

World birth defects day

Towards a national registry for birth defects in Saudi Arabia

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In 2010, the World Health Organization (WHO) defined congenital anomalies or birth defects (BDs) as “structural or functional anomalies, including metabolic disorders, which are present at the time of birth”.¹ This definition includes structural malformations detected antenatally or discovered after birth by routine screening tests, and inherited enzymatic deficiency disorders detected later in life. The epidemiological implication is that the current prevalence of BDs is underestimated, and there are wide regional and ethnic variations. Birth defects lead to an even greater financial burden, unanticipated psychological trauma, family dynamics disruptions, and social tension. In the 2014 update,² the WHO indicated that BDs affect one in 33 newborn infants, leading to an estimated 3.2 million BD-related disabilities, and an estimated 270,000 deaths during the first 4 weeks of life. In industrialized and developed countries, BDs contribute significantly to infant mortality rates, and are the leading cause of perinatal mortality in some countries.³ The cause of BDs can be environmental, genetic, infectious, or multifactorial. Most often it is difficult to identify the precise cause(s) of specific defects, although many BDs are preventable. Accordingly, the WHO regional office for South-East Asia with 11 organizations declared the 3rd of March, 2015 as the “World Birth Defects Day” to grab the attention of healthcare providers and the public at large. The aims are: to raise awareness of birth defects, to develop and implement primary prevention programs, and to expand referral and care services.

A “Birth defect national registry” will help achieve these aims by disseminating surveillance data collected by local investigators to the public and to healthcare providers and stakeholders for initiation of preventive

plans, and also help in monitoring trends and clusters of birth defects.

The global picture of birth defects. With a reduction in the rate of neonatal infections and intrapartum complications in the industrialized countries, BDs have contributed significantly to perinatal mortality and to the “under 5 mortality,” and to morbidity. In Finland, for example, the most common cause of mortality in infants under one-year of age is BDs.⁴ The WHO reported the causes of death in 3.08 million infants under one-year of age from 193 countries. In 9% of all deaths, BDs were the cause.² Over the past 3 decades, several national and international BD registries have been initiated; the European countries registry (www.eurocat-network.eu), the United States Centers for Disease Control and Prevention (www.cdc.gov), the International Clearinghouse for Birth Defects Surveillance and Research (www.icbdsr.org), the Latin-American Collaborative Study of Congenital Malformations, the Victorian Birth Defects Register in Australia, and the Japan Association of Obstetricians and Gynecologists program. In addition, several national and regional registries have been initiated in many parts of the world. These institutions are engaged in data collection, trends, and cluster detection, data analysis, dissemination of information, and holding regular meetings and conferences on various aspects of BDs from causation to impact and to prevention. They also collaborate with the WHO on disease classification and the formulation of new initiatives and policies. In 2013 and 2014, the national birth defects prevention network in USA used the slogan “BDs are common, costly and critical” during their national birth defects prevention month (www.nbdpn.org/bdpm2014.php).

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This year's campaign (January 2015) used the theme "Making Healthy Choices to Prevent Birth Defects - Make a PACT for Prevention". The P - in PACT stands for plan ahead, A - for avoid harmful substances, C - for choose a healthy life style, and T - for talking to your healthcare provider. On the websites of these registries and institutions there is a wealth of information on how to implement these steps, and information on BDs and their prevention in general.

The current status of birth defects in KSA. Most studies on BDs from Saudi Arabia are retrospective in nature. They focused only on structural malformations and reported prevalences per 1000 live births (LB) not total births. They did not include still births (SB) with malformations, elective terminations of pregnancy for fetal anomalies (ETOPFA), metabolic or endocrine disorders, and inherited blood diseases. In a recent prospective hospital-based study in the Riyadh region of Saudi Arabia, Majeed-Saidan et al⁵ reported a prevalence of 41.1/1000 total births (LB + SB with BDs + ETOPFA). This indicates that one in 24 births were affected by one form of a major BD. This prevalence may also be an underestimate, as it did not include inherited blood disorders. The March of Dimes, in its global report on BDs, reported a prevalence of 81.3/1000 births (one in 12 born is affected by a BD) from the Kingdom of Saudi Arabia (KSA).⁶ Given an annual delivery rate of 600,000 in Saudi Arabia, this would indicate that around 48,000 babies are born each year with a major BD. In addition to the cost and disability, BDs are one of the leading causes of death in infancy. In recent studies,^{7,8} lethal malformations were the direct cause of deaths in up to 44% of babies admitted to 2 neonatal intensive care units in the KSA, and in Oman. The spectrum of the various BDs encountered in Saudi Arabia from a recent prospective cohort study includes; cardiovascular system (22.5%, 14.8/1000 births), renal system (20.3%, 11.3/1000 births), CNS (13.6%, 5.6/1000 births), genital system (10.7%, 4.4/1000 births), limb malformations (8.4%, 3.4/1000 births), chromosomal aberrations (6.6%, 2.7/1000 births), gastro-intestinal tract (6.3%, 2.5/1000 births),

oro-facial cleft (5.5%, 1.8/1000 births), musculo-skeletal (4.4%, 1.4/1000 births), genetic syndromes and microdeletions (5.0%, 2.0/1000 births), and others (7.0%, 4.2/1000 births).⁹

The most common risk factors for BDs in a case-control study nested within the above-mentioned prospective cohort study are: consanguinity ($p<0.000$); advanced maternal age ($p<0.01$), poor periconceptional intake of folic acid (FA) supplements ($p<0.000$), male gender ($p<0.000$), and overt maternal diabetes mellitus ($p=0.05$).⁵ Other factors may have contributed to the pathogenesis of BDs, and in over 50% of such defects, no cause could be determined.

What is needed in Saudi Arabia and other developing countries? The creation of a national BDs registry where national data on BDs can be collected, analyzed, and then reported to healthcare policy makers is imperative; plans can be formulated, trends and clusters of BDs can be detected, their causes identified and dealt with. It is important to maintain a dialogue with the religious scholars to widen the scope of ETOPFA, especially with the introduction of the promising molecular testing on fetal DNA in maternal blood early in pregnancy to diagnose various inherited disorders and chromosomal aberrations. Premarital counseling, especially in families with known inherited disorders, is important in addition to preimplantation genetic diagnosis in families with a previously affected child. Mandatory staple food fortification with FA should include all the flours on the market (locally-produced and imported), wheat products, cereals, and rice. The recent Saudi Medical Journal supplement on neural tube defects (NTDs)¹⁰ clearly shows that the prevalence of NTDs is still unacceptably high despite flour fortification, which was started in 2001. Mandatory fortification of all staple foods will lead to a reduction in NTDs and some other major BDs as previously reported.¹¹ It is vital to start health education programs in schools and colleges targeting women of childbearing age with regards to proper FA supplement intake, the importance of planned pregnancies, and the avoidance of harmful substances or drugs during pregnancy, especially the first trimester.

Currently, the web-based BDs registry related to the Medical Services Directorate of the Saudi Military, inaugurated in January 2013, is operational, and collects data from 16 major Saudi military hospitals. In addition, there are several other programs that are either hospital-based or malformation-specific that collect data on various BDs. These programs need to be integrated

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into one national registry or into 4-5 regional registries to obtain a comprehensive national dataset and a clearer picture of the current prevalence of BDs in accordance with the latest WHO case definitions.

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