National Guidelines on Birth Defects Surveillance

Sri Lanka has achieved low levels of infant mortality. Further reduction of infant mortality necessitates a cause-specific approach. Birth defects assume a leading cause of infant mortality and several strategies are available for prevention and control of birth defects. Unavailability of quality birth defects data is a challenge in further reducing mortality and caring for the affected children.

Definition of Birth Defect (Congenital Anomaly / Congenital Abnormality) is a structural or functional anomaly of organs, systems, or parts of the body that occurs during intrauterine life, and is caused by genetic or environmental factors or both, evident before birth, at birth or later in life.

Birth defects may be either structural or functional;

Structural -Malformation, deformation, disruption and dysplasia (e.g., spina bifida, cleft lip, heart defects, club foot, Achondroplasia, Down syndrome).

Functional -Developmental disabilities (eg. some types of cerebral palsy, deafness), Metabolic diseases (e.g., phenylketonuria, galactosemia) or Hematologic diseases: (e.g., sickle cell anemia, thalassemia, glucose-6-phosphate dehydrogenase [G-6-PD] deficiency)

Surveillance of birth defects involves the ongoing, systematic collection, analysis, and interpretation of data related to birth defects, essential to the planning, implementation and evaluation of public health practice, closely integrated with the dissemination of these data to those who need to know and linked to prevention and control of such defects. The aim of birth defects surveillance is to improve health of children.

The **objectives of the surveillance** are to make available quality birth defects data, to utilize such data effectively at different levels (field, hospital, district and national levels) and to translate lessons learnt in to practice by dissemination to all stakeholders of maternal & child health and related areas.

Epidemiologic:
- Develop baseline and monitor prevalence
- Detect and investigate temporal trends and clusters
- Provide basis for ecologic and etiologic studies
- Document the distribution among groups of population (e.g.: geographic area, maternal age)

Health care and clinical:
- Refer children to specific services
- Evaluate utilization of services
- Detect changes in health practice
- Provide data for follow-up studies and other clinical research studies

Educational and social:
- Inform public about burden of congenital malformations
- Inform parents about resources and care facilities
- Provide data for studies of economic impact
- Promote education of health care providers

Planning and evaluation:
- Provide basis for service planning
- Identify research needs and facilitate epidemiologic or lab research
- Evaluate effectiveness of preventive and treatment services

Ministry of Health has introduced a birth defects surveillance mechanism, with Family Health Bureau (FHB), as the national nodal point. Figure 1 schematically depicts the national birth defects surveillance mechanism.

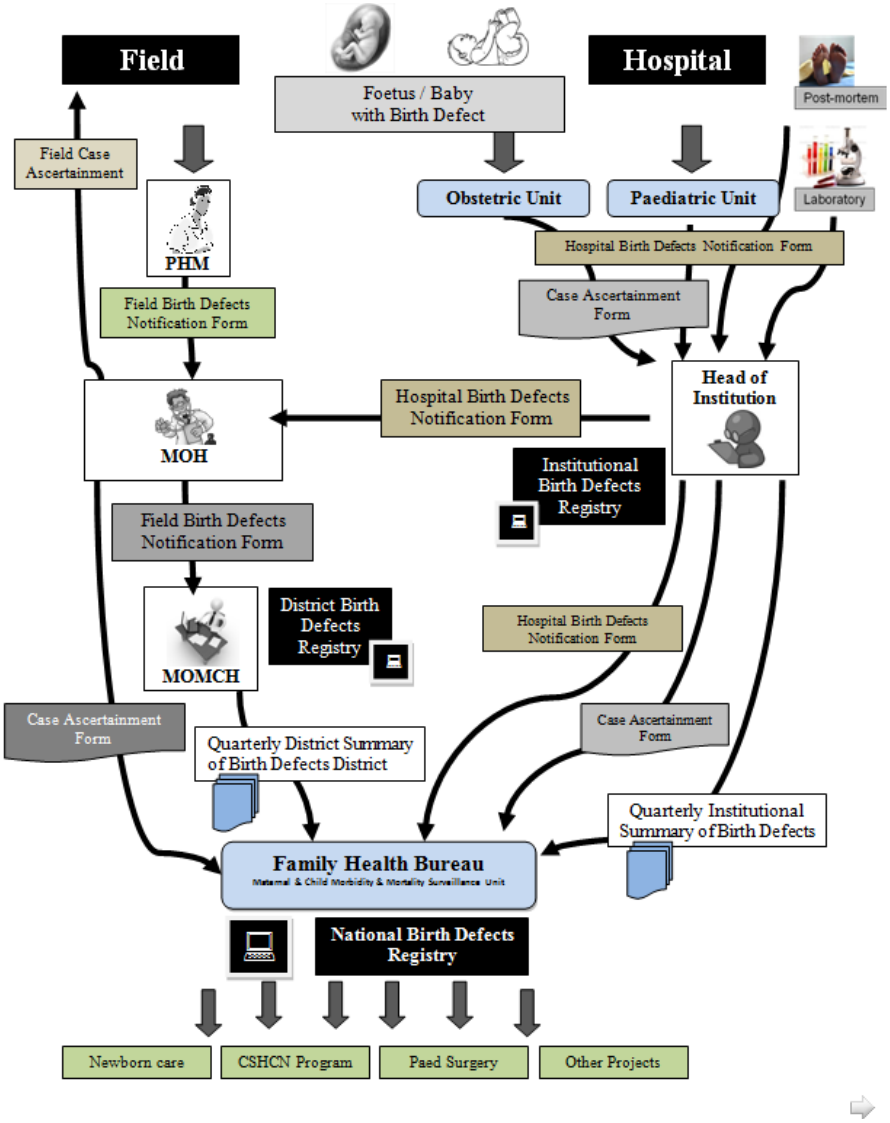

Figure 1 - National Birth Defects Surveillance Mechanism

NB: Only the hospital sector of the surveillance mechanism will be implemented from 1st of June 2017

1.0 Eligible cases – any structural or functional anomaly of organs, systems, or parts of the body that occurs during intrauterine life, and is caused by genetic or environmental factors or both, evident before birth, at birth or later in life up to 2 years.
This includes all foetal anomalies and other birth defects detected after births up to the age of two years.

Minor malformations
Minor malformation is a condition that does NOT have serious medical or social consequences, usually does not require treatment.
Minor malformations are important:

- May serve as indicator of altered morphogenesis.
- Are valuable clues for clinical geneticists, in the diagnosis of a specific pattern of congenital conditions (syndrome).

Minor malformations should be coded and included in the surveillance. However they will not be counted in the birth prevalence estimates of major birth defects in the surveillance program.

2.0 Capture points

Children with birth defects may be captured at different service delivery points in a hospital setting:

1. Out-patient Department
2. Clinics
3. Obstetric
4. Neonatology
5. Paediatric
6. Cardiology
7. Intensive care
8. Radiology
9. Surgical
10. Laboratory / Pathology
11. Forensic (post-mortem)
12. Any other relevant unit

Hospital administrators should introduce a mechanism to capture and document children with birth defects considering the practical aspects of each health facility.

3.0 Notification of birth defects (Government and private hospitals)

3.1 Within the hospital

All **birth defects** detected at both government and private hospitals should be notified to the head of the institute by the highest level clinician / medical officer of the respective unit (eg. obstetric / paediatric / radiology / surgical / laboratory / post-mortem) using the Birth Defects Notification Form (BD-1) within 24 hours. A copy of the BD-1 should be kept at the respective unit for future reference.

Sister/Nurse in Charge of the unit where the birth defect detected should ensure that the format is completed by the relevant officers and sent to the head of the institute under the guidance of the consultant in charge.

3.2 From hospital to national level

Upon receipt of a Birth Defects Notification Form (BD-1), the head of the institution should sign, date stamp and forward the all collected notification forms at the end of each month to the Director – MCH (Family Health Bureau).

Family Health Bureau will assign a birth defect identification number to the case notified and send an acknowledgement letter to the head of the institute. The birth defect identification number should be noted and quoted in all the correspondence.

4.0 Birth defects case ascertainment

The head of the institute, should identify the focal clinician who will be responsible for the overall management of the case and facilitate obtaining all the clinical details and relevant documents to the above clinician.

The highest level clinician (eg. Obstetrician / Neonatologist / Paediatrician / Surgeon / Radiologist / Forensic Pathologist / Pathologist etc) should complete the Birth Defects Case Abstraction Form (BD-2) with the latest available data from all sources within 14 days of detecting the case. The completed BD-2 should be forwarded to the head of the institute.

Copies of BD-2 of the index case should be kept with the clinician and with the head of the institute to update data in the future course of management or in the event of death.

4.1 From hospital to national level

Upon receipt of a Birth Defects Case Abstraction Form (BD-2), the head of the institution should sign, date stamp and forward the all collected Case Abstraction forms at the end of each month to the Director – MCH (Family Health Bureau).

Sister/Nurse in Charge of the unit where the birth defect detected should ensure that case abstraction formats are completed by the relevant officers and sent to the head of the institute under the guidance of the consultant in charge.

4.2 Foetal Anomalies

The obstetric unit should maintain data related to birth defect during the course of pregnancy and share the Birth Defects Case Abstraction Form (BD-2) and other details with the paediatrician / neonatologist after a live birth.

4.3 Foetal / Infant deaths

A post mortem or pathological post mortem should be conducted wherever possible in cases of foetal and infant deaths when a definitive cause of death is not determined. Relevant officials should proceed for a pathological post mortem with a probable cause of death.

The forensic pathologist or pathologist is responsible for providing details for filling Post-Mortem / Pathological Examination Findings and Cause/s of Death in the BD-2. Other details should be filled by the relevant clinician.

Upon receipt of a completed Birth Defects Case Abstraction Form (BD-2), the head of the institution should sign, day stamp and send the form within 24 hours to the Director – MCH (Family Health Bureau).

5.0 Custody of documents related to the birth defect and management

Once a case (pregnant mother / infant) of birth defect is reported in an institution (government or private hospital), the head of the institution should maintain a register with basic detail along with the assigned birth defect identification number. A file with copies of the Birth Defects Notification Form (BD-1), clinical notes, investigations and all other relevant documentation related to the mother and/or infant should be maintained at the hospital.

6.0 Coordination of birth defects surveillance

The institutional birth defects surveillance activities should be coordinated by the Medical Officer – Public Health (MO – PH) or a medical officer designated by the head of the institution.

7.0 Web-based National Birth Defects Information Management System (NBDIMS)

A web-based birth defects information management system is implemented in parallel. All the important variables of birth defects information should be entered in this system.
Web link: ***http://124.43.17.168:19667/nbds***
Director – Maternal and Child Health and National Program Manager on Child Morbidity & Mortality Surveillance act as database custodians.
At hospital level, Medical Officer – Public Health (MO – PH) or a medical officer designated by the head of the institution should enter and maintain data in the on-line system. Necessary guidelines, passwords and system administration will be communicated to head of the hospital.

8.0 Coding

ICD – 10 (International Statistical Classification of Diseases and Related Health Problems Tenth Revision) will be used for coding identified birth defects. ICD – 10 is available on: http://apps.who.int/classifications/icd10/browse/2010/en. Chapter XVII includes (Q00-Q99): malformations, deformations and chromosomal anomalies. Coding extensions developed by Royal College of Paediatrics & Child Health – RCPCH will also be used for greater specificity.

For further clarification of birth defects surveillance, you may contact Maternal & Child Morbidity & Mortality Surveillance Unit of Family Health Bureau.