Workshop on
An Introduction to Quality Assurance
in Genetic Diagnostic Laboratories

Molecular Genetics, Cytogenetics, Biochemical Genetics
& Newborn screening

PROGRAMME BOOK

6 & 7 February 2012

Colombo, Sri Lanka

Organized by the Human Genetics Unit, Faculty of Medicine
University of Colombo
Sri Lanka
In collaboration with the
European Molecular Genetics Quality Network
Workshop on
An Introduction to Quality Assurance in Genetic Diagnostic Laboratories

Molecular Genetics, Cytogenetics, Biochemical Genetics & Newborn screening

6 & 7 February 2012

Colombo, Sri Lanka
TABLE OF CONTENTS

Messages

Director, Human Genetics Unit 1

Chairman, European Molecular Genetics Quality Network 2

Former Chairman, European Molecular Genetics Quality Network 3

Sponsors, Contributors and Endorsements 5

Aims of the workshop 7

Programme

Inauguration 9

Day 1 11

Day 2 13

European Faculty 15

Sri Lankan Faculty 21

Contents of the Delegate Pack 23

List of Participants 25
MESSAGE FROM THE DIRECTOR, HUMAN GENETICS UNIT

I am delighted that the Human Genetics Unit was able to organise this workshop in Colombo in collaboration with colleagues from Europe at a time when genetic diagnostic laboratories in Sri Lanka are looking at improving the quality of the services that they provide.

I am particularly pleased that scientists working in all genetic diagnostics laboratories in Sri Lanka have accepted our invitation to the workshop. I hope that this would be the beginning of networking between the genetic diagnostics laboratories in Sri Lanka and that that would lay the foundation for the formal establishment of the network.

I am particularly pleased that so many organizations have come together to support this workshop. The endorsement of the Sri Lanka Accreditation Board (SLAB) is particularly significant, because the local network that is established following the workshop can work with SLAB to establish a genetics quality network in Sri Lanka for both cytogenetics and molecular genetics.

I am delighted to note that Prof. Vajira H. W. Dissanayake, has as usual, been able to muster the support of so many organizations to ensure that a workshop of the highest standard is being conducted in Sri Lanka.

I thank the organizations sponsoring and endorsing this workshop as well as the European resource persons for their valuable contribution to enhance the quality of genetic diagnostic services in Sri Lanka.

Prof. Rohan W Jayasekara MBBS, PhD (N’cle), C.Biol, MSB(Lond.)
Chair and Senior Professor of Anatomy
Director, Human Genetics Unit
Dean, Faculty of Medicine
MESSAGE FROM THE EUROPEAN MOLECULAR GENETICS QUALITY NETWORK

EMQN is delighted to be involved in the Workshop on Quality Assurance in Genetic Diagnostic Laboratories, along with colleagues from CEQA and ERNDIM. EMQN’s mission is to promote quality in genetic testing worldwide. Genetic testing is now part of mainstream laboratory diagnostics, but it retains its unique characteristics: most genetic tests are only carried out once in a person’s lifetime and the results may have far-reaching implications for the patient and their family. These two aspects of genetic testing combine to bring quality sharply into focus - it is essential that we have systems in place to ensure that we get the correct result to the right patient every time.

We hope that you find this workshop useful and that it will be just the start of a long and fruitful relationship between EMQN and the laboratory geneticists of Sri Lanka.

Prof. David Barton
Chief Scientist, National Centre for Medicinal Genetics, Dublin, Ireland.
Chairman, European Molecular Genetics Quality Network
MESSAGE FROM THE EUROPEAN MOLECULAR GENETICS QUALITY NETWORK

On behalf of my colleagues from the European Quality Assessment organizations we would like to thank Professor Dissanayake and his colleagues for inviting us to Colombo and for the opportunity to talk about the ideas behind laboratory quality assurance, about our experience and some prospects for the future. Genomic analysis will be at the centre of modern medical practice in the 21st century and it is of the utmost importance that we practice the highest standards in order to retain the confidence of the public in this new and exciting technological and scientific area.

Dr. Rob Elles
Clinical Director, Genetic Medicine, Manchester UK
Former Chairman, Board Member, European Molecular Genetics Quality Network
SPONSORS, CONTRIBUTORS AND ENDORSEMENTS

EMQN: The European Molecular Genetics Quality Network

CEQA: Cytogenetic European Quality Assessment

ERNDIM: Biochemical Genetics EQA

EuroGentest

Human Genetics Unit, Faculty of Medicine, University of Colombo

National Science Foundation, Sri Lanka
Sri Lanka Medical Association

Sri Lanka Accreditation Board
AIMS OF THE WORKSHOPS

Quality assurance is central to the safe and efficient operation of all medical laboratories. This workshop will introduce scientists, technicians and managers from genetic diagnostic laboratories to the development and operation of a Quality Management System compliant with international standards. In addition the workshop will consider international guidelines specific to genetic laboratories. There will be a focus on how External Quality Assessment (Laboratory Proficiency Testing) can be used by a laboratory to compare its performance against its peers and improve its practices.

TOPICS WILL INCLUDE:

• The place of a Quality Management System in laboratory practice and achieving accreditation
• The international standards for Quality Management – ISO17025/ISO15189
• Guidelines and Regulatory frameworks for Genetic Testing in Europe, Sri Lanka and India
• Concepts in Quality Assurance
• Developing a Quality Management System, the role of the Quality Manager and the Quality team, the individual responsibility of each member of staff.
• Systems for document control
• Test Validation. The role of controls and reference materials
• Laboratory organization related to quality, risk analysis and the role of Internal Quality Control procedures
• External Quality Assessment including EQA for new technologies in Molecular Genetics, Cytogenetics, Biochemical Genetics
• Integrating EQA into routine practice
• Practical exercise in internal audit
• Prospects for developing Quality Assurance practices in genetic laboratories Sri Lanka
CONTINUED PROFESSIONAL DEVELOPMENT

The workshops have been approved by the Royal College of Pathologists of London for CPD purposes to a maximum of 11 credits (excluding refreshment breaks).

Medical staff and clinical scientists in career grade posts who are enrolled with one of the UK Royal Colleges for CPD purposes and attend the meetings will be entitled to receive CPD credits.

LOCAL HOST

Prof. Vajira H. W. Dissanayake, Professor, Human Genetics Unit, Faculty of Medicine, University of Colombo
Inauguration

February 06, 2012
Venue: New Lecture Theatre, Faculty of Medicine, University of Colombo

09.00  Arrival of Invitees

09.05  Welcome Address
Prof. Rohan W Jayasekara
Dean, Faculty of Medicine
University of Colombo

09.10  Address by the Head of the European Molecular Genetics Quality Network
Prof. David Barton
Chairman, European Molecular Genetics Quality Network

09.20  Address by the Guest of Honour
Prof. Kshanika Hirimburegama
Vice Chancellor, University of Colombo

09.30  Address by the Chief Guest
Mr. Thilak Wickremasinghe
Director/CEO, Sri Lanka Accreditation Board

09.40  Vote of Thanks
Prof Vajira HW Dissanayake
Professor, Department of Anatomy/ Human Genetics Unit
Faculty of Medicine
University of Colombo

09.45  Refreshments
Day 1
February 06, 2012
Venue: Department of Anatomy, Faculty of Medicine, University of Colombo

10.00 - 10.40 Guidelines and Regulatory frameworks for Genetic Testing in Europe and Sri Lanka
OECD and Council of Europe, Direct to consumer testing - examples of National legislation e.g. HGC framework for DTC.
Dr. Rob Elles

Legal framework and regulation in Sri Lanka
Prof. Vajira HW Dissanayake

10.40 – 11.40 Concepts in Quality Assurance
The Deming cycle
Quality Improvement
Planning
Review
Dr. Rob Elles

The place of a Quality Management System in laboratory practice and achieving accreditation
Planning for accreditation
A long term effort
The audit – what to expect
Dealing with non conformance
Maintaining a level of compliance
Dr. Ros Hastings

ISO9000, ISO17025, ISO15189 etc
Certification and accreditation
CAP and other standards
Mr. Thilak Wickremasinghe

12.40 – 13.40 Lunch
13.40 – 14.10  **Practical exercise in internal audit (1)**
Reporting policy
Dummy reports
Audit form
*Dr. Simon Patton*

14.10 – 14.40  **Developing a Quality Management System, the role of the Quality Manager and the Quality team, the individual responsibility of each member of staff**
A starting point
The Quality Manager friend or foe?
The Quality Team
No time for quality?
Measuring the costs
Measuring the benefits
*Dr. Simon Patton*

14.40 – 15.10  **Systems for document control and operating a QMS**
Paper based systems
Q-Pulse and other electronic systems
Document control as a starting point
Policies, SOPs and Forms
Document review

15.10 – 15.30  **Tea Break**

15.30 – 16.15  **Test Validation. The role of controls and reference materials**
Controlled In Vitro Diagnostics and Laboratory Developed Tests
Validation of a LDT/guidelines
Documentation of validation
Controls and Reference Materials – availability and use
*Prof. David Barton*
Day 2  
February 07, 2012

09.00 – 09.45  Quality Assurance in Biochemical Genetics and New-born screening  
EQA and IQC in Biochemical Genetics – examples  
Precision and accuracy  
Relevance of Laboratory Quality Assurance to the outcome for patients  
Why EQA needs to be international  
Future requirements  
Prof. Brian Fowler

09.45-10.10  Tea Break

10.10 – 10.40  External Quality Assessment including EQA for new technologies in: Molecular Genetics and EMQN  
Prof. David Barton

10.40 - 11.10  External Quality Assessment including EQA for new technologies in: Cytogenetics and CEQA  
Dr. Ros Hastings

11.10 – 11.40  External Quality Assessment including EQA for new technologies in: Biochemical Genetics and ERNDIM  
Prof. Brian Fowler

11.40 - 13.00  Lunch

13.00 – 13.40  Integrating EQA into routine practice  
Regular participation  
Attitude to the EQA sample  
Reporting – Deadlines  
Review of results  
Learning and improving  
If things go wrong  
Appealing
Making participation public
Dr. Ros Hastings

13.40 – 15.00
Practical exercises in internal audit (2)
Results of the audit (groups)
Lessons
Dr. Simon Patton

15.00 -15.20
Tea Break

15.20 – 15.50
Prospects for developing Quality Assurance practices in genetic laboratories in Sri Lanka - round table
Dr. Rob Elles and Prof. Vajira HW Dissanayake

15.50 – 16.10
Conclusions and close
Dr. Rob Elles and Prof. Vajira HW Dissanayake
EUROPEAN FACULTY

Dr. Rob Elles PhD, FRCPath
Clinical Director - Genetic Medicine, Manchester UK
Member - Service Development Group, Human Genomics Strategy
Group (UK Department of Health)
Department of Health Chief Scientist’s award for delivering quality 2009
Former Chairman - British Society for Human Genetics, and European Molecular Genetics Quality Network

Rob Elles is a Fellow of the Royal College of Pathologists and Past Chairman of the British Society for Human Genetics; representing healthcare professionals in the UK National Health Service and scientists in diagnostics and research.

He is currently the Clinical Director of Genetic Medicine in Manchester, UK. Genetic Medicine is an integrated Clinical, Laboratory Service and Research Department. It employs 250 Medical, Counseling, Scientific and support staff offering Genetic Services to a Regional population of nearly 5 million and many national specialty services.

He worked as a research technician in the Department of Biochemistry, University of London in the early days of applying recombinant DNA technologies to Human Genetics. In 1983 he moved to the Genetics Service in Manchester. He completed his PhD whilst helping build the Molecular Genetics laboratory into a group of over 40 scientists and technologists. In 2002 his Laboratory was awarded one of two Department of Health contracts to develop National Genetics Reference Laboratories. The NGRLs support the UK Genetic Testing Network in informatics, technology assessment and quality assurance.

Dr Elles has been involved in training and professional standards and many areas of development of diagnostic genetics in the UK. He was secretary of the Clinical Molecular Genetics Society and in 1998 coordinated a
European Commission research project which developed into the self supporting European Molecular Genetics Quality Network which offers 28 Laboratory Proficiency Testing (LPT) schemes to nearly 800 laboratories in 42 countries. He is a partner in the EuroGentest work-group centered on LPT for molecular genetics. He was co-chair of the expert steering group on Quality Assurance in Molecular Genetic Testing of the Organization for Economic Co-operation and Development that adopted guidelines for genetic testing in 2007.


Dr. David E Barton, PhD, DipRCPath
Chief Scientist, National Centre for Medical Genetics, Dublin, Ireland
Chairman, European Molecular Genetics Quality Network
Member, Steering Committee, EuroGentest Network of Excellence
Member, Genetic Services Quality Committee, European Society of Human Genetics
Past Member, Steering Committee, UK NEQAS in Molecular Genetics
Past Treasurer, Clinical Molecular Genetics Society

David Barton is Chief Scientist (Director) of the Molecular Genetics Laboratory at the National Centre for Medical Genetics in Dublin, Ireland, and Adjunct Associate Professor at University College Dublin. He took over from Rob Elles as Chairman of EMQN in March 2011. Having trained in Trinity College Dublin, he carried out medical genetics research at Yale University and Cambridge University before setting up the NHS molecular genetics diagnostic laboratory in Cambridge. He returned to Dublin to set up his current laboratory at the National Centre for Medical Genetics in 1995.

Dr Barton has been involved in work to monitor and improve the quality of genetic testing for many years, working with UK NEQAS, the European
Molecular Genetics Quality Network, the OECD and EuroGentest. He coordinated the EU CRMGEN project, developing certified reference materials for genetic testing. In EuroGentest he continues to work on reference materials development and also has responsibility for examining the role and impact of IVD regulation in genetic testing.

David Barton has published over 100 papers in peer-reviewed journals on many aspects of the molecular genetics of inherited disorders. Research interests include the genetics of vesicoureteral reflux, male infertility and the development of novel DNA diagnostic devices.

He was a member of the expert steering group on Quality Assurance in Molecular Genetic Testing of the Organization for Economic Co-operation and Development (OECD) that adopted guidelines for genetic testing in 2007.

Related web links: www.genetics.ie; www.emqn.org; www.eurogentest.org;

Prof. Brian Fowler, PhD
Head of Laboratory, University Children’s’ Hospital (UKBB), Basle, Switzerland
Chairman ERNDIM (Biochemical Genetics External Quality Assessment Scheme)
Member, Genetic Services Quality Committee, European Society of Human Genetics
President of the Swiss National Society for Inherited metabolic disorders
ERNDIM member within the EuroGentest Network

Brian Fowler is currently Head of Labs and Biochemist at the University Children's Hospital Basel and has a long standing interest in inherited metabolic disease, specializing in disorders of homocysteine and related vitamin metabolism.

He was born in Manchester, England in 1946 where he studied
biochemistry. He attained the degree of Ph.D. at the Dept. of Medical Biochemistry, University of Manchester in 1972. Post doctoral studies at the Royal Manchester Children's Hospital were followed by a period as Research Associate at Yale University. In 1976 he returned to the Royal Manchester Children's Hospital as Head of the laboratory section of the Willink Biochemical Genetics Unit until 1990 when he moved to take up his present post. Between 1988 and 1999 he was honored to be first, Honorary Secretary and then Chairman of the Society for the Study of Inborn Errors of Metabolism. From 1999 to 2008 he has undertaken the role of Chairman of ERNDIM, the European network for quality control in inherited metabolic disorders and is currently

**Related web link:** www.erndim.org

**Dr. Ros Hastings PhD, FRCpath**  
Scheme Co-ordinator Cytogenetics European Quality Assessment (CEQA)  
Scheme Organiser UK NEQAS for Clinical Cytogenetics  
Chair of ESHG Genetic Services Quality Committee  
ESHG Board Member  
Unit leader within the EuroGentest Network

Ros Hastings is a state registered scientist and is a Fellow of the Royal College of Pathologists. She is the Scheme Organiser/Co-ordinator of the European EQA scheme, CEQA (Cytogenetics European Quality Assessment) and the National UK NEQAS for Clinical Cytogenetics EQA scheme. Both Schemes are based at the John Radcliffe Hospital, Oxford and between them have more than 450 registered laboratories.

Dr Hastings is Chair of the ESHG Diagnostic Service Quality Committee whose main aim is to improve the quality and provision of Genetic services in Europe for the benefit of patients. She coordinates the ESHG Workshop on Diagnostic Cytogenetics held annually at the ESHG conference. In addition, she is one of the co-ordinators of the ECA Permanent Working Group on Cytogenetics and Society, and co-author of the European Cytogenetic Guidelines. In 2010 and 2011, Dr Hastings coordinated two
Symposia (ESHG satellite meetings) on the ‘Changing landscape of genetic testing’ and ‘Array in daily practice; promises and pitfalls’

Dr Hastings has more than 30 years experience in Constitutional (Postnatal and Prenatal), Cancer and Molecular Cytogenetics, both in a research and diagnostic setting. At the beginning of her career she researched into Cancer and immunological disorders at the Imperial Cancer Research Fund, London, the MRC Cytogenetics Unit, Edinburgh and the Cancer Research Campaign Laboratory, Manchester. Dr Hastings has also worked in four diagnostic Cytogenetics Laboratories within the UK, in senior positions ranging from Head of Prenatal Section to Deputy Director, and was responsible for signing out cytogenetic reports for all aspects of Cytogenetics. For the last 10 years Dr Hastings has been involved in External Quality Assessment.

CEQA is a European EQA scheme that started in 2006 through EU funded EuroGentest Network. The UK NEQAS for Clinical Cytogenetics has been affiliated to the UK NEQAS consortium for 30 years. Both EQA schemes cover constitutional and acquired cytogenetics and participating laboratories are from Europe, Asia, Australia and South America. The EQA process involves the distribution of EQA samples and the assessment of Clinical Cytogenetics reports from Amniotic Fluids, CVS, QF-PCR, Bloods, Solid Tissues, AML, ALL, CML, MDS, Solid Tumours, B-cell Lymphoproliferative Disease, Fanconi Anaemia, Preimplantation diagnosis, Rapid aneuploidy FISH and Microarray. The EQA schemes assess laboratory performance and are both educational and useful for benchmarking performance. The Cytogenetics EQA schemes have published guidelines and their findings in the scientific journals and books.

Related web links:
www.ceqa-cyto.eu
www.ccneqas.org.uk
www.eshg.org
www.biologia.uniba.it/eca/www.eurogentest.org
www.eurogentest.org
Dr. Simon Patton PhD
Executive Administrator and Board Member – European Molecular Genetics Quality Network (EMQN)
Quality Manager – Genetic Medicine, Manchester UK
Member, Steering Committee, UK NEQAS in Molecular Genetics

Simon Patton is currently the Executive Administrator of the European Molecular Genetics Quality Network (EMQN), a post he has held since 1999. The network is the leading provider of Quality Assurance Schemes to Diagnostic Molecular Genetics Laboratories in the world. It has over 950 registered laboratories participating in its activities and has recently become accredited to the international EQA standard ISO 17043. The EMQN is based at St Mary’s Hospital in Manchester (UK), which is an accredited laboratory, providing a diagnostic service to a population of 4.2M in the eastern part of the North West Region of the UK. The laboratory has a long track record in the development of quality assurance systems for diagnostic molecular genetics.

He is also involved in many other Quality Assurance activities including lecturing, course development and consultancy.

Related web links:
www.emqn.org.uk
www.ngrl.org.uk
SRI LANKAN FACULTY

Prof. Vajira H W Dissanayake  MBBS (Colombo), PhD (Nottingham)
Professor, Department of Anatomy
Medical Geneticist, Human Genetics Unit, Faculty of Medicine, University of Colombo.
Medical Geneticist, Asiri Centre for Genomic and Regenerative Medicine, Asiri Group of Hospitals
President, Sri Lanka Medical Association
President, Health Informatics Society of Sri Lanka

Vajira H W Dissanayake established the Molecular Genetic Diagnostic laboratory in the Human Genetics Unit in 2004 and the Asiri Centre for Genomic and Regenerative Medicine, at the Asiri Group of Hospitals in Colombo in 2006. He has been in the forefront of introducing new genetic techniques to the Country.

He leads a research group in the University of Colombo focusing on genetics of reproductive disorders, especially pre eclampsia, fetal growth restriction and recurrent pregnancy loss. Prof. Dissanayake coordinates M.Sc. courses in Biomedical informatics, Clinical Genetics and Genetic Diagnostics in the University of Colombo and is a member of the steering committee of the Forum for Ethics Review Committees in Asia and the Western Pacific, President of the Health Informatics Society of Sri Lanka and President of the Sri Lanka Medical Association. He was the principal investigator in the team that sequenced the first Sri Lankan Personal Genome.

Related web links:
www.hgucolombo.org
www.asiri.lk/ACGRM
Thilak Wickremasinghe  BSc (Hons), MSc, MBA

Director/Chief Executive Officer, Sri Lanka Accreditation Board
Lead Assessor, Accreditation of testing and calibration Laboratories, medical laboratories and certification bodies
Core tutor on Assessor training for accreditation of laboratories and certification bodies
APLAC evaluator of accreditation bodies

Related web links:
www.slab.lk
CONTENTS OF THE DELEGATE PACK

- Programme
- Copy of the OECD Guidelines
- Copy of the Council of Europe Statement
- Reprints of Eurogentest test validation guidelines – Mattocks et al.
- Reprints of lecture PowerPoint hand-outs
- Audit exercise documentation:
  - Introduction to the audit exercise
  - A laboratory reporting policy
  - OECD Guidelines on result reporting
  - Forms for recording reporting audit exercise (locally printed)
- Workshop feed-back form
- Attendance and Continued Professional Development credit certificate (where requested)
- Information on EQA schemes (provided by EMQN, ERNDIM and CEQA)
LIST OF PARTICIPANTS

Human Genetic Unit, Faculty of Medicine, University of Colombo

1. Prof Vajira H W Dissanayake
2. Dr Hemali W W Goonasekera
3. S P N Sandamali Senanayake
4. D A J Niranjala Kumari
5. A A G S Abeysekara
6. U G Imalki Upamalika Kariyawasam
7. Jeewani sanketha Opanayaka
8. Dr Prasadi N De Silva
9. Dinuka Markalanda
10. Vindya Lankika Kumari
11. Dr Nilakshi Samaranayake
12. Gayani Galhena
13. E M Nihal Rosa de Silva
14. P K D S Nisansala
15. Manjula Niluka Rajarathna

Molecular Medicine Unit, Faculty of Medicine, University of Kelaniya

1. Dr Nilmini Silva Gunawardene
2. Dr Aresha Manamperi

Genetic Laboratory, Asiri Surgical Hospital, Colombo

1. Shalani Thirukeshwaran
2. Kumarathy Kumaraguru
3. Suresh De Silva
Genetic Laboratory, Durdans Hospital, Colombo

1. Miss. Hemanthini Kalubowila

GENETECH Sri Lanka

1. Manisha Gunasekara
2. Dilshani Fernando

Sri Lanka Accreditation Board

1. Mr Thilak Wickramasinghe
2. Mr Rahal Widanagamage
3. Ms Manisha Wickramasinghe

Department of Obstetrics and Gynaecology, Faculty of Medicine, University of Colombo

1. Prof. Sumeda Wijerathne

Department of Basic Sciences, Kotalawala Defense University

1. Maj. WMMS Bandara
2. AJIS Rathnayake

Faculty of Medicine, University of Jaffna

1. Dr Thurayratnam Chenthuran

Molecular Genetics Laboratory, Medical Research Institute

1. Mr. R. Ramesh