

# Birth Defects, the Challenges Ahead for Malaysia

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Birth defects are structural abnormalities which develop in foetuses during their intrauterine life. Depending on the underlying cause, birth defects may affect one or more organ systems and sometimes present as recognizable syndromes. According to large series studies, defects of the heart (affecting about 1 in every 100 to 200 livebirths), neural tube (affecting 1 of every 1000 pregnancies), lip and palate (affecting 1 in 700 to 1000 livebirths), and male genitalia, and Down Syndrome are by far the most common<sup>1-4</sup>.

Because of the social, economic, psychological and educational cost associated with birth defects, knowing their incidence in a community and the associated risk factors will greatly help a country in carrying out preventive measures and cost-effective management of this group of medical conditions. Although most major birth defects can be detected during antenatal period in pregnant women by imaging techniques and/or blood testing, some are detectable only after birth. An efficient, reliable and cost-effective system of screening services during both the antenatal and postnatal period is, therefore, of paramount importance to help determine the actual incidence of various birth defects in a particular community. Such a system encompasses not only competently trained personnel, adequate number of equipment for screening purpose, and a clear line of referral pathway for suspicious cases, but also online linkage of findings to provide reliable national data for preventive measures and management without duplication of workload.

In Malaysia, a national system of screening and documentation for birth defects during both the antenatal and postnatal period is not yet in place. Most of the reported data on antenatal and postnatal screening are government hospital-based, using a descriptive study model<sup>2,7</sup>. Because of manpower and financial constraints, the extent and types of antenatal

screening is variable and carried out mainly on high risk pregnancies in most government hospitals. Postnatally, screening examination of the newborn is usually carried out by the most junior doctors, the freshly graduated house officers. The latter's exposure to neonatal screening examination during the undergraduate period is at best a two-week posting during their five year undergraduate medical course. Cyanotic heart diseases and renal abnormalities may be missed without ultrasonography during the asymptomatic neonatal and infancy period. Furthermore, the definition of birth defects is not uniform. For instance, in the survey of birth defects in a tertiary center reported by Noraihan et al in this Journal<sup>5</sup>, glucose-6-phosphate dehydrogenase deficiency and thalassemia were included as birth defects. This variable definition makes comparison of incidence of birth defects reported in other centers and countries difficult.

Studies in human and animals show that environmental pollution is significantly associated with birth defects<sup>8-10</sup>. With increasing industrialization and the frequently recurrent haze problems in Malaysia, it is not certain whether there is an increase in the incidence of birth defects and whether there is any significant association between environmental pollution and birth defects in our country. An accurate data on the incidence of birth defects in the different parts of the Malaysia in relation to factories sites may provide some clues on this.

According to the annual mortality data collected by the Malaysian Statistics Department in recent years,<sup>11</sup> birth defects are among the top three common causes of perinatal death. This suggests that this group of conditions has a significant impact on the distribution of limited health resources in the care of sick infants and children. There is, therefore, an urgent need to collect reliable national data to help identify obvious

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preventable risk factors so that both short-term and long term national plans to minimize this group of conditions can be instituted. Setting up a Malaysian birth defect registry is the obvious first step forward.

Taking into consideration the limited financial and manpower resources, and learning from the experience gathered by other illness registries in this country, the types of condition to be included in the birth defect registry during the initial phase should not be too many. It is better to have good quality data collected on a few types of conditions than to include all types of birth defects without clearly defined and robust definitions, with resultant large masses of unreliable and unusable data. One good model of national medical registry is the Malaysian Cancer Registry<sup>12</sup> which has only seven items for medical and health practitioners to report. Despite the small number of items collected, the data collected provide very useful and reliable information on cancers in Malaysia which help national planners and health providers in cancer prevention and research.

Based on studies conducted in various Malaysian hospitals, one of the most common types of severe birth defects are those affecting the neural tubes<sup>5,7</sup>. Gross neural tube defects such as anencephaly and meningomyeloceles can be readily detected either antenatally by ultrasonography and/or postnatally by physical examination. Furthermore, current evidence showed that folic acid supplement plays a significant role in reduction of its incidence in some countries<sup>13</sup>. For a start, perhaps the Malaysian Birth Defects Registry

should focus on data related to the neural tube defects and its associated potential risk factors in this country.

As obstetricians, paediatricians, pathologists and maternal child health personnel are involved in care and/or diagnosis of patients with birth defects, all four groups working in both government and private healthcare institutions should be represented in the committee on the National Birth Defect Registry when it is set up. Besides designing a user-friendly data collection proforma with clearly defined diagnostic criteria, and setting up of an on-line database entry system, education campaign should be carried out to help health providers in fine-tuning their antenatal diagnosis through ultrasonography and postnatal screening examination of this group of conditions. In addition public education is important to create awareness of this registry. This will ensure that as many cases are detected and reported as possible. Various maternal risk factors (such as folic ingestion, smoking, alcohol consumption, illegal drug(s) use, maternal diabetes, and herbal ingestion during pregnancy, relationship with industry and occupations, and infection) should be included to help identify potential risk factors for future research with the aim of prevention and treatment.

As this is a national project and its findings have huge impact on the wellbeing of the population, the main source of funding for initiation of the Malaysian Birth Defects Registry should be from the Ministry of Science, Technology and Innovation (MOSTI).

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