Newborn Screening Information Infrastructure

How to Communicate Across Programs and Share Our Knowledge

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Backdrop
Global Globin 2020 Challenge

Goals of the project fall into the following main areas:

1. To see growth in the quality and quantity of curated inputs from low- and middle-income countries participating in the project into internationally recognized genetic databases. Tackling haemoglobinopathies is an ideal entry point for these countries to develop the necessary infrastructure and expertise that can expand genetics and genomic medicine into other areas of health-service delivery.

2. To harmonize the sharing of all relevant variant data between countries in accordance with international best practice that integrates all the relevant ethical and regulatory frameworks and policies required to serve and protect patients at the same time that the necessary biotechnical systems and procedures are developed.

3. To ensure that the storage, curation and sharing of the relevant DNA variation information is sustainable in the medium and longer term by expanding and strengthening the international network of professionals, including curators, researchers, clinicians, bioinformaticians, counsellors, patient groups and health bureaucrats.

Countries: Belgium, Brunei, China, Cyprus, Egypt, France, Italy, Malaysia, Mexico, Mozambique, Nepal, The Netherlands, Nigeria, The Philippines, Portugal, South Africa, Thailand, Venezuela and Vietnam.
The ITHANET portal is an expanding resource for clinicians and researchers dealing with haemoglobinopathies.

It integrates information on news, events, publications, clinical trials and haemoglobinopathy-related organisations and experts and, most importantly, databases of variations (IthaGenes), epidemiology (IthaMaps) and HPLC data (IthaChrom).
Newborn Screening Infrastructure
How Does It Work?

Follow-up:
- Obtain test results
- Get results to family
- Repeat test(s) if needed
- Ensure diagnostic testing
NEWBORN SCREENING IS MORE THAN A TEST

It is an entire system of services to ensure timely follow-up and intervention
SYSTEM - Newborn Screening

- Physician
- Birthing Facility
- Dried Blood Spots
- Medical Home (Physician)
- Services
- Long-Term Follow-up
- Other Databases
- Follow-up Coordinator(s)
Building Information Infrastructure for Follow-up and Data Sharing

• An opportunity to use an established analytical infrastructure

• Proven to accelerate clinical validation, foster data sharing, peer comparison, and ultimately performance improvement

• “crowdsourcing” with researchers, clinicians, public health professionals
  • Research infrastructure
  • Follow-up
Going Forward

- Standardize case definitions
- Standardize screening and diagnostic protocols
- Establish a registry
  - Parents/individuals
  - Health care providers
  - Public health professionals
  - Researchers
- Standard protocol[s] for treatment
- Need for informed consent??
- Regional approach?
Resources

Newborn Screening Translational Research Network
http://nbstrn.org/
NBSTRN Tools

**VRDBS**
- The Virtual Repository of Dried Blood Spots (VRDBS) is an open-source, web-based tool that enables NBS researchers to search over 2.9 million DBS from participating states.

**LPDR**
- The Longitudinal Pediatric Data Resource (LPDR) is a secure informatics system designed to enable enhanced data collection, sharing, management and analysis for conditions identified as part of newborn screening or for conditions that may benefit from newborn screening.

**R4S**
- The Region 4 Stork tool is a web-based application developed to improve detection of true positive cases of diseases detected through newborn screening. The R4S project provides postanalytical interpretation of complex analyte profiles. It has been widely adopted into the routine practice of newborn screening laboratories worldwide.
LPDR Mission

• The majority of NBS conditions are rare and translating new discoveries into clinical practice requires prospective collection, aggregation and sharing of health information.

• To facilitate this translation the NBSTRN developed the Longitudinal Pediatric Data Resource which includes:
  – Data Sets
  – CDEs and Standardization
  – Informatics System
  – Discovery Interface
  – Data Warehouse
Benefits of NBS standardization

• Electronic result reporting
  – Much faster than mail
  – Can send results to multiple recipients at the same time
  – Automatically goes into a patient’s record

• Sharing data
  – For follow-up

• Border countries, disaster preparedness

• Enabling cross-referencing with other data sources
  – Birth certificates, hearing results, immunization registries

• Aggregating data for research and quality assurance
Process for standardizing

- Consolidate the terms that are the same
  - For example, “birth date” and “date of birth” OR “gestational age” and “weeks’ gestation”
- Mapped each term to the appropriate vocabulary standards
  - LOINC for lab tests, other clinical measurements, card variables
  - SNOMED CT for conditions
- Many NBS terms might have no matching term in the standards, so you obtain new LOINC codes for analytes, card variables, and SNOMED CT codes for condition names
- Many cycles of mapping, obtaining codes, and feedback
- Created HL7 messaging template for reporting NBS results
APHL
Resources

• NLM’s NBS website
  – LOINC NBS panel and annotated sample HL7 message available for download
  – Lists of conditions, associated analytes and their LOINC codes, as well as UCUM and SNOMED CT codes where appropriate
  

• LOINC: http://loinc.org

• UCUM: http://unitsofmeasure.org

• HL7 Standards:
  http://www.hl7.org/implement/standards/
International Journal of Neonatal Screening

an Open Access Journal by MDPI

The International Journal of Neonatal Screening (IJNS) is the official journal of the International Society for Neonatal Screening (ISNS) and the German Society for Neonatal Screening (DGNS).

IJNS aims to improve the knowledge of the natural course of target diseases for newborn screening, as well as the knowledge about methods used in newborn screening laboratories. IJNS also seeks to improve the communication between newborn screening laboratories worldwide, and to distribute technical notes and improvement of existing screening methods and assays.

Members of ISNS or DGNS will enjoy a 20% discount for full paper publication in IJNS, the other attendees will enjoy a 10% discount, please contact: ijns@mdpi.com

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