Creating the Ecosystem for Taking Genetics from Bench to Bedside in a Developing Country: A Personal Experience from Sri Lanka

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University of Colombo
Mihintale Hospital - The most ancient hospital in the World
437 – 367 BC
Colombo Medical School [Established 1870] - The second oldest Medical School in Asia

Sri Lanka Medical Association [Established 1887]  
The oldest National Medical Association in Asia and Australasia
Only Medical Genetics Center in Sri Lanka
Provide Clinical/Diagnostic Genetic Services, Provide Undergraduate and Postgraduate Training, and Conduct Research by itself and in collaboration with academic and the private sector both nationally and internationally

Serving a Population of 20.1 Million People
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Vision
To help Sri Lankan families live healthier and happier lives.

Mission
To be the leading Translational Genomic Medicine Institution in South Asia by facilitating the transfer of technology from bench to bedside in the field of genomic medicine by developing clinical genetics and genetic diagnostic services; training medical and allied health staff to provide medical genetics services; conducting genetic research and engaging in advocacy to promote universal access to and availability of genetic services.
International Collaborations

Intentional Genetic Education Network (IGEN)
Forum for Ethics Review Committees in Asia and the Western Pacific (FERCAP)
European Molecular Quality Network (EMQN)
Pan Asian Personal Genomics Initiative
Indo-UK Genetics Network

Canada
University of British Columbia

USA
University of Houston
Pennsylvania State University
University of Texas

Italy
Cure2Children Foundation

UK
Kings College, London
University of Durham
University of Leeds

Norway
University of Oslo

France
Pasteur Institute

India
Institute of Genomic & Integrative Biology
Manipal Institute of Regenerative Medicine

Nepal
National Academy of Medical Sciences
Global Hospitals

Singapore
National University of Singapore

International Collaborations with
16 institutions in 8 countries in 3 continents
5 Networks
Services Available

Medical Genetics

- Genetic Evaluation and Counseling
- Cytogenetic Diagnostics
- Molecular Genetic Diagnostics
- Functional Genetics
- Bioinformatics

1983 Established
- Karyotyping
- FISH

2004 Established
- Automated Karyotyping
- Real Time PCR
- Sanger Sequencing
- NGS

2010 Established
- Established

2011 Established
- Established
Medical Genetics
- Genetic Evaluation and Counseling
- Bioinformatics
- Functional Genetics
- Molecular Genetic Diagnostics
- Cytogenetic Diagnostics
- QA
  - ISO15189
  - EMQN
  - External QA

..taken to bedside

Karyotyping
- FISH – 22q11.2 deletion

Thrombophilia
- JAK2 V617F & Exon 12
- Haemachromotosis
- Thalassaemia
- Duchenne Muscular Dystrophy
- Spinocerebellar Ataxia
- Huntington Disease
- HLA-B27

Companion Diagnostics
- K-Ras
- Haemato-oncology
  - Bcr-Abl
- Oncology
  - BRCA1/2
  - RB1
  - MYCN
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Development and implementation of a web-based continuing professional development (CPD) programme on medical genetics

Gumindu GAK Kolutunga, Rohana B Marasinghe, Indika M Karunathilake and Vajira HW Dissanayake

Summary

We developed, implemented and evaluated a web-based continuing professional development (CPD) programme on medical genetics. Development of the CPD programme followed the ADDIE model, i.e. Analysis, Design, Develop, Implement and Evaluation. An invitation to participate in a needs analysis survey was sent to all doctors on the email list of the Sri Lanka Medical Association. A total of 129 completed surveys was received (57% of the 228 who accessed the online survey). The average age of respondents was 42 years (range 27-81). The male:female ratio was approximately 2:1. Almost all respondents (96%) selected web-based CPD programmes, or web-based and conventional lectures, as their preferred method of learning. The programme was piloted on a group of 10 doctors. The average pre-knowledge score was 40.2 and the post-knowledge score was
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Translational Goals for 2014-15

Expand Cancer Genetic Services

**Rationale**

Cancer incidence in Sri Lanka has increased 200% in the past 10 years.
The national cancer drug budget is increasing at an alarming rate.
Oncologists are interested in improving cancer genetics services including optimising therapy based on genetic profile of tumours to improve patient outcomes.

**Strategies**

Promote using family history as a tool for identifying families with inherited cancer syndromes in collaboration with the National Cancer Prevention Programme [Already commenced].

Provide CME to oncologists in partnership with the Sri Lanka College of Oncologists [Already commenced]

Improve haemato-oncology diagnostic service
   [Training already planned in collaboration with King’s College, London]

Introduce companion diagnostics for cancer
   k-RAS [already Introduced]
   EGFR mutation testing and HER2/NEU testing [identified for introduction]

Introducing NGS and Clinical Bioinformatics services for tumour tissue genomics.
Translational Goals for 2014-15

Prevent and ‘Cure’ Thalassaemia

Rationale

60-100 children are born with thalassaemia in Sri Lanka every year. [15 % 2nd child in the family]

The total thalassamic patient population of approximately 3000 in Sri Lanka take up 5% of the annual drug budget of the national health service for blood transfusions and ion chelating therapy which is provided free of charge to the patients.

Current HPLC based national screening programme is expensive and as such has not achieved the expected coverage.

Pregnancy termination is not legal in Sri Lanka. Even if legalised would be culturally unacceptable.

Cost of one BMT is equal to one year’s cost of blood transfusions and iron chelation.

Strategies

Introduce a cost effective population based screening programme for carrier detection using high throughput SNP genotyping and counseling.

Strengthen counseling services for families with thalassaemia.

Support the initiative to establish a bone marrow transplantation center for thalassaemia.

Conduct research to characterise clinical outcomes with primary and secondary genetic modifiers of thalassaemia with the view to using the genetic profile to optimise treatment converting the disease from a fatal one to a non fatal manageable illness.
Translational Goals for 2014-15

Prevent and Control Birth Defects and Inborn Errors of Metabolism

Rationale
Sri Lanka is unable to meet MDG4 because birth defects account for 10.1% of Neonatal Death and 18% of Infant Death.

Pregnancy termination is not legalised and even if legalised would be culturally unacceptable.

Most cases of birth defects go undiagnosed (Diagnostic yield of chromosome culture and karyotyping is approximately 5%. Microarray can improve this up to approximately 30%.)

Take up of genetic testing is low as genetic tests in the national health service are out of pocket for patients.

There is no nationwide new born screening programme although there are limited programmes for neonatal screening for hypothyroidism.

Strategies
Increase awareness about birth defects and prevention and control of birth defects

   International Conference on Birth Defects will be held from 9 to 12 February 2014

Introduce diagnostic microarray testing.

Advocate with the Ministry of Health for reimbursement for genetic testing.

Advocate with the Ministry of Health to establish a country wide newborn screening programme.
Translational Goals for 2014-15

Improve Genetic Literacy Among Medical and Allied Health Professionals

Rationale

Although there are 8 Medical Schools there is only one academic medical genetics department. All medical consultations in Sri Lanka before board certification have to serve a mandatory period of at least 1 year abroad (usually in UK or Australia), but when they return, although ‘genetically literate’, they do not know what genetic services are available. Genetics is not taught in most allied health courses.

Strategies

Introduce an online CME courses (Already commenced)
Conduct symposia and workshops with annual sessions of professional medical colleges and associations. (Ongoing)
Advocate for adoption of a core-curriculum in genetics in medical schools and in allied health courses (Already commenced with the IGEN conference in June 2013).
Establish the South Asian Genetics Education Network to promote interaction between South Asian Genetics Educators (Already commenced with the IGEN conference in June 2013. A proposal has been submitted to External Affairs Ministry for submission to the South Asian Secretariat).
Establish HGU-NAMS collaborating center in Kathmandu, Nepal (Already commenced).
Thank you