EQA scheme coordination centre
KU Leuven, Department of Public Health and Primary Care
Biomedical Quality Assurance Research Unit
Herestraat 49, box 602, 3000 Leuven, Belgium
Tel. +32 (0)16 33 01 43 - Fax: +32 (0)16 34 71 90
E-mail: cf.network@med.kuleuven.be

GENERAL REPORT

CYSTIC FIBROSIS
EXTERNAL QUALITY ASSESSMENT SCHEME 2012

Scheme period: June 2012 – April 2013

Assessment meeting: April 18 – 19, 2013, Madrid

Date of report: July 25, 2013

Report version: 2

Changes compared to version 1: p4, average genotyping score

Drafted by: Prof. Dr. Els Dequeker, Dr. Marie des Georges, Dr. Emmanuelle Girodon, Dr. Christof Meyer-Kleine, Dr. Michael A. Morris, Dr. Martin Schwarz, Lien Tembuyser, Dr. Raina Yamamoto and Sofie Delen

Authorized by: Prof. Dr. Els Dequeker and Dr. Emmanuelle Girodon

Announcement:

This year the assessment meeting of the CF EQA scheme was held together with the assessment meeting of the European Molecular Genetics Quality Network (EMQN). Since 2012 we are working closely together to use the benefits in harmonization and efficiency for both organizations. The re-developing of the EQA databases and websites are essential before we can offer the same tool for registration, data submission and evaluation. This cooperation includes that we are also changing our timing of the scheme. More information on the timetable changes will be communicated with the participants when available.

Note that the general report comes with the resources listed below:

- **Assessment table 1 – genotype and interpretation results, and clerical/reporting accuracy: ***
  Genotypes were evaluated together with interpretation and clerical/reporting accuracy. The results of this assessment can be found in assessment table 1.
- **Individual comments: ***
  Laboratories that submitted reports containing errors or elements that should or could be improved, received individual comments from the assessors.
- **Certificate of participation:** **
  All laboratories received a certificate of participation. It is indicated on the certificate whether or not the laboratory participated successfully. Laboratories that submitted written reports, including correct genotype data and no serious interpretation or risk calculation errors, were deemed to be successful. The genotype, interpretation and clinical/reporting scores are printed on the certificate.
- **Letter of persistent poor performance: ****
  Laboratories that did not successfully participate in the current year and at least once in the two previous years, received an additional letter inviting them to contact the assessors to discuss their results and ways to improve their performance.

Please check if the data in the compiled resources are in agreement with the data that were sent out of the laboratory. If you feel any mistakes were made in the assessment, please contact cf.network@med.kuleuven.be **before June 24, 2013**. The CF Network will respond after the 15th of July. Thereafter the marks become final.

* Resources available at http://cf.eqascheme.org, after logging in

** Resources sent to the laboratory by mail

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CONTENT

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OVERALL RESULTS

Participants:

- In total, 218 participants received a set of 3 purified DNA samples (15µl, concentration ~ 0.33µg/µl). Of these, **participants from 215 laboratories in 34 countries returned their results**.

Country	Number of laboratories that participated in the scheme in			Country	Number of laboratories that participated in the scheme in		
	2010	2011	2012		2010	2011	2012
Australia	10	11	11	Netherlands, The	8	8	8
Austria	6	5	5	New Zealand	1	1	1
Belgium	7	7	8	Norway	0	1	1
Croatia	2	2	2	Poland	7	7	5
Cyprus	1	1	1	Portugal	2	2	2
Czech Republic	3	4	4	Russia	1	0	0
Denmark	2	2	2	Saudi-Arabia	0	0	0
Estonia	2	2	2	Serbia	2	2	1
Finland	1	1	1	Slovakia	1	0	1
France	30	31	32	Slovenia	1	1	1
Germany	51	47	50	Spain	18	15	13
Greece	5	6	5	State of Qatar	0	0	0
Hungary	2	3	3	Sweden	1	4	3
Ireland	1	1	1	Switzerland	12	12	14
Israel	0	1	1	Turkey	2	1	2
Italy	25	26	25	Ukraine	1	1	1
Latvia	1	1	1	United Kingdom	7	6	4
Lithuania	1	1	1	United States	3	4	2
Macedonia	1	1	1	**TOTAL**	**218**	**218**	**215**

Assays used:

- **82.3% of laboratories (n=177) used a commercial assay** to test for the presence of *CFTR* mutations in CF EQA sample 1 (CF12-1).

		Number	
Assay	**Company**	**2011**	**2012**
RDB-INNO LiPA CFTR17+Tn Update and CFTR19	Innogenetics	50	51
RDB-INNO LiPA CFTR19	Innogenetics	7	7
RDB-INNO LiPA CFTR17	Innogenetics	7	5
RDB-INNO LiPA CFTR17+Tn Update and CFTR19 + CFTR Italian Regional	Innogenetics	5	4
RDB-INNO LiPA CFTR17+Tn Update and CFTR19 + INNO LiPA CFTR Deletions + 6	Innogenetics	1	0
RDB-INNO LiPA CFTR Deletions + 6	Innogenetics	1	0
Cystic Fibrosis Genotyping Assay	Abbott Molecular	65	58
Elucigene CF29v.2	Gen-Probe Life Sciences	14	9
Elucigene CF-EU2	Gen-Probe Life Sciences	9	23
Elucigene CF30	Gen-Probe Life Sciences	5	5
Elucigene CF4	Gen-Probe Life Sciences	1	0
Elucigene CF-EU1	Gen-Probe Life Sciences	0	0
AC023/25 Cystic Fibrosis	Nuclear Laser Medicine s.r.l.	5	6
xTAG® Cystic Fibrosis 71 kit v2	Luminex Molecular Diagnostics	2	2
xTAG® Cystic Fibrosis 39 kit v2	Luminex Molecular Diagnostics	2	1
Reverse hybridization StripAssay	ViennaLab Diagnostics	2	3
MassARRAY MALDI-TOF	Sequenom	2	1
Devyser CFTR Core	Devyser	0	1

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Myriapod® Cystic Fibrosis	Diatech Pharmacogenetics	0	1
MALDI TOF Mass Spectrometry home-brew	/	1	0
SNP extension assay (SNuPe&MALDI-TOF)	/	1	1
DNA sequencing	/	18	17
Real time PCR	/	4	3
Restriction Enzyme Analysis (REA)	/	4	1
Heteroduplex Analysis (HA)	/	3	2
Amplification Refractory Mutation System (ARMS)	/	3	2
Arrayed Primer Extension (APEX)	/	2	2
Capillary Electrophoresis (CE)	/	1	3
Restriction Fragment Length Polymorphism (RFLP)	/	1	1
Allele Specific PCR (AS-PCR)	/	1	2
Allele Specific Oligonucleotide PCR (ASO-PCR)	/	0	1
Allele Specific Primer Extension (ASPE)	/	1	2
Single-Strand Conformation polymorphism (SSCP)	/	0	1
Unknown (no answer received)	/	0	0
TOTAL		**218**	**215**

Successful participation:

- **194 of 215 laboratories (90.2%) successfully participated** in the CF EQA scheme.
- **9 of 215 participants (4.2%) were identified as persistent poor performers**.

Nature of error	**2010 (218)**	**2011 (218)**	**2012**
Genotype error	4 (1.8%)	6 (2.8%)	12 (5.6%)
Genotype error + error in risk calculation	1 (0.5%)	0	0
Genotype error + no, wrong or insufficient interpretation	0	0	0
Genotype error + no reports received	0	1 (0.5%)	0
Genotype error + no, wrong or insufficient interpretation + error in risk calculation	1 (0.5%)	0	0
No, wrong or insufficient interpretation	7 (3.2%)	11 (5.0%)	4 (1.9%)
No, wrong or insufficient interpretation + error in risk calculation	5 (2.3%)	0	0
No, wrong or insufficient interpretation + no reports received	0	0	0
Serious error in risk calculation	30 (13.8%)	8 (3.7%)	2 (0.9%)
Datasheet submitted, but no reports received	4 (1.8%)	4 (1.8%)	2 (0.9%)
Analytical failure for all samples		1 (0.5%)	1 (0.5%)
TOTAL no successful participation	**52 (23.9%)**	**32 (14.7%)**	**21 (9.8%)**
Successful participation	**166 (76.1%)**	**186 (85.3%)**	**194 (90.2%)**

Genotyping:

- **12 of 215 laboratories (5.6%) made genotype errors**.

Genotype error	**2012**
False negative	9
Wrong genotype	3

- **The average genotyping score for the 2012 scheme is 1.960 / 2.000** (on a total of 645 samples).

Interpretation:

- **4 of 215 laboratories (1.9%) provided incorrect, insufficient or no interpretation.** Sufficient interpretation of the result should provide an answer to the questions asked in the referral, along with recommendations for genetic counselling and further testing, if relevant, as well as implications for family members (OECD guidelines for quality assurance in molecular genetics testing). Interpretation in genetics

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reports is important as they can have a very long life-time. Interpretation should be understandable by any recipient and not only, for example, by specialized geneticists (OECD guidelines).

- **The average interpretation score for the 2012 scheme is 1.812 / 2.000** (out of a total of 619 samples).
- In the event of a laboratory making a genotype error for one of the cases, the interpretation for that case was not marked; all fields in assessment table 1 will be black for that case. Consequently, the laboratory is not penalized for scoring of interpretation and the maximum score for interpretation is 4.00/4.00.
- **2 of 215 laboratories (0.9%) participated unsuccessfully because of an error in the risk calculation.**
- **Inappropriate/misleading reporting:** 71 of 215 laboratories provided inappropriate or erroneous comments that were not included in the expected interpretation elements and were penalized; for example: mentioning or offering PND in case 1 when only 1 mutation is detected. For this type of error, points are deducted from the interpretation score of the relevant case.

Clerical/reporting accuracy:

- **18 of 215 laboratories (8.4%) made clerical/reporting errors which could lead to a serious risk of mis-identification or misinterpretation.**
- **The average clerical/reporting accuracy score for the 2012 scheme is 1.971/ 2.000** (out of a total of 619 reports).
- Laboratories will receive, apart from the genotyping and interpretation score, a Clerical accuracy score. This score was calculated based on 3 different error types (columns in assessment table 1). This year, we had **1 laboratory of 215 (0.5%)** we evaluated as a poor performer for clerical accuracy.
    - Serious clerical error(s): wrong patient names, wrong date of birth, …
    - Mutation(s) sometimes written incorrectly: F508 (c.1521_1523delccT), 551D, …
    - Wrong nomenclature(s): c.1898+1G>A was written instead of 1898+1G>A, …
- **From 2013 onwards**: all errors regarding nomenclature will be included in the genotyping score.

Nomenclature:

- **It is recommended that reports include a description of identified sequence variants in both HGVS and traditional nomenclature.**
- HGVS nomenclature should be used correctly. This implies:
    - Describing sequence variations at the nucleotide level, the cDNA level.
      Example: F508del is equivalent to c.1521_1523del or c.1521_1523delCTT.
    - Specifying the nucleotide reference sequence on the report, including the version number. For *CFTR*, the reference sequences currently recommended are NM_000492.3 for cDNA and NG_016465.1 for gDNA.
    - Exon numbering. The numbering of the *CFTR* exons has a historical basis, with the result that, for example, “exon 10” is the 11th exon (www.genet.sickkids.on.ca). However, because of the precision of HGVS nomenclature (employing a nucleotide definition of any mutation), **reference to exon numbers is no longer appropriate, once a mutation is accurately reported**. Alternatively, the system used for exon numbering should be clearly indicated.
- Laboratories should be aware that HGVS recommendations are being regularly modified and expanded. Therefore, when using HGVS nomenclature, it is good practice to regularly consult the latest version on the HGVS website: http://www.hgvs.org/mutnomen/
- **Recent changes in the HGVS rules** (HGVS V2.0), which occurred after the publication “Berwouts et al., Hum Mutat. 2011 Nov;32(11):1197-203)” include:
    - Use of “*” instead of “X” for a stop codon. Therefore p.Gly542X becomes p.Gly542*.
    - Use of “;” instead of “+” in genotypes. Therefore c.[1521_1523del]+[=] becomes c.[1521_1523del];[=].

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- We recommend informing your clinical colleagues about the use of HGVS and major changes whenever necessary.

Other reporting elements:

- Unique identification: **name**, **gender, date of birth and patient identifier number**. The first (given) and last (family) name should be distinguished.
- **Ethnic / geographic origin,** which should be differentiated from place of birth.
- **Sample reception date,** which should be differentiated from **sample collection date**.
- Laboratory's **own sample identification number**.
- **Date of the report**.
- **Signatures** or a mention of who was responsible for the electronic signature.
- **Unique identifier on each page** if the report contains more than one page. This could be an ID or name of the patient.
- **Page number** and **total number of pages**: 1/1 or 1 of 2 and so on.
- **Name of** the **referring person**.
- **Nature of sample.** A number of laboratories identified it as blood, presumably because this is the laboratory's usual sample type. It is important to accurately record the precise sample type, especially when DNA has been extracted by a third party.
- **Reason for testing** re-stated in full and not abbreviated ('CF testing' is not sufficient).
- **Mutations tested and method of testing**.
    - Where a **commercially-available kit** is used, this should be clearly identified in the report (or traceable for the user), including the **version** of the kit, as modifications may have improved detection of certain mutations.
    - The **quoted list of mutations** should be checked and updated, according to the version of any kit being used. This information can be appended as a footnote to the report, rather than in the body of the text.
- **Detection rates (diagnostic sensitivity),** which should be taken from the scientific literature or from WHO reports (http://www.who.int/genomics/publications/en/) and which should reflect the patient's population when known. Laboratories are therefore recommended to indicate the mutation detection rate of the test used, which may vary according to the ethnic or geographic origin of the individual. Laboratories should not use their own (local) population figures for all patients, as these may not be applicable to other ethnic origins.
- The report **title** refers to *CFTR* gene study or to cystic fibrosis molecular diagnosis and can be clearly distinguished from the rest of the report. Mentioning EQA in the title is not appropriate.
- **The main message of the report should include the results and their interpretation, in the context of the indication for testing.** Additional technical information could be in tables or footnotes. Similarly, tables of risks should be avoided if they are not relevant to the particular patient.

Laboratories could consult the following resources in order to improve their reports:

- Best practice guidelines for molecular genetic diagnosis of cystic fibrosis and CFTR-related disorders-updated European recommendations – Dequeker *et al.*, *Eur J Hum Gen*, 2009, 17(1): 51-65.
- Best practice guidelines on reporting in molecular genetic diagnostic laboratories in Switzerland – Swiss Society of Medical Genetics (www.sgmg.ch).
- Recommendations for reporting results of diagnostic genetic testing (Biochemical, Cytogenetic and Molecular Genetic) - On behalf of the ESHG Quality committee, M Claustres et al., *Eur J Hum Gen*, accepted 2013.
- Example reports on the CF Network website: http://cf.egascheme.org/info/public/education.xhtml

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EXPECTED GENOTYPE AND INTERPRETATION PER CASE

Case 1:

- Danielle Wittmer is a neonate suspected of being affected with cystic fibrosis, based on positive sweat tests. Her maternal aunt died of cystic fibrosis 30 years ago. The search for frequent mutations was negative in Danielle's mother. A molecular analysis is requested for Danielle to confirm the diagnosis.
- **CF12-1:** Danielle Wittmer - Innsbruck, Austria, DOB 02/06/2012
- **Expected genotype result:**
  c. [827_828delinsAA(;)1521_1523del]
  or c.[827G>A(;)828C>A(;)1521_1523del]
  (F508del / C276X)
- **Expected interpretative criteria:**

In case two mutations are detected

- Confirmation of CF diagnosis (1.00)
  The confirmation of the diagnosis of cystic fibrosis should be clearly stated.
  Example text: *the patient is highly likely to be compound heterozygous for c.1521_1523delCTT (p.Phe508del, F508del) and c.827_828delinsAA (p.Cys276*, C276X), which would confirm the diagnosis of cystic fibrosis.*
- Qualification of Genotype (0.25)
  Qualification of the genotype should be included in the report, by mentioning that both parents need to be tested to confirm compound heterozygosity and, thereby, the diagnosis of CF in the child. In the present case, it would also help confirm the novel double mutation c.827_828delinsAA. It has also implications for the parents and relatives if prenatal diagnosis is required or if testing of relatives is to be undertaken.
- Suggest/offer PGD/PND to the parents for next pregnancy (0.25)

In case one mutation is detected

- Suggest/offer further testing (1.25)
  In case only one mutation is found, advice for further testing (in another laboratory) should be written in the report.
- Sensitivity is discussed or given (0.25)
  The diagnostic sensitivity of the test used should be given and refer to DW's geographic origin.
  There are different figures, notably depending on the region of Austria (95% in Tyrol: and 75% in Styria: (Stuhrmann et al. Clin Genet, 1997). Both were accepted. Sensitivity applying to the European or Caucasian population was scored as +/- in the Table. Sensitivity applying to a local population, for example Spain, was scored as -.
- Inappropriate/misleading comment (marks deducted depending on the context)
  Mentioning or offering PND in case only one mutation was found is potentially dangerous (without mentioning any further specifications: e.g. it can be done in case a 2nd mutation is found or if linked intragenic markers are identified) (-0.5).
  A number of laboratories provided a residual risk for DW of carrying a 2^{nd} mutation, solely based on the diagnostic sensitivity of the test. This was considered as a misleading comment (-0.5) as, based on clinical symptoms and positive sweat tests, there was a high suspicion of CF and, thereby, the presence of a 2^{nd} CF-causing mutation.

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In case one or two mutations are detected

- Cascade screening in relatives (0.25)
  The possibility of cascade screening for relatives should be mentioned in all cases in which an individual is positive for a CF-causing mutation.
- Genetic counselling for the parents (0.25)

Case 2:

- CF carrier testing is requested for Elisa Pennec who is a healthy woman and whose partner, Erwan Pennec, has a nephew diagnosed with cystic fibrosis at birth. Erwan Pennec was found to be heterozygous for c.1521_1523del, p.Phe508del (F508del) in your laboratory. The couple is planning a pregnancy and would like to know their risk of having a child with CF. There is no history of CF in Elisa's family, but she knows that her brother is followed for infertility. Elisa and Erwan are both from Brittany.
- **CF11-2:** Elisa Pennec - Brittany, France, DOB 10/03/1986
- **Expected genotype result:**
  - c.[350G>A(;)1521_1523del] (F508del/R117H)
    with neutral variants c.1210-12T[7];[9] (T7/T9)
  - Likely genotype: c.[350G>A;1210-12T[7]];[1521_1523del;1210-12T[9]]
    Whenever c.350G>A (p.Arg117His, R117H) is detected, it is recommended to test the polyT variant at c.1210-12 and report on it. Whether p.Arg117His is associated with the splicing c.1210-12T[5] variant or with the normal c.1210-12T[7] variant may influence genetic counselling for the couple.

- **Expected interpretative criteria:**
  - Confirmation of CF carrier status (0.50)
    The confirmation of the cystic fibrosis carrier status should be clearly stated, associated with the finding of c.1521_1523del (p.Phe508del, F508del).
  - Comment on variable phenotype associated with F508del/R117H (0.25)
    The disease risk associated with the compound heterozygous c.[1521_1523del];[350G>A;1210-12T[7]] (F508del/R117H;T7) genotype actually varies from **mostly** no symptom to CFTR-related disorders, such as CBAVD, and very rarely to classical CF (Thauvin-Robinet et al. J Med Genet 2009).
  - Risk figure for the couple of having a child with classical CF (0.25)
    Clear statement of classical CF for a child (0.25) It is important to distinguish between the different possible outcomes for pregnancies: having a child with classical CF, associated with the c.1521_1523del homozygous genotype (risk ¼), or having a child at risk of a CFTR-related phenotype, associated with the c.[350G>A;1210-12T[7]];[1521_1523del] genotype (risk ¼; low penetrance and variable phenotype).
  - Suggest/offer PND/PGD to the parents for the next pregnancy (0.25)
    The availability of prenatal diagnosis (PND) or pre-implantation genetic diagnosis (PGD) for future pregnancies should be stated for the risk of classical CF.
  - Cascade screening in relatives (0.25)
    The possibility of cascade screening for relatives should be mentioned in all cases in which an individual is positive for a CF-causing mutation. Based on literature data, it may not be recommended to suggest testing c.350G>A (R117H) on a T7 background in relatives (Thauvin-Robinet et al. J Med Genet 2009).
  - Genetic counselling for EP and his partner (0.25)
  - Inappropriate/misleading comment

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A number of laboratories still consider c.350G>A (R117H) as a CF-causing mutation. Stating that the diagnosis of CF is confirmed in EP or that she carries two CF-causing mutations or that she has a 50% chance of having a CF child was scored as a misleading comment (-0.5).

Case 3:

- Catalina Sacharov is the healthy partner of Andrei Dobre, a CF patient who is homozygous for c.1521_1523del, p.Phe508del (F508del). The couple requests assisted reproduction to have children and they would like to know their risk of having a child with cystic fibrosis. CF carrier testing is requested for Catalina. There is no history of CF in Catalina's family. Catalina and Andrei are both from Romania.
- **CF10-3:** Catalina Sacharov - Bucharest, Romania, DOB 14/09/1983
- **Expected genotype result:** c.[3909C>G];[=] or N1303K/normal

- **Expected interpretative criteria:**
  - Confirmation of CF carrier status (0.5)
  - Risk figure for the couple of having a child with CF (0.75)
    The risk of having a child with cystic fibrosis is 1/2.
  - Suggest/offer PND/PGD to the couple (0.25)
  - Cascade screening in relatives (0.25)
  - Genetic counselling for the couple (0.25)

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ANNEX 1 – ORGANIZATION OF THE SCHEME

CF EQA Coordination:

KU Leuven, Department of Public Health and Primary Care, Biomedical Quality Assurance Research Unit, Herestraat 49, box 602, 3000 Leuven, Belgium
Fax: +32 (0)16 34 71 90, E-mail: cf.network@med.kuleuven.be, Website: http://cf.eqascheme.org/
Co-ordinator: Prof. Dr. Els Dequeker, Tel: +32 (0)16 34 58 81, els.dequeker@med.kuleuven.be
Assistant co-ordinator: Sofie Delen, Tel: +32 (0)16 33 01 43, sofie.delen@med.kuleuven.be
Administrative assistant: Romy Gentens, Tel: +32 (0)16 34 60 33, romy.gentens@med.kuleuven.be

Medical/technical expert:

Dr. Emmanuelle Girodon
Hôpital Henri Mondor, UF de Génétique, Service de Biochimie et de Génétique, Créteil, France,
emmanuelle.girodon@hmn.aphp.fr

Assessors:

Dr. Marie des Georges
Institut Universitaire de Recherche Clinique, Public Health - Hospital Medical School, Laboratoire de Génétique Moléculaire, Montpellier, France, marie.desgeorges@inserm.fr

Dr. Christof Meyer-Kleine
Synlab MVZ Bad Nauheim, Bad Nauheim, Germany, Christof.Meyer-Kleine@synlab.com

Dr. Michael A. Morris
Synlab FAMH génétique médicale, Directeur diagnostic moléculaire, Lausanne, Switzerland,
michael.morris@synlab.com

Dr. Martin Schwarz
St Mary's Hospital, Genetic Medicine, Regional Molecular Genetics Service, Manchester, UK,
Martin.Schwarz@cmft.nhs.uk

Dr. Raina Yamamoto
Laboratoriumsmedizin Dortmund, Dr. Eberhard und Partner, MVZ, Molekulargenetik, Dortmund, Germany,
yamamoto@labmed.de

Prof. Dr. Els Dequeker, Dr. Emmanuelle Girodon

Assistant assessors:

Sofie Delen & Lien Tembuyser
KU Leuven, Department of Public Health and Primary Care, Biomedical Quality Assurance Research Unit, Leuven, Belgium, lien.tembuyser@med.kuleuven.be

Sponsoring:

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The CF Network is grateful to Abbott and Hologic Gen-Probe. Thanks to their sponsoring, 12 laboratories from a country with a Human Development Index below 0.810 (Lithuania, Latvia, Croatia, Serbia, Ukraine, Macedonia, Turkey) were able to participate at a reduced fee.

(HDI is an index defined by the United Nations: en.wikipedia.org/wiki/List_of_countries_by_Human_Development_Index)