
</tr>
<tr>
<td>D56.0</td>
<td>282.43</td>
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<tr>
<td><strong>Synonyms</strong></td>
<td><strong>Alpha thalasemia</strong></td>
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<tr>
<td>Alpha plus thalasemia, nondeletion type</td>
<td>Alpha plus thalasemia, nondeletion type</td>
</tr>
<tr>
<td>Alpha plus thalasemia, single gene deletion</td>
<td>Alpha plus thalasemia, single gene deletion</td>
</tr>
<tr>
<td>alpha Thalasemia</td>
<td>alpha Thalasemia</td>
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<td>alpha^+^ Thalasemia, deletion type</td>
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</tr>
<tr>
<td>alpha^+^ Thalasemia, nondeletion type</td>
<td>alpha^+^ Thalasemia, nondeletion type</td>
</tr>
<tr>
<td>Homozygous alpha thalasemia</td>
<td>Hemoglobin H disease</td>
</tr>
<tr>
<td>Thalasemia, alpha</td>
<td>Hgb H disease</td>
</tr>
<tr>
<td>Thalasemia, alpha plus, deletion type</td>
<td>Thalasemia, alpha plus, deletion type</td>
</tr>
<tr>
<td>Thalasemia, alpha plus, nondeletion type</td>
<td>Thalasemia, alpha plus, nondeletion type</td>
</tr>
<tr>
<td>Thalasemia, alpha, homozygous</td>
<td>Thalasemia, alpha, homozygous</td>
</tr>
<tr>
<td><strong>Includes</strong></td>
<td><strong>Includes</strong></td>
</tr>
<tr>
<td>Alpha thalasemia major</td>
<td>Alpha thalasemia major</td>
</tr>
<tr>
<td>Hemoglobin H Constant Spring</td>
<td>Hemoglobin H Constant Spring</td>
</tr>
<tr>
<td>Hemoglobin H disease</td>
<td>Hemoglobin H disease</td>
</tr>
<tr>
<td>Hydrops fetalis due to alpha thalasemia</td>
<td>Hydrops fetalis due to alpha thalasemia</td>
</tr>
<tr>
<td>Severe alpha thalasemia</td>
<td>Severe alpha thalasemia</td>
</tr>
<tr>
<td>Triple gene defect alpha thalasemia</td>
<td>Triple gene defect alpha thalasemia</td>
</tr>
<tr>
<td><strong>Use additional code, if applicable, for hydrops fetalis due to alpha thalasemia</strong> (P56.99)</td>
<td><strong>Hydrops fetalis due to isoimmunization</strong> (P56.0)</td>
</tr>
<tr>
<td><strong>Excludes:</strong></td>
<td><strong>Excludes:</strong></td>
</tr>
<tr>
<td>alpha thalasemia trait or minor (D56.3)</td>
<td>alpha thalasemia trait or minor (282.46)</td>
</tr>
<tr>
<td>asymptomatic alpha thalasemia (D56.3)</td>
<td>hydrops fetalis due to isoimmunization (773.3)</td>
</tr>
<tr>
<td>hydrops fetalis due to isoimmunization (P56.0)</td>
<td>hydrops fetalis not due to immune hemolysis (778.0)</td>
</tr>
</tbody>
</table>
Contacts:

Feasibility study non-DNA tests: reimbursement, reference laboratories
- Nathalie Vandeveld

Feasibility study Quality genetic laboratories
- JeanBernard Beaudry

- Feasibility study Quality clinical activities genetic centres

Database on tests/samples: portfolio of Belgian genetic testing offer
- Kim Van Roey

Central Registry Rare Diseases
- Annelies Mallezie

Orphanet
- Elfriede Swinnen

Rare Disease Coding
- Montse Urbina Paz
Extra slides on specific actions
Action 1 – focus on non-DNA analyses for rare diseases

**Problem:** Non-reimbursed (expensive) analyses in Belgium or outsourced crossborder
No reference laboratories

**Main Objective:** To improve the diagnosis and follow-up of rare diseases

**Current status:** Feasibility study comprising of:

- A complete list of non-DNA analyses of clinical biology prescribed by Belgian clinicians for the diagnosis and/or follow-up of rare diseases
- A “Short-list” of non-DNA analyses eligible for reimbursement based on their clinical relevance and analytical efficiency.
- Proposal Selection criteria and procedure for future Reference Laboratories of Clinical Biology, recognized to perform the selected non-DNA analyses
- Financing proposition for selected non-DNA analyses and future Reference Laboratories

(*) : identification of gene mutations, measurement of the gene expression,... are excluded
Action 1 – focus on non-DNA analyses for rare diseases

Example: Priority for reimbursed according to national expert opinion

Coagulation and hemostasis

- Biochemie
- Hematologie
- Coagulatie & hemostase
- Immuno-hematologie & niet-infectieuze serologie
- Endocrinologie
- Therapeutische monitoring

>> nathalie.vandevelde@wiv-isp.be
**Action1 - Illustration in the context of anemia's:**

**Comparison of the clinical relevance & analytical efficiency of different analyses:**

<table>
<thead>
<tr>
<th>Diseases</th>
<th>Not reimbursed analyses</th>
<th>Reimbursed comparator</th>
</tr>
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<tbody>
<tr>
<td>Membrane disorders (e.g. sferocytosis)</td>
<td>• eosin-5’maleimide (EMA) test</td>
<td>• flow cytometry osmotic fragility</td>
</tr>
<tr>
<td></td>
<td></td>
<td>• cryohemolysis test</td>
</tr>
<tr>
<td>Paroxysmal nocturnal hemoglobinuria</td>
<td>• Transferrin isoforms analysis with Isoelectric focusing</td>
<td>• Transferrin isoforms analysis with capillary zone electrophoresis</td>
</tr>
<tr>
<td></td>
<td>• Apolipoprotein C3 phenotyping with Isoelectric focusing</td>
<td>• /</td>
</tr>
</tbody>
</table>

**Selection for a reimbursement?**

**Added value?**
### Comparison of the clinical relevance & analytical efficiency of different analyses:

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<td></td>
<td>• Apolipoprotein C3 phenotyping with Isoelectric focusing</td>
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### Reimbursed analyses with different methods:

<table>
<thead>
<tr>
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<th>Reimbursed analyses with different methods</th>
</tr>
</thead>
<tbody>
<tr>
<td>Sickle cell disease, Thalassemia’s and other forms of hemoglobinopathies</td>
<td>Hemoglobin analysis and phenotyping :</td>
</tr>
<tr>
<td></td>
<td>- Isoelectric focusing</td>
</tr>
<tr>
<td></td>
<td>- Electrophoresis</td>
</tr>
<tr>
<td></td>
<td>- Chromatography</td>
</tr>
<tr>
<td></td>
<td>- Spectrophotometric dosage</td>
</tr>
</tbody>
</table>
Illustration with some analyses performed in the context of anemia's:

**Example 1: Spherocytosis (auto-hemolytic anemia)**

- Several non-genetic analyses to distinguish different Hemoglobinopathies:
  - eosin-5'maleimide (EMA) test
  - flow cytometry osmotic fragility (FC OF)
  - cryohemolysis test
  - Hemoglobin/Mean corpuscular hemoglobin concentration ratio
- Different analyses with different methodologies
- Already compared in the scientific literature & laboratory practice

What's the more efficient analysis and best candidate for a reimbursement?
→ Survey
→ Discussion about the results
→ Field needs?

<table>
<thead>
<tr>
<th>Survey results</th>
<th>EMA test</th>
<th>FC OF test</th>
<th>Cryohemolysis test</th>
</tr>
</thead>
<tbody>
<tr>
<td>Reimbursement ?</td>
<td>NO</td>
<td>YES</td>
<td>YES</td>
</tr>
<tr>
<td>Total annual volume of prescription in Belgium ?</td>
<td>350</td>
<td>?</td>
<td>358</td>
</tr>
<tr>
<td>Turnaround time ?</td>
<td>3.5 ± 0.7 days</td>
<td>?</td>
<td>3.5 ± 0.7 days</td>
</tr>
<tr>
<td>Priority level for getting a reimbursement/increasing the reimbursement (between 1 [low priority] to 5 [high priority]) ?</td>
<td>5 ± 0</td>
<td>?</td>
<td>4 ± 1</td>
</tr>
</tbody>
</table>

A) Feasibility study: Improve Quality assurance of their laboratory activities

- Analysis current situation
  - All Genetic centres accredited (ISO-15189)
  - Particularities of genetic testing offer:
    - Diversity, rarity
    - EQA offer not available for many parameters
    - Statistical comparison not feasible
  > Need for more development of method-based EQA
  > Need for centralization of EQA schemes through WIV-ISP

- Costs estimation: Impact of Quality Systems on spendings
  - Costs of audits, EQA schemes
  - Participation in committees (guidelines etc)
  - Training, education of personnel

> Technical development causes challenges, new needs
  - Increased data storage capacities for large datasets (NGS)
  - Development of centralized databases for genetic variants
  - Uniform procedures for (long-term) storage of biological material and information
  - Need for clear guidelines (informed consent, diagnostic routing, incidental findings...)

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Action 2: Focus on Genetic centres – Evaluation of existing Quality Systems

• WIV-ISP in cooperation with the 8 genetic centres:
  • Improvement quality of care by the use of indicators
  • Centralized quality system:
    • Quality manual
    • System internal quality evaluation
    • Quality coordinator
    • Set up of internal and external quality controls
  • Estimation investments to be done by Centres ~EU norms
A) Quality assurance of their clinical activities

- Analysis existing situation, examples abroad
  - All
Quality genetic counselling:

**1) Additional Protocol on genetic testing to the Biomedicine Convention, EU level**

- Contains provisions to guarantee the quality of genetic centres in Member States of the Council of Europe
- Concentrates on protection of quality and safety of the services, respect for informed consent and protection of confidentiality

While respecting the wide variety of different systems, the EU wants its Member States to define clear quality and safety standards for health care provided on their territory

- Regulation quality genetics still national level

**2) EuroGentest:**
- Recommendations for genetic counselling

**3) ESHG (European Society of Human Genetics):**
- Guidelines and recommendations

Quality system in the centers of Human Genetics

6th European Symposium on Rare Anaemias - 1st Dutch-Belgian meeting for patients and health professionals
Proposition quality system

to be decided by expert panel by use of RAND/UCLA method

**Quality system** – 3 parts:

1. **Quality indicators**
   - registration – central evaluation
   - transparency
   - feedback centres → improvement, detection problems

2. **Audits**
   - Intern
   - Extern (peer review) – each 5 years

3. **Guidelines**
   - national guidelines
   + Quality coordinator per centre
Proposition quality system – how to proceed

RAND/UCLA methodology (= modified Delphi method)\(^6\,7\,8\)
= robust and valid method for quality indicator development.

5 steps:
1. **Systematic literature review** -> to identify a preliminary set of quality indicators
2. **Recruitment of experts** reflecting the variety of stakeholders in the area of interest
3. **1st round homework assignment**: the experts score the preliminary set of indicators for ‘appropriateness’ on a scale from 1 (not appropriate) to 9 (very appropriate)
4. **2nd round of scoring in a face-to-face panel meeting**. Only indicators with a median score of 4, 5, 6 or higher but not reaching consensus need to be discussed, modified where necessary and rescored. The indicators with a median score of 7, 8 or 9 are automatically added to the quality indicator set.
5. Using the second round scores to **decide on the quality indicator set**
6. References

4. EuroGentest. Summary of guidelines for genetic counselling
   • http://www.eurogentest.org/fileadmin/templates/eugt/pdf/summaryofguidelinesMay06.pdf
5. ESHG European Society of Human Genetics. https://www.eshg.org/eshgdocs.0.html
Proposition quality system clinical activities

to be decided by expert panel by use of RAND/UCLA method

**Quality system** – 3 parts:

1. **Quality indicators**
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4. **Quality coordinator per centre**