Expanding Newborn Screening Programs Globally: Current Initiatives and Future Directions

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Vision
A healthier world through quality laboratory practice.

Mission
To promote the role of public health laboratories in shaping national and global health objectives, and to promote policies, programs, and technologies which assure continuous improvement in the quality of laboratory practice and health outcomes.
APHL Newborn Screening and Genetics

- Strengthens the role of NBS PHL’s
- Facilitates implementation
- Policy statements
- Provides input to CDC’s Newborn Screening Quality Assurance Program (NSQAP) on quality control and proficiency testing issues
Infant Morbidity and Mortality

- Malaria
- Diarrhea
- Malnutrition
- Upper Respiratory Diseases
- Infectious Diseases
- Low Birth Weight
- Sickle Cell Disease
The State of Sickle Cell Disease In Africa
## Estimated prevalence of hgb gene variants and affected conceptions

<table>
<thead>
<tr>
<th>Region</th>
<th>Pop mil</th>
<th>CBR</th>
<th>BR/y mil</th>
<th>MR &lt;5y</th>
<th>% pop w sig variant</th>
<th>Sickle cell conceptions /1000</th>
<th>% &lt;5 MR affected</th>
</tr>
</thead>
<tbody>
<tr>
<td>Africa</td>
<td>586</td>
<td>39.0</td>
<td>22 895</td>
<td>168</td>
<td>18.2</td>
<td>10.68</td>
<td>6.4</td>
</tr>
<tr>
<td>America</td>
<td>853</td>
<td>19.5</td>
<td>16 609</td>
<td>27</td>
<td>3.0</td>
<td>0.49</td>
<td>2.0</td>
</tr>
<tr>
<td>World</td>
<td>6 217</td>
<td>20.7</td>
<td>128 814</td>
<td>81</td>
<td>5.2</td>
<td>2.28</td>
<td>3.4</td>
</tr>
</tbody>
</table>

CBR crude birth rate
MR mortality rate
Sickle Cell Disease

- SCD affects ~ 100 million people worldwide
  - 5% of world’s population are carriers
- Over 200,000 infants/yr are born with SCD in Africa.
  - 60% will die as infants
- Equatorial Africa has 10-40% prevalence of sickle trait
  - Highest prevalence is in West Africa
  - Ghana & Nigeria estimates are 15-40%

Ghana

Population: 23,351,000

Birth rate: 28 births/1,000 population

Annual births: 757,000

Death rate: 9 deaths/1,000

Infant mortality: 51 deaths/1,000
Ghana

• Newborn screening for SCD pilot study in the mid 90’s

• Sponsored by the Ministry of Health, various hospitals in Ghana, the Sickle Cell Disease Association of Ghana, Children’s Hospital in Philadelphia
  • Supported by a grant from NIH/NHLBI.
Ghana

- Screening laboratory is based in Kumasi
- Expansion continues and the country is moving towards a national program
- Over 300,000 infants have been screened
- ~2% of screened infants have SCD
  - Over 85% of the newborns with SCD are tracked and followed up
- Collaboration with APHL and CDC
MOU with Ghana

• Cooperative relationship among the five organizations:
  – Ghana Ministry of Health
  – Sickle Cell Foundation of Ghana
  – Komfo Anokye Teaching Hospital
  – Noguchi Memorial Institute for Medical Research
  – Association of Public Health Laboratories
  – Newborn Screening Quality Assurance Program, CDC

• Collaborative initiatives and laboratory support for sickle cell disease and other hemoglobinopathies for newborn screening.
Liberia

Population: 3,955,000
Birth rate: 38 births/1,000 population
Annual births: 145,000
Death rate: 10 deaths/1,000
Infant mortality: 100 deaths/1,000
Liberia

• ~ 4000 deaths annually from SCD infants under five according to WHO

• Previous studies in Liberia have estimated the sickle cell gene prevalence at 15%

• Pilot study proposed to determine prevalence of SCD in Monrovia through neonatal screening and develop a clinical program for education and health supervision
Liberia

• Joint collaboration between researchers at the Children’s Hospital Boston, the University of Liberia and the Liberian Ministry of Health and Social Work
• Short and long term objectives
• Other avenues for collaboration and assistance, including training and quality assurance
Nigeria

Population: 154,729,000
Birth rate: 40 births/1,000 population
Annual births: 6,028,000
Death rate: 16 deaths/1,000
Infant mortality: 96 deaths/1,000
SCD - Nigeria

- Nigeria accounts for 75% of all SCD births in Africa
- Nigeria ranks #1 among sickle cell endemic countries in Africa and worldwide.
- ~24% of Nigerians have sickle cell trait
- Prevalence of SCD in Nigeria is 20 per 1000 births
  - ~150,000 infants are born in Nigeria with SCD/yr
  - ~100,000 infants will die annually from SCD in Nigeria alone
    - Representing almost 80% of infant deaths from SCD in Africa.

(59th WHO assembly 2006)
Newborn Screening Initiative in Nigeria

• Goal: To reduce morbidity and mortality related to diseases detectable by newborn screening in Nigeria, using sickle cell disease as a model.

• Discussions started with three states in June 2009
Objectives of Initiatives

• To provide NBS for hemoglobinopathy conditions for newborns in several states in Nigeria.
• To provide prophylactic and comprehensive SCD management to newborns identified.
• To provide education and counseling to families whose children are identified with significant hemoglobinopathies through NBS.
NBS Activities in Nigeria

- Three isoelectric focusing platforms and materials were donated to three states in Nigeria (Oyo, Kaduna and Anabara) by PerkinElmer Inc.

- Training of laboratorians, follow up coordinators, nurses and genetic counselors scheduled July/August 2011
Tanzania

Population: 42,746,000
Birth rate: 32 births/1,000 population
Annual births: 1.462 million
Death rate: 12 deaths/1,000
Infant mortality: 70 deaths/1,000
Tanzania

- 8,000 to 11,000 SCD births year
- No newborn screening (yet)
- Prevalence of HbAS and HbSS is estimated at 12.1% and 0.7%, respectively.
- Muhimbili Sickle Cell Programme established in 2004
  - SCD clinic with over 2000 patients
Newborn Screening in Tanzania

• Collaboration with partners
  – Tanzania Ministry of Health and Social Welfare
  – Diagnostic Services
  – Family and Child Health
  – CDC Tanzania

• Provide the necessary equipment, materials, training to initiate a newborn screening program in Tanzania
Next Steps

• Formation of the Newborn Screening Advisory Committee of Stakeholders
• Collaborate with partners to finalize available resources for the initiative
• Coordinate training activities with laboratory staff and nurses
• Determine initiative start date
• Coordinate similar initiatives in other developing countries.
Future activities

• Collaborate and assist new newborn screening programs in developing countries
• Coordinate efforts with state newborn screening programs in the U.S., CDC and manufacturers
• Engage and collaborate with ISNS on newborn screening initiatives worldwide
• Develop twining initiatives