EQAS for Rare and Congenital Anaemias

Barbara De la Salle

UK NEQAS General Haematology
West Herts Hospitals NHS Trust
Objectives

• Haematology WG proposal
• ENERCA
• Questionnaire Results
• New proposal – PK EQAS
• Problems of providing cross boundary EQAS
• Experience – UK NEQAS Molecular Haemoglobinopathies
Haematology WG Proposal

European survey of existing EQAS for rare diseases diagnostic tests

TITLE
The provision of External Quality Assessment Schemes (EQAS) for the diagnostic tests associated with rare anaemias in Europe

AIMS AND OBJECTIVES
1. To survey EQA providers within European Union member states to determine the provision of EQAS for rare and congenital anaemias.
2. To discuss with EQA providers the use of reference methods for haematological parameters within EQAS and how IVDD companies use reference methods in the calibration and development of instruments and kits.
3. To work in collaboration with the European Network for Rare and Congenital Anaemias (ENERCA) to produce a collated catalogue of relevant EQAS.
4. To present the outcome of the survey at the EQALM annual meeting, 2011.
European Network for Rare and Congenital Anaemias

Rare anaemias (RA) :
Prevalence less than 5 per 10,000 individuals in a given community.
ENERCA 1  2002
ENERCA 2  2005
ENERCA 3  2009
WP1 –
WP2 – Quality of patient care
WP3 –
WP4 –
WP5 –
WP6 –
WP2 Specific Objectives 1

• To establish close collaborative links with recognised European and International organisations

• To improve the quality of laboratory data on RA by linking methods to higher order reference materials
WP2 Specific Objectives 2

- To facilitate the participation of Expert Centres in EQA
- To provide educational EQAS
- To prioritise the preparation of guidelines for the laboratory diagnosis of RA
- To develop a European Registry of RA
QUESTIONNAIRE TO EUROPEAN EQA PROVIDERS
ENQUE-Harmonisation-2

Dear Colleague,

At last year’s EQALM meeting, the Haematology Working Group adopted a new project to examine the provision of EQAS and the use of reference methods for the laboratory investigations used in the diagnosis of Rare Anaemias throughout Europe. This work is a collaboration with the European Network for Rare and Congenital Anaemias (ENERCA) with the objective of providing improved patient diagnostics through standardisation and harmonisation.

Sent June 2011
Core List of Tests

- Rare anaemia groups – ENERCA website
- Rationalised into disorders using same types of diagnostic testing
- Background research
  - Literature search for tests used
  - Abstracts
<table>
<thead>
<tr>
<th>General Laboratory Tests</th>
<th>Red cell enzyme disorders</th>
<th>RBC membrane disorders</th>
<th>PNH</th>
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<td>Bilirubin</td>
<td>Liver function test</td>
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<td>Haemoglobin</td>
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<td>LDH levels</td>
<td>Dilute electrophoresis</td>
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<tr>
<td>Serum LDH</td>
<td>Hb S screening tests</td>
<td>Molecular haplotype</td>
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<td>Urea urea nitrogen</td>
<td>Hb A2, Hb F, Hb S-quantitation</td>
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<td>Haemoglobinuria</td>
<td>HPLC</td>
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<tr>
<td>Urine albuminuria</td>
<td>Electrophoresis and elution</td>
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<tr>
<td>Urine acetone</td>
<td>Heat and test</td>
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<tr>
<td>Serum folate</td>
<td>Hb H bodies</td>
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<td>Red cell folate</td>
<td>Heinz bodies</td>
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<tr>
<td>Cobaltin</td>
<td>Whole blood elution</td>
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<tr>
<td>Neutrophils, lymphocytes</td>
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<td>Platelet count</td>
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</tbody>
</table>

**General Laboratory Tests**

**Blood film morphology**

- Leukocyte count
- Platelet count
- Neutrophils
- Lymphocytes
- Monocytes
- Eosinophils
- Basophils
- Megakaryocytes

**Red cell enzyme disorders**

- Glucose-6-phosphate dehydrogenase (G6PD) deficiency
- Pyruvate kinase deficiency (PKD)
- Pyrimidine-5'-nucleotidase deficiency (PNK)

**RBC membrane disorders**

- Demonstration of red cell membrane proteins by sodium dodecyl sulfate polyacrylamide gel electrophoresis (SDS-PAGE)
- Chronic hemolytic anemia (Hb, He)

**PNH**

- Fluo cytochemistry for CD55 and CD59

**Please list any additional tests here - Applicable to rare and congenital anaemia diagnosis**

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</tbody>
</table>

**UK NEQAS**
## General Laboratory Tests

<table>
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<tr>
<td>Blood film morphology</td>
<td>Hb variant detection</td>
<td>Glucose-6-phosphate dehydrogenase (G6PD) deficiency</td>
<td>Demonstration of red cell membrane proteins by sodium dodecyl sulfate polyacrylamide gel electrophoresis (SDS-PAGE)</td>
<td>Ham test (modified serum test)</td>
</tr>
<tr>
<td>CBC (complete blood count)</td>
<td>HPLC</td>
<td>Met Hb test</td>
<td>Chromatography of erythrocyte membrane proteins (ZEBC)</td>
<td>Sucrose lysis test (sugar-water test)</td>
</tr>
<tr>
<td>Sialic acid content</td>
<td>Hb electrophoresis</td>
<td>Fluorescent spot test</td>
<td>Proportion of spectrin dimers and tetramers in red cell membranes (Hb)</td>
<td>Flow cytometry for CD38 and CD45</td>
</tr>
<tr>
<td>Uric acid content</td>
<td>Isoelectric focusing</td>
<td>Cytochemical demonstration of spectrin deficiency (ZEBC)</td>
<td>Trypsin digestion of spectrin (Hb)</td>
<td>FLAER assay (fluorescently labeled spectrin test)</td>
</tr>
<tr>
<td>Serum haptoglobin</td>
<td>Capillary electrophoresis</td>
<td>Quantitative assay for G6PD activity</td>
<td>Chromatographic assay for G6PD (especially Hb)</td>
<td></td>
</tr>
<tr>
<td>LDH levels</td>
<td>Globulin electrophoresis (GE)</td>
<td>Chromatography of spectrin (Hb)</td>
<td>Autohemolysis test (Hb)</td>
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<tr>
<td>Serum LDH</td>
<td>Hb S screening tests</td>
<td>Molecular diagnostic</td>
<td>Acidified glycerol lysis test (AGLT) (Hb)</td>
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<tr>
<td>Urea nitrogen</td>
<td>Whole blood sedimentation test, soluble activity test</td>
<td>Molecular diagnostic</td>
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<td>Haemoglobinuria</td>
<td>Hb A2, Hb F, Hb S quantitation</td>
<td>Pyruvate Kinase deficiency spot test</td>
<td>Pyruvate Kinase deficiency (PKD)</td>
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<tr>
<td>Urea concentrations</td>
<td>Electrophoresis and elution</td>
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<tr>
<td>Urea tests</td>
<td>Unstable haemoglobin: heat stability test</td>
<td>Quantitative assay for PK activity</td>
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<tr>
<td>Urea folate</td>
<td>Hb H bodies</td>
<td>Molecular diagnostic</td>
<td></td>
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<tr>
<td>Red cell folate</td>
<td>Methemoglobin bodies</td>
<td>Pyridine-4-carboxylic acid deficiency (Pyridine-4-carboxylic acid deficiency)</td>
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<tr>
<td>Cobalt uptake</td>
<td>Kleihauer test (Hb detection)</td>
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<td>Serum ferritin</td>
<td>Flow cytometry for Hb-F cells</td>
<td>Other red cell glycolytic enzyme deficiencies</td>
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</table>

## Hb disorders

### Hb variant detection

<table>
<thead>
<tr>
<th>Hb variant detection</th>
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</thead>
<tbody>
<tr>
<td>Immunophenotypic assay</td>
<td>Glutathione stability</td>
</tr>
<tr>
<td>Serum iron and total iron binding capacity (TIBC)</td>
<td>Hb A1B Hb detection using absorption spectrophotometry (ZEBC)</td>
</tr>
<tr>
<td>Liver iron concentration (SQID or MRT); Moeskert iron concentration (MRT)</td>
<td>Electrospray ionization mass spectrometry (ESI-MS)</td>
</tr>
<tr>
<td>Serum ferritin (SF) &amp; transferrin saturation</td>
<td>Methemoglobinemia</td>
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<tr>
<td>Beta-thalassemia mutations</td>
<td>Glutamine synthetase deficiency (Remithand)</td>
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<tr>
<td>Structural variants</td>
<td>Quantitative assay for methionine synthase activity</td>
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<tr>
<td>Molecular diagnosis</td>
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<td>DNA analysis</td>
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</table>

## Additional Tests

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<tr>
<th>General Laboratory Tests</th>
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<th>PMH</th>
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## UK NEQAS
### General Laboratory Tests

<table>
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<tr>
<td>Blood film morphology</td>
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<td>Glucose-6-phosphate dehydrogenase (G6PD) deficiency</td>
<td>Demonstration of red cell membrane proteins by sodium dodecyl sulfate polychromatolytic gel electrophoresis (SDS-PAGE) (Hb Hb, hemoglobinopathies)</td>
</tr>
<tr>
<td>CBC (complete blood count)</td>
<td>HPLC</td>
<td>HPLC</td>
<td>HPLC</td>
</tr>
<tr>
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<td>HPLC</td>
<td>HPLC</td>
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<tr>
<td>Blood urea</td>
<td>HPLC</td>
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</tbody>
</table>

### Red cell enzyme disorders

**Glucose-6-phosphate dehydrogenase (G6PD) deficiency**
- Reduced glutathione (GSH) assay
- Electrophoresis
- Spectrophotometric assay

### Additional Tests

- DNA analysis
- Alpha thalassemia mutation
- Structural variants

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**UK NEQAS**

Please list any additional tests here - applicable to rare and congenital anaemia diagnosis
### General Laboratory Tests

<table>
<thead>
<tr>
<th>Test Description</th>
<th>Methodology</th>
</tr>
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<tbody>
<tr>
<td>Hemoglobin level</td>
<td>HPLC</td>
</tr>
<tr>
<td>Platelet count</td>
<td>Fluorometric</td>
</tr>
<tr>
<td>Erythrocyte count</td>
<td>Digital</td>
</tr>
</tbody>
</table>

### RBC membrane disorders

**Demonstration of red cell membrane disorders**

1. **Ham test** (modified serum test)
2. **Serum protein electrophoresis**
3. **Coombs test** (direct and indirect)
4. **Flow cytometry** for CD55 and CD65

**RBC enzyme disorders**

1. **Glucose-6-phosphate dehydrogenase deficiency**
2. **Pyruvate kinase deficiency**
3. **G6PD deficiency**

**PNH**

1. **Coombs test** (direct and indirect)
2. **Flow cytometry** for CD55 and CD65

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**Please list any additional tests here - applicable to rare and congenital anaemia diagnosis**

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<td>HPLC</td>
<td>NBT spot test</td>
<td>Glucosyltransferase test</td>
<td>Sucrose lysis test (sucrose-water test)</td>
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<td>Red blood cell count</td>
<td>Hb electrophoresis</td>
<td>Fluorescent spot test</td>
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<td>Flow cytometry for C5b and C9b</td>
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<td>10-electro focusing</td>
<td>Cytoskeletal demonstration of G6PD deficiency</td>
<td>Trypsin digestion of spectrin (Hb)</td>
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<td>Capillary electrophoresis</td>
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<td>FLAER assay (fluorescence-labeled erythrocyte) test</td>
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<td>LDL levels</td>
<td>Glutathione peroxidase</td>
<td>Chromatation inhibition test</td>
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<tr>
<td>Serum LDH</td>
<td>Hb A2, Hb F, Hb S electrophoresis</td>
<td>Molecular diagnosis</td>
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<td>Autohemolysis test (Hb)</td>
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<tr>
<td>Serum ferritin</td>
<td>Hb A2, Hb F, Hb S electrophoresis</td>
<td>- HPLC</td>
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<tr>
<td>Urate and other anions</td>
<td>Hb A2, Hb F, Hb S electrophoresis</td>
<td>- RFLP</td>
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<td>Uric acid</td>
<td>Hb A2, Hb F, Hb S electrophoresis</td>
<td>- -SSCP</td>
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<td>- RFLP</td>
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<td>Hb H bodies</td>
<td>- RFLP</td>
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<td>Zinc-protoporphyrin</td>
<td>Hb H bodies</td>
<td>- RFLP</td>
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<td>Bone marrow iron stain</td>
<td>Hb H bodies</td>
<td>- RFLP</td>
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</table>

**PNH**

- Haptoglobin deficiency
- Serologically positive attacks
- Autoimmune hemolytic anemia
- Platelet dysfunction

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**Please list any additional tests here - applicable to rare and congenital anemia diagnosis**

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**UK NEQAS**
1.1 What is the name of the EQA programme or survey provided?

Full Blood Count

1.2 What tests from the 'core list of tests' are covered?

CBC - Hb, WBC, RBC, Hct, MCV, MCH, MCHC, Platelets

1.3 How frequently do you distribute specimens?

12 times per year

1.4 How many specimens are distributed each year?

24

1.5 What is the type of survey material provided?

Human whole blood - partially fixed

1.6 How many participants are registered?

1,360

1.7 Do you accept participants from outside your own country?

Yes

1.8 Is there a higher order reference method for this test?

Yes

1.9 How do you establish your target value?

Higher order reference method

2.1 Off the tests that you do NOT provide EQAS for, list the 5 that you think would benefit most from EQAS provision.

- TSH
- Free thyroxine
- Free triiodothyronine
- Thyroid stimulating hormone
- T4

2.2 Would you be prepared to offer EQAS for rare anaemias in collaboration with another EQAS provider?

Yes

2.3 Are there any other tests that should be included in the core tests list?

No

2.4 About your EQA scheme

Organization name

UK NEQAS General Haematology

Nature of the organisation

Private company

2.5 Name of the Scheme Organiser/Director

Professor Keith Hyde

2.6 Address

PO Box 14, Watford, WD18 9FJ, UK

Telephone number

44 1923 217676

Fax number

44 1923 217676

2.7 Email

neqas@ukneqas.org.uk

2.8 Website

www.ukneqas.org.uk

2.9 Name of the person completing this form

Barbara De la Faille

2.10 Would you agree to your scheme being listed on the European Network for Rare and Congenital Anaemias website (www.enerca.org)?

Yes
Questionnaire

- Analytes
- Frequency
- Type of survey material
- Number of participants
- Performance monitoring
- EQAS ‘wishlist’
- Potential for collaboration
- Accreditation status
Responses by September 2011

Response rate

<table>
<thead>
<tr>
<th>Number of organisations</th>
<th>Responded</th>
<th>Asked</th>
</tr>
</thead>
</table>

Countries (some sent 2 replies)

- Slovenia
- Denmark
- Romania
- France
- Spain
- Canada
- Sweden
- UK
- Switzerland
- Norway
- Ireland
- Russia
- Czech Republic
- Croatia

50% (16/31)
EQAS Available for General Laboratory Tests

- Vitamin B₁₂
- Bilirubin
- CBC
- Serum Fe
- Folate
- Ferritin
- Haptoglobins
- Urinary haemosiderin
- Bone marrow Fe
- LDL
- LDH
- Blood morphology
- Post analytical interpretation
- Reticulocytes
- Transferrin
- TIBC
What’s not provided from the core list?

- Urine ferrioxamine iron
- Serum Transferrin Receptor
- Liver iron
- Myocardial iron
- Zinc protoporphyrin
But .............

Are all tests clinically useful?
EQAS Available for Hb Disorders

- Sickle cell solubility
- Hb variant identification
- Hb A2 %
- Hb F %
- Hb S %
- Hb H bodies
- Newborn sickle screening
- Molecular Haemoglobinopathies (DNA)
Only 3 provide services outside their own country
EQAS FOR MOST TESTS

...........Except: Hb F by flow, Kleihauer, Methaemoglobin, Haemoglobinuria, PNH by flow
EQAS Providers - Other tests

- Hb F by flow
- Kleihauer
- G6PD
- Methb
- Hburia
- PNH by flow
Do you accept participants from outside your own country?

** ALSO - 11 out of 15 EQAS providers would be prepared to offer new specialist services in collaboration**
### Frequency of Provision

<table>
<thead>
<tr>
<th>Test</th>
<th>Range</th>
<th>Specimens/yr</th>
</tr>
</thead>
<tbody>
<tr>
<td>CBC</td>
<td>2 - 24</td>
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<tr>
<td>Bilirubin</td>
<td>1 - 26</td>
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</tr>
<tr>
<td>Hb A_2</td>
<td>1 - 18</td>
<td></td>
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</tbody>
</table>
EQA Wishlist

Available within EQALM

• Hb Variant detection
• Hb A2, Hb F, Hb S
• G6PD
• Kleihauer
• Flow cytometry for Hb F
• Retics
• Red cell folate
• Serum folate
• Cobalamin
• Serum ferritin
• Serum Haptoglobin
• Blood Film Morphology

Not available within EQALM

• Unstable Hbs
• Heinz bodies
• Serum transferrin receptor
• PK activity

QUESTION
Of the tests that you do NOT provide EQAS for, list the 5 that you think would benefit most from EQAS provision?
Barriers to EQAS provision

• Survey material
  – Availability
  – Stability
• Insufficient demand in a single country
• Restriction of services to own country only
  – Funding restrictions
• Cost of transportation
• Customs difficulties
• Local medical practice
• Language
New PK scheme proposal

• European collaboration
• Normal and PK deficient patient material

Development phases:
  – Survey material development
    • Storage, stability, volumes etc.
  – Small scale survey with selected labs
  – Recruitment of interested participants
  – Pilot exercise(s) to refine scheme design
  – Performance assessment methods
How will we provide pan-European services?

• Direct sale and delivery to individual participants
• Provision via an intermediary agent or distributor
• In collaboration with another EQA provider
  – Supported by ¾ responders to questionnaire
UK NEQAS Molecular Haemoglobinopathies Scheme

- Approximately 9 labs in the UK
- Boosted to 25 with European labs
- Survey material – development of cell line library of cases

Problems
- Cost
- Survey material
- Matching EQA cases to local incidence
- Performance assessment
Summary

• **For rare anaemias**
  – Adequate provision of most general tests within EQALM
  – Some provision for specialist / rare anaemias:
    • Haemoglobinopathy EQAS
    • PNH EQAS
    • G6PD EQAS

• **EQALM next steps**
  – Facilitate participation and collaboration
  – Offer guidance on new provision
  – Help recruit specialist laboratories to ENERCA
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Any more responses?

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