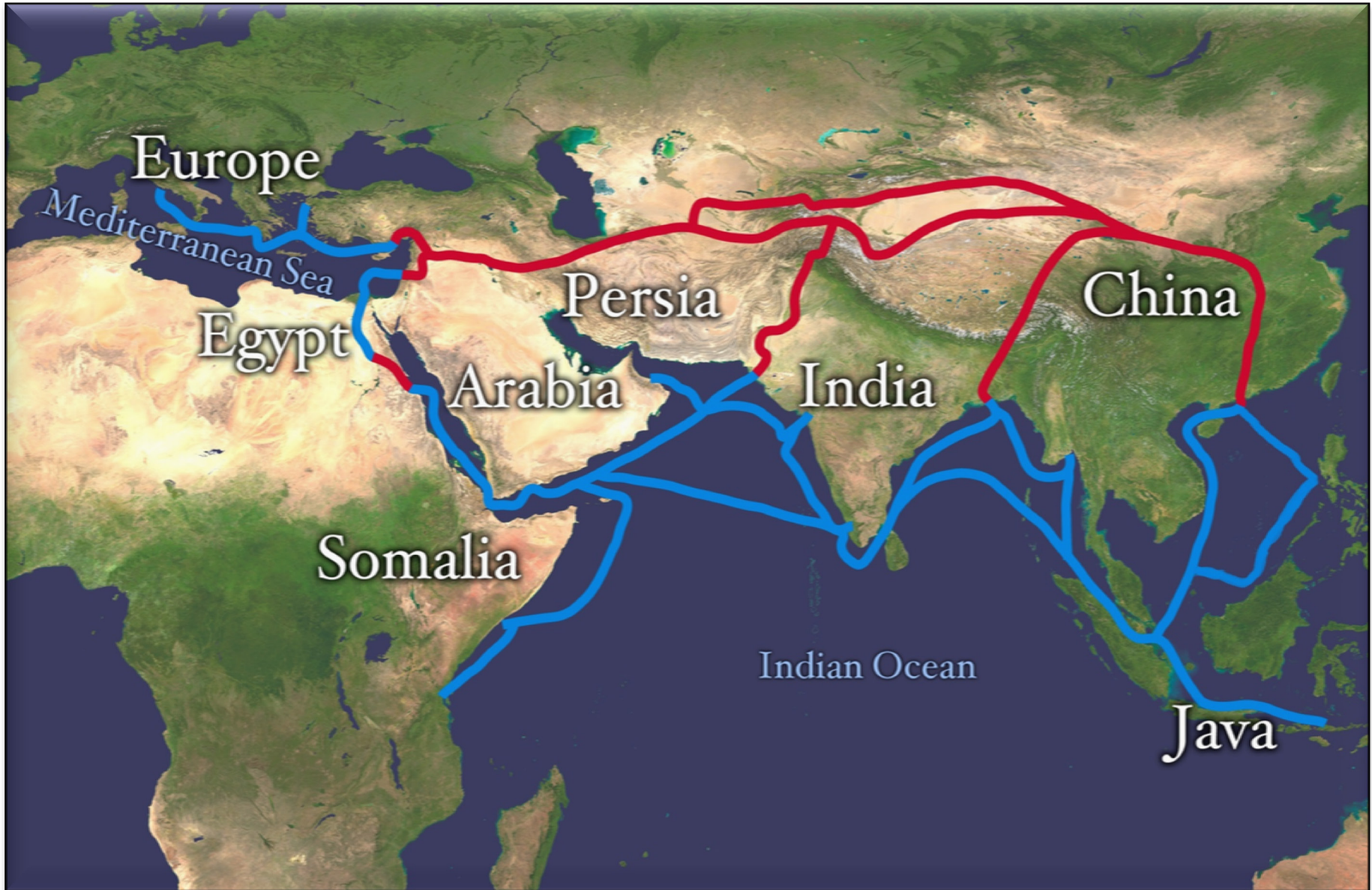


***Creating the Ecosystem for Taking Genetics from  
Bench to Bedside in a Developing Country:  
A Personal Experience from Sri Lanka***

***Prof. Vajira H. W. Dissanayake MBBS, PhD, FNASSL  
Professor and Medical Geneticist  
Human Genetics Unit, Faculty of Medicine  
University of Colombo***





Europe

Mediterranean Sea

Egypt

Arabia

Persia

India

China

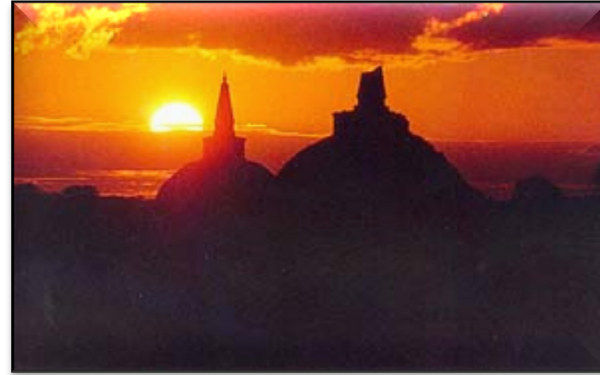
Somalia

Indian Ocean

Java



**300 BC**



**161-137 BC**



**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**

# UPCOMING

mark your calendars

## International Conference on Birth Defects 2014

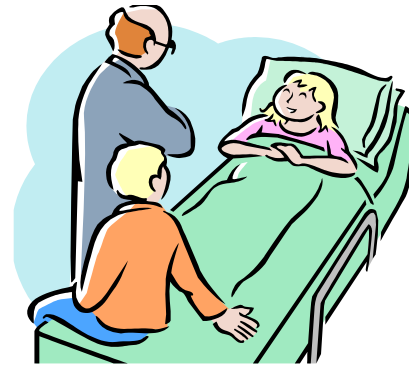
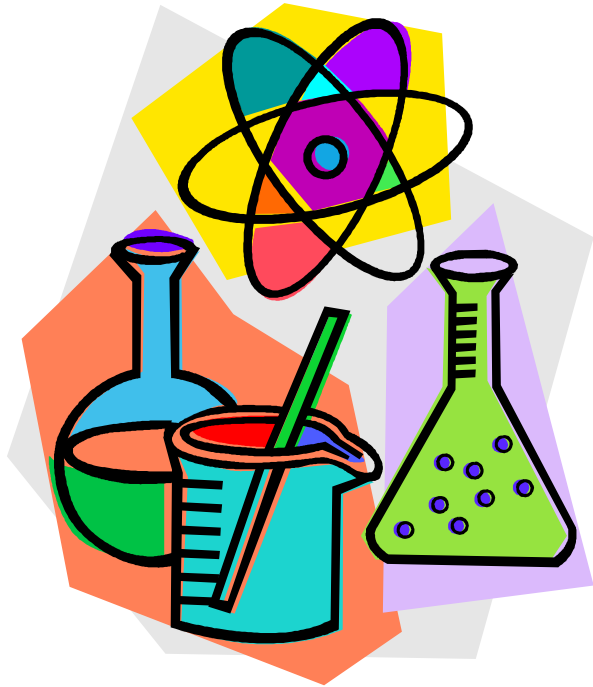
Upcoming Conferences

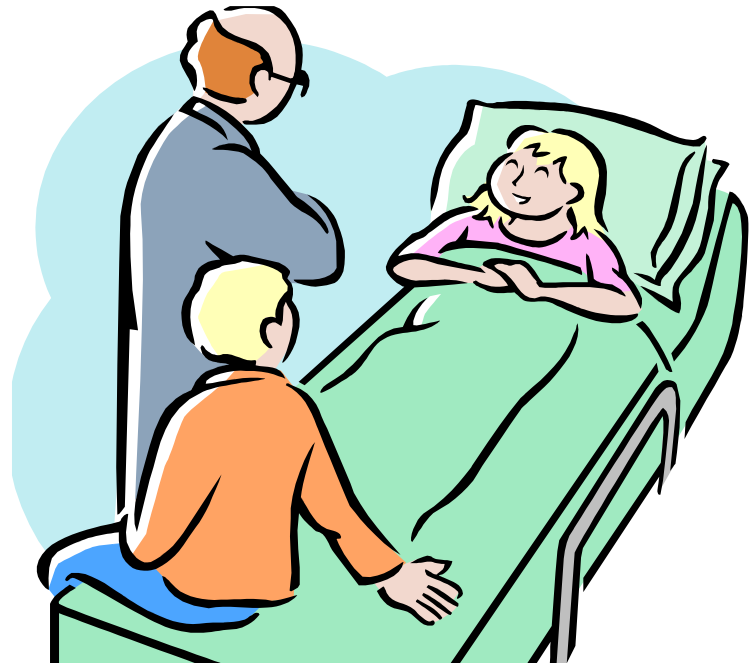
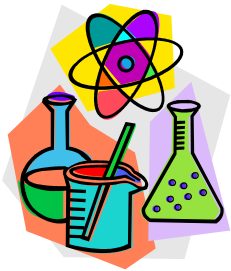
Human Genetics Unit of the Faculty of Medicine University of Colombo is the only centre in Sri Lanka dedicated to providing clinical genetic services including genetic testing and genetic counselling. In addition we conduct both undergraduate, postgraduate, and CME courses in genetics for medical and allied health professionals and programmes aimed at raising genetic awareness among the public. We have an active research programme in reproductive genetics. We play in advocacy to make genetic services available to the people of Sri Lanka and contribute to policy formation in the field of genetics in Sri Lanka.

**Only Medical Genetics Center in Sri Lanka**

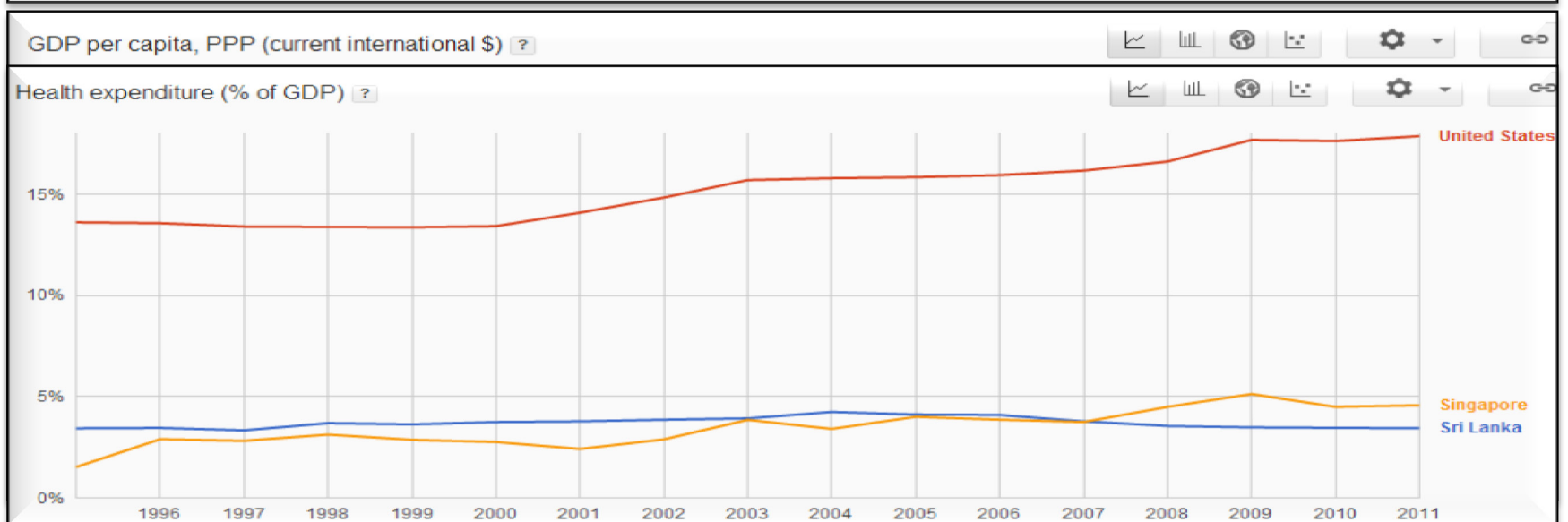
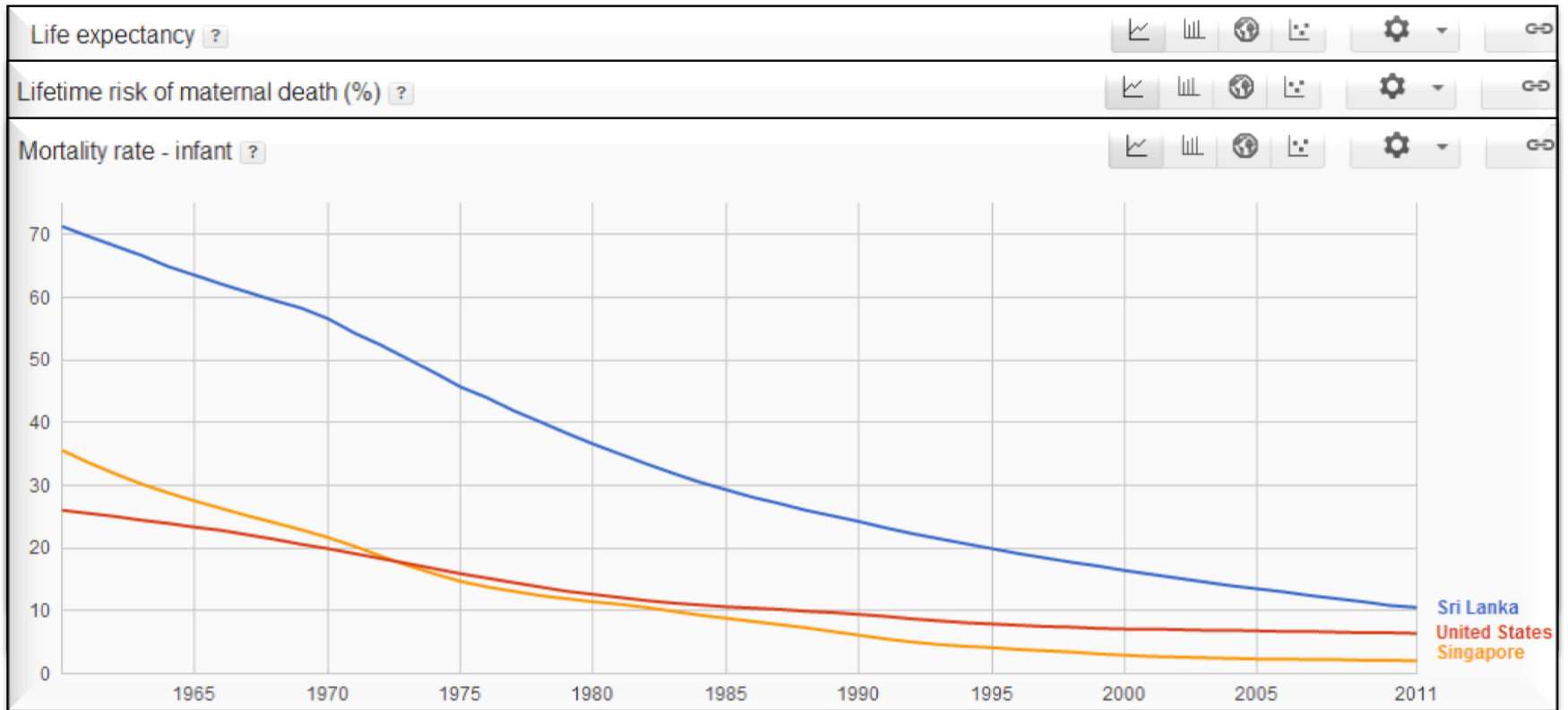
**Provide Clinical /Diagnostic Genetic Services, Provide Undergraduate and Postgraduate Training, and Conduct Research by it self and in collaboration with academic and the private sector both nationally and internationally**

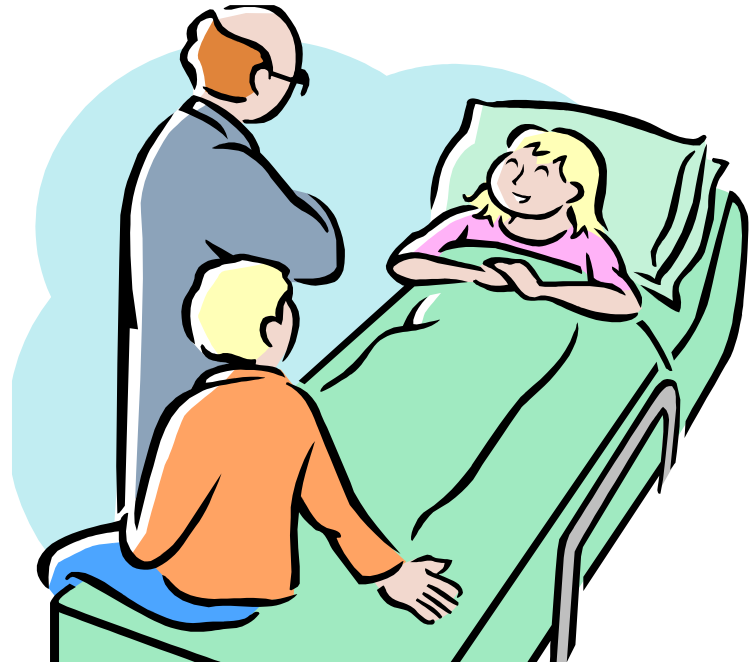
**Serving a Population of 20.1 Million People**











# UPCOMING

mark your calendars

## International Conference on Birth Defects 2014

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**Serving a Population of 20.1 Million People**

# VÍSIÓN

To help Sri Lankan families live healthier and happier lives.

# MÍSSIÓN

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

## International Networks

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

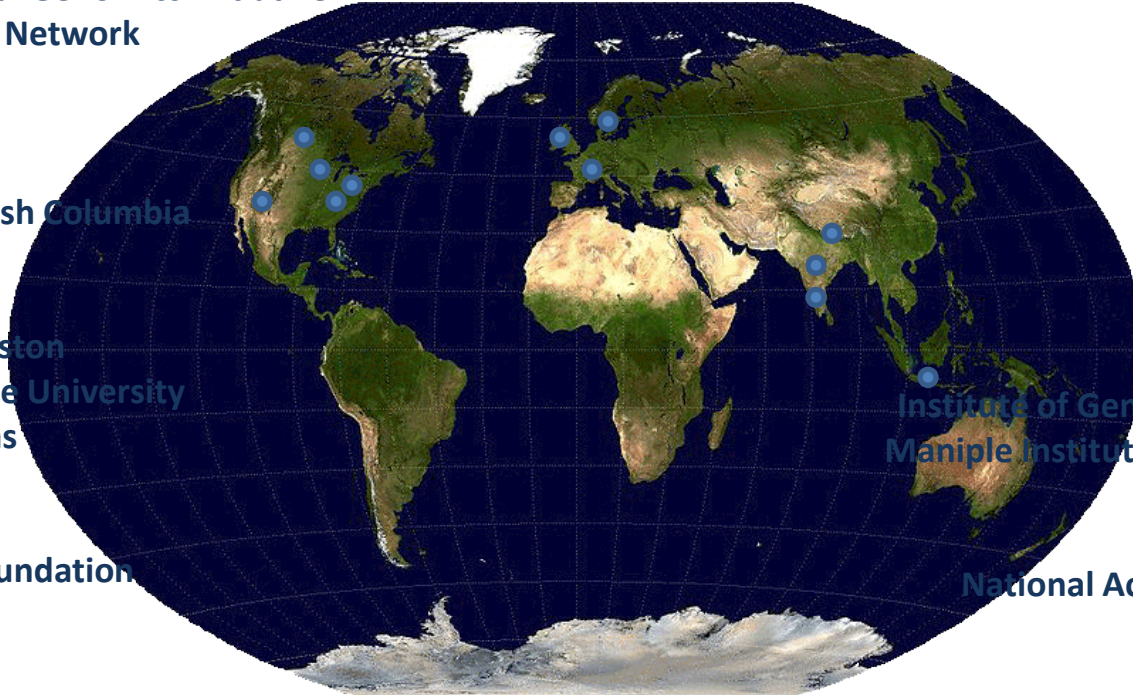
University of British Columbia

## 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

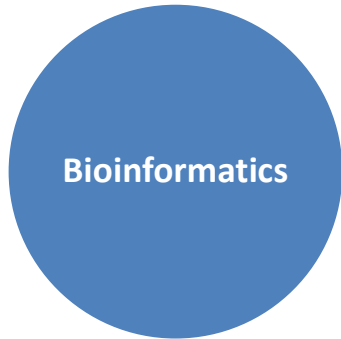
National University of Singapore

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

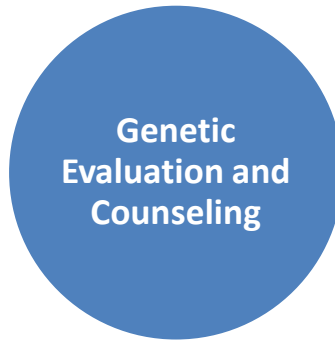


# Services Available

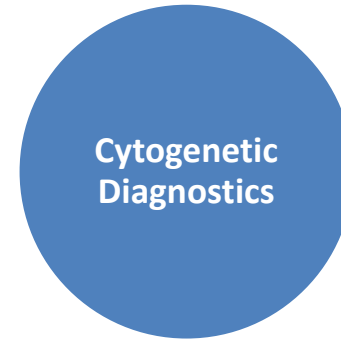
**2010 Established**



Bioinformatics



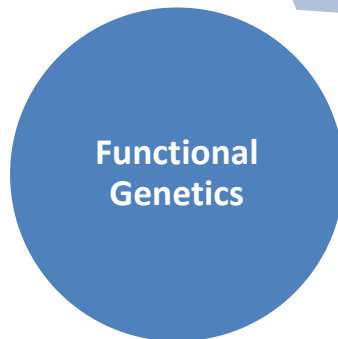
Genetic  
Evaluation and  
Counseling



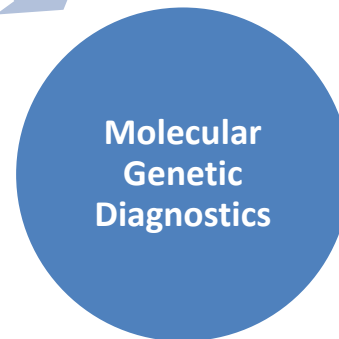
Cytogenetic  
Diagnostics



Medical  
Genetics



Functional  
Genetics



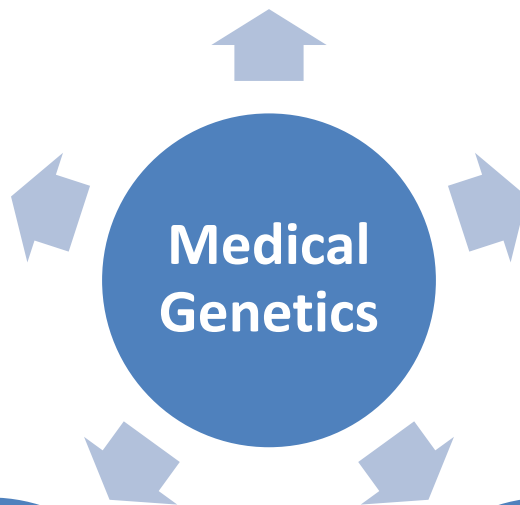
Molecular  
Genetic  
Diagnostics

**1983 Established**  
**2004 Automated**  
**Karyotyping**  
**2013 FISH**

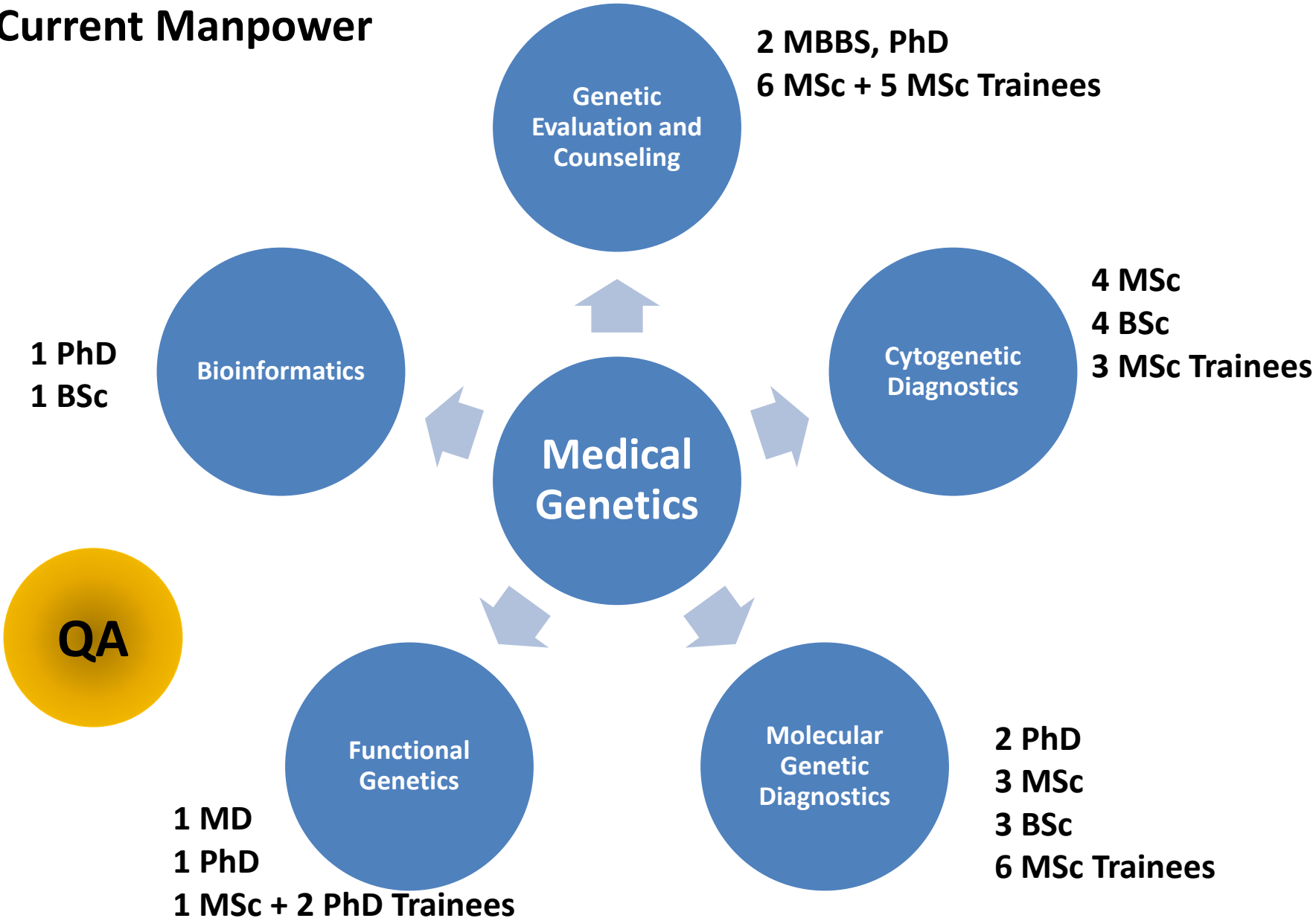
**2004 Established**

**2006 Real Time PCR**  
**2006 Sanger Sequencing**  
**2013/14 NGS**

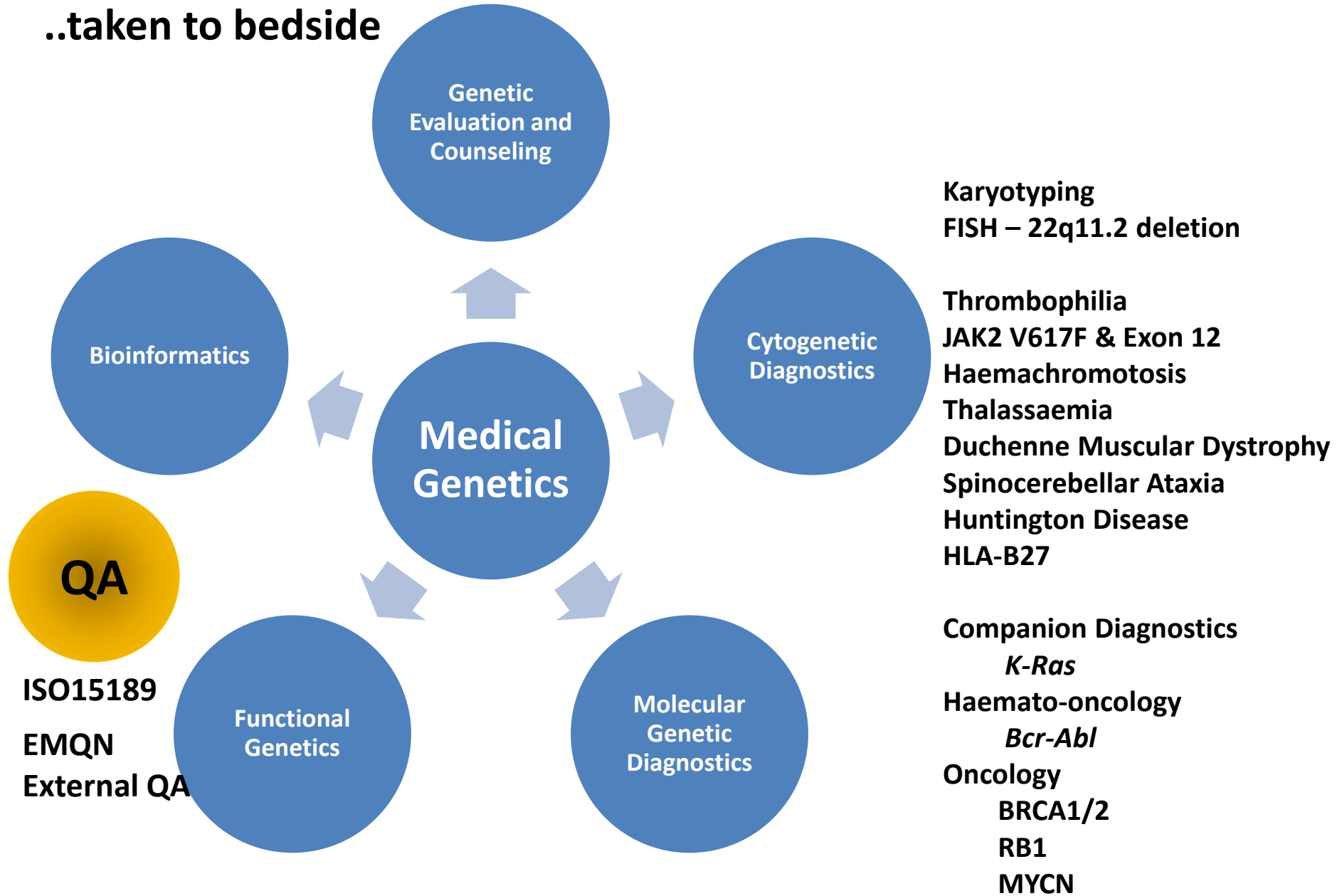
**2011 Established**



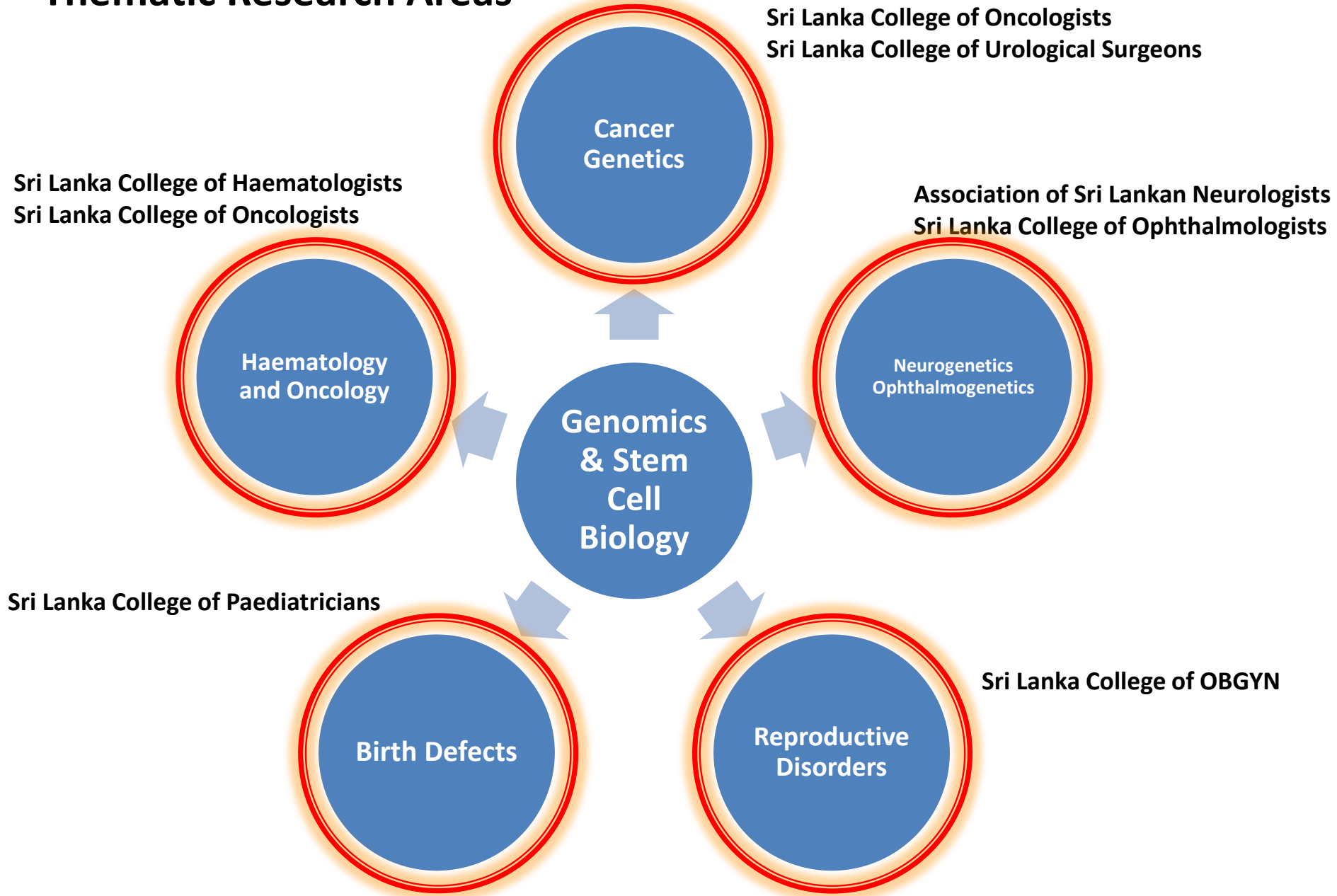
# Current Manpower



**..taken to bedside**



# Thematic Research Areas



# International Collaborations

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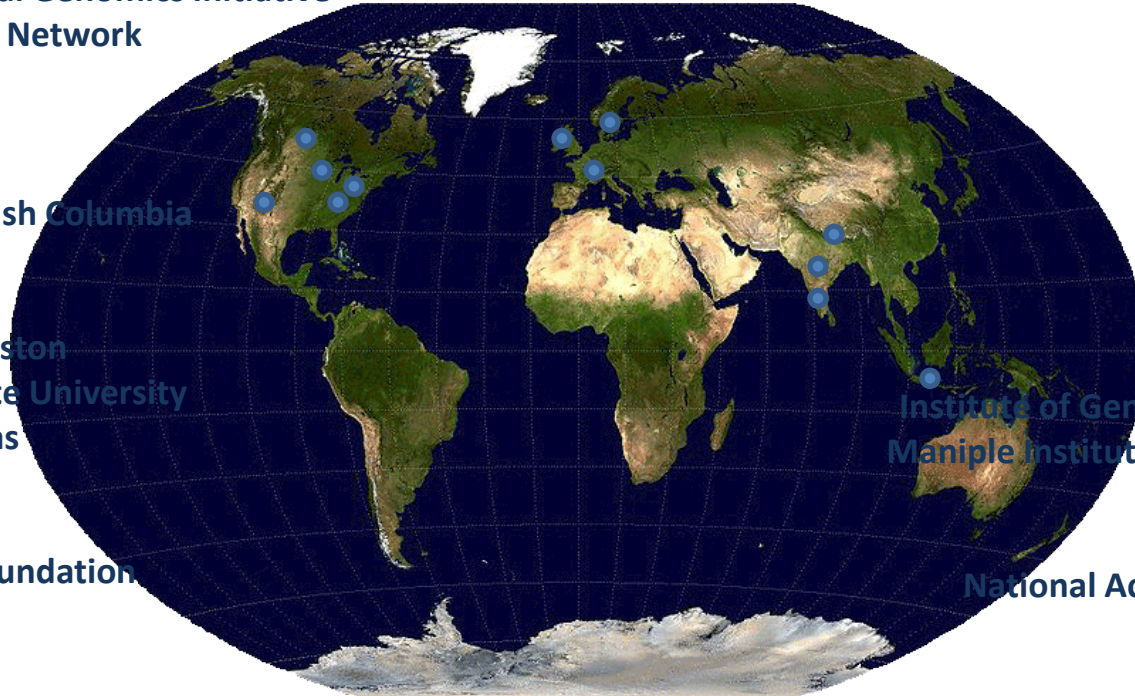
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# Translational Goals for 2014-15

## 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 iron 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**

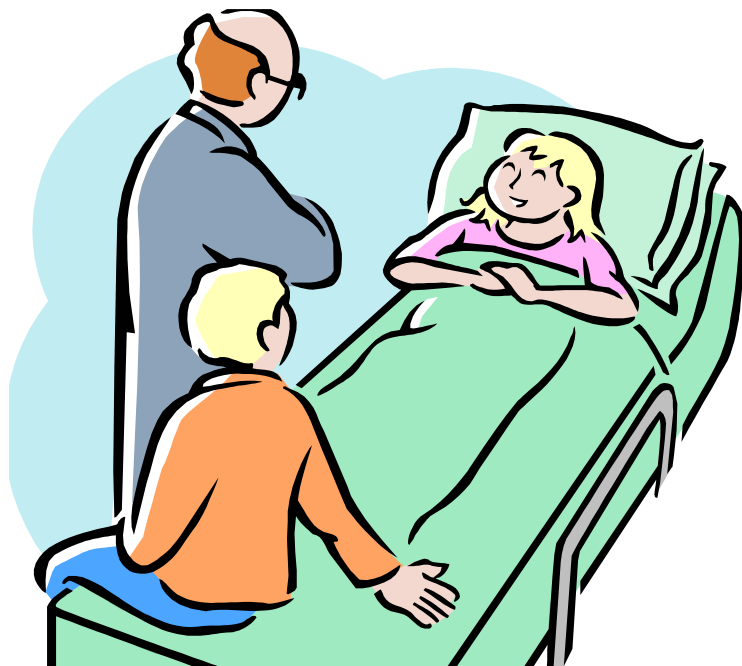
Introduce an online CME courses (Already commenced)

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