Epidemiology of Birth Defects in Women's Health University Center Assiut –Egypt: An Observational Cross-Sectional Study

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Abstract: The objective of this study was to determine the descriptive epidemiology of birth defects in Women's Health University Center – as a unique tertiary hospital in upper Egypt to estimate the real impact of this problem on Assiut governorate. The design was observational cross- sectional study. The study was performed at the antenatal clinic of obstetrics and the labour room from September 15th 2009,On 8696 cases, including 123 babies with congenital malformations, making a frequency of 14/1000 deliveries. No previous chromosomal study was performed to any women scheduled for this study. The commonest congenital malformations were neural tube defects especially hydrocephalus (55.3% of cases) the second commonest was multifoetal abnormalities (44.7 % of cases). Antenatal diagnosis of these anomalies was made by antenatal ultrasonography in 84.4% of cases while 16.6% cases were diagnosed postnatally. In the light of the previous data we concluded our recommendations to overcome the birth defects problem in upper Egypt by trying more advance in basic reproductive health care services, educating couples about avoidable risks of such defects and discouraging reproduction after the age of 35 and introducing more advance and availability of genetic services.

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Keyword: Birth defects – Antenatal care.

1. Introduction

More than 4 million children are born with birth defects each year all over the world[(11)]. There is a little doubt that birth defects cause enormous harm in settings where risk factors for many conditions are raised and health care resources are limited. Yet today there is an unprecedented opportunity to prevent many birth defects and reduce the consequences of those that occur, and to do so at reasonable cost[(22)]. For example congenital rubella syndrome can be prevented through the immunization of children and women, and also the impaired mental development secondary to iodine deficiency during pregnancy can be, prevented by the administration of the low cost iodinized table salt[(18)].

Birth defects account for a significant proportion of morbidity and mortality among infants and children, particularly in areas where infant mortality due to more common causes has been reduced[(15)].

The prevalence of specific birth defects varies widely in different populations. In countries where basic public health services are not available, the prevalence of serious birth defects is generally higher than in developed countries[(21)].

Genetic screening can detect risk factors associated with birth defects before conception as well as prenatally. For confirmed severe birth defects, legal termination of pregnancy can be offered after nondirective counseling to support each women in the decision that is appropriate for her[(14)]. Several developing countries are making significant progress toward prevention and care of birth defects and reducing infant mortality[(15)]. Birth defects could be classified into[(4)]:

I- Genetic birth defects:

A) Chromosomal disorders like trisomy 21 (Down Syndrome).
B) Single-gene disorders which are classified by mode of inheritance as autosomal recessive or dominant as thalassemias or as X=linked recessive or dominant as glucose-6-phosphate dehydrogenase deficiency (G6PD).

II- Birth defects of environmental origin:

Exposure to a teratogen during embryonic or foetal life can cause functional disorders and malformations. Teratogens could include either infectious agents as *toxoplasma gondii*, medications, or maternal illness as iodine deficiency disorders.

III- Birth defects of complex and unknown origin:

As neural tube defects, congenital heart disease, cleft lip and/or palate, or talipes or club foot.

The process of reducing the impact of birth defects in developing countries can be undertaken in three stages[(7)].

1- Introduction of low-cost preventive interventions.
2- Provision of improved treatment for children and adults with birth defects.
3- Introduction of screening programs to identify genetic birth defects that can be prevented or treated.

Discouraging pregnancy in women over 35, folic acid fortification, salt iodinization, routine

immunization against rubella, detailed foetal ultrasonography and neonatal screening programs are examples of the above stages[9].

However, even relatively simple efforts to monitor the birth prevalence of common birth defects along with associated death and disability, can highlight priority areas and changes in those areas over time. This information is a key to identify priority interventions and their success over time[20].

2. Patients and Methods

This observational cross-sectional study was conducted between September 15th 2009 and September 15th 2010 at the antenatal clinic of obstetric and labour room of Women's Health Center, Assiut University, Assiut, Egypt, and comprised 8696 cases including 123 babies with congenital malformation making a frequency of 14/1000 deliveries.

Inclusion criteria include:

1- All pregnant women attending the antenatal care clinic with gestational age between 16-36 weeks gestation.
2- All pregnant women who were admitted to the labour room to have either vaginal or caesarean delivery.

Patients were identified antenatally using (Medison 128 B®) ultrasound apparatus with a curved abdominal probe of 3.5 MHz frequency or postnatally by detailed clinical examination for all newborn babies (either living or stillbirth) in collaboration with paediatric team. A detailed history and basic investigation for cases highly suspicious of TORCH infection (Toxoplasma, rubella, cytomegalovirus and herpes simplex) and diabetes mellitus. The points elaborated during history taking were age, parity, infection or fever with rash in the early pregnancy, recurrent abortions, consanguineous marriage, family history of abnormal babies, or exposure to drugs like warfarin phenytoin or streptomycin … etc during first trimester.

Table (1): Complete obstetrical ultrasound report:

Patient demographic data	- Name. - indication for consultation. - First day of last normal menstrual period. - Examination date. - Name of interpreting sonographist.
Foetal biometry (in millimeter)	- Biparietal diameter. - Head circumference. - Abdominal circumference. - Femur length. - Acknowledged.
Foetal Anatomy	- Cranium. - Cerebral ventricles. - Posterior fossa. - Face & lips. - Spine. - Diaphragm. - Four-Chamber heart. - Heart axis and cardiac outflow tracts. - Stomach, kidneys and urinary bladder. - Abdominal cord insertion and cord vessels. - Arms, hands, legs and feet. - Genitals.
Amniotic fluid	- Index and clarity.
Placenta	- Position as well as relationship to the cervical internal os and (if applicable) to a uterine scar.
Maternal anatomy	- Uterus, cervix, kidneys

Table (2): Newborn physical examination:

Skin	Head	Neck
- Macerated. - Intact. - Blistered. - Rash present. - Meconium staining. - Other (describe).	- Large head - Small head. - Encephalocele. - Others (describe).	- Webbing. - Masses or lump. - Others (describe).
Body	**Eyes**	**Back**
- Body wall defect. - Abdominal distention. - Abnormal umbilical cord. - Hernias. - Abdominal masses. - Others (describe).	- Lids fused. - Lids open. - Size and any abnormality. - Others (describe)	- Symmetrical. - Spina bifida. - Patient anus. - Other (describe).
Nose	**Genitalia**	**Ears**
- Nostrils patent. - Nostrils not patent. - Others (describe).	- Normal male. - Normal female. - Other (describe)	- Normally situated. - Abnormal form. - Other (describe).
Limbs		
- Absent limbs. - Hypertrophied. - Curving limbs. - Fractures. - Abnormal digit shape. - Other (describe).		

3. Results

During the study period 8696 cases were conducted including 123 babies with congenital malformations making a frequency of 14/1000 deliveries. Table (3) showed the risk factors among the studied population.

Risk factor	No.	%
1- Maternal age > 30 y.	55	44.7
2- Consanguineous marriage.	78	63.4
3- Past history of abortion.	11	8.9
4- Family history of congenital anomalies	22	17.9
5- Drug intake (anti-epileptics ,Warfarin)	3	2.4
6- Fever in 1st trimester of pregnancy.	2	1.6
7- Diabetes mellitus.	13	10.6
8- Smoking (husband)	61	49.6
9- Body mass index ≥ 30	35	28.5
10- Paternal (husband)	43	34.95

Patients who had not scheduled antenatal care at their primary health care centers were (84%) of cases and were coming to woman's health center for an isolated antenatal visit for different obstetric complains. Among them 24 (19.5%) were primiparous, 45 (36.5%) were multiparous and 54 (43.9%) were grandmuliparous (> 5 parity). No patient had any genetic testing. Majority of patients (76.4%) had vaginal delivery while 23.6% of cases had caesarean section for malpresentation or hydrocephalus. There were (58) live births (43%), (59) still births (47.9%) and (11) novonatal deaths (8.9%). The commonest congenital malformations were neural tube defects seen in 55.3% of cases; among them, hydrocephalus was predominant (32 cases) as shown in the next table(4):

Anomaly	No.	%
Neural tube defects (Mainly hydrocephalus 32 cases approx. 26%).	68	55.3
Abdominal wall defects.	4	3.24
Gastrointestinal anomalies.	7	5.7
Cardiovascular anomalies.	21	17.08
Genitourinary anomalies.	11	8.92
Skeletal dysplasia	2	1.62
Others	10	8.14

4. Discussion

One child, out of 55 born has major structural abnormality. Such abnormalities can contribute up to 15% of perinatal deaths. Genetic disorders and congenital abnormalities can occur in about 2.5% of all the live births, and account for up to 30% of paediatric

hospital admissions and about 50% of childhood deaths in industrialized countries[22]. In our study, the prevalence of congenital abnormalities was 14/1000 deliveries or 1.4%.

One of the important predisposing factor associated with congenital abnormalities is the consanguineous marriage. The consanguinity, which is defined as the marriage between relatives, has received a great deal of attention as a potential risk factor for many adverse health outcomes. Consanguineous marriage has declined remarkably in many parts of the western world, but it is still very common in the middle east, especially among Arab communities where it is believed to preserve family ties[2].

Consanguineous marriages was found in our study to be around 64%. Children of consanguineous parents may be over represented in patients with severe mental retardation, blindness, hearing impairments and deaf-mutism [8].

Increasing maternal age is associated a high incidence of congenital malformation. The overall reported incidence of congenital anomalies increases significantly over 40 years of age. Croen and Shaw[6] found that there was a strong association between advanced maternal age and chromosomal defects. However Leck[12], found that the rates for most congenital anomalies were higher in teenager pregnancies than in adult ones. In our study 45% of cases were above 30years of age and 10% were below 18 years old.

Offspring of women with DM are at increased risk for congenital malformations mainly attributable to the poor periconceptional glycaemic control[13]. The maternal DM per se, through the adverse effects on maternal metabolism is the responsible factor for the increased incidence of congenital malformation in the offspring[5].13 cases (about 10.6%) of our patient has history of D.M.

Perinatal infections that may be teratogenic include herpes simplex, viral hepatitis, mumps ,rubella, varicella and toxoplasmosis and account for 2-3% of all congenital anomalies. TORCH, which includes toxoplasmosis, other (syphilis, varicella – Zoster, parvovirus), Rubella, cytomegalovirus and Herpes infection, are some of the most common infections associated with congenital anomalies. Most of TORCH infections cause mild maternal morbidity, but have serious foetal consequences, and treatment frequently has no impact on foetal outcome[17]. In our study 2 patients reported high- grade fever in 1st trimester with positive serology for CMV and toxoplasma (1.62%).

Maternal exposure to drugs or environmental chemicals may be responsible for 46% of congenital anomalies or approximately 1 in 400 liveborn infants. Maternal use of antiepileptic drugs during pregnancy has been associated with an increased risk of major congenital anomalies in foetus. Yet in many women planning for pregnancy, antiepileptic drugs cannot be discontinued to avoid seizures during pregnancy[3]. In the current study 3 women were on antiepileptics or warfarin during their pregnancy (2.4%).

There is no clear accepted definition of advanced paternal age. A frequently used criterion is any male, age ≥40 years old at the time of conception, as opposed to the current population mean paternal age of 27 years[1]. Previous studies have suggested that some groups of birth defects may be associated with advanced paternal age. Moreover, some studies have suggested that the risk of genetic defects, specifically, sporadic dominant single – gene mutations, is 4-5 times greater for fathers aged 45 and above than for those 20-25 years old. [19]. In our study, above 35% of husbands were above 50 years old.

Katherine *et al.*[10] conducted a systemic review and meta-analysis study through which investigated the effect of greater than recommended maternal weight, on congenital anomaly risk. They demonstrated that obese mothers were at increased adds of pregnancies affected by neural tube defects, spina bifida, cardiovascular anomalies, septal anomalies, cleft lip and or palate, anorectal atresia and limb reduction anomalies. In our study, more than 25% have body mass indices ≥ 30.

In our study, passive maternal smoking due to husband smoking was found to be the second risk factor as it was found in more than 50% of patients. This was consistent with the study of Scott [16] who concluded that, the offspring of smokers and passive smokers had a 56% increase in the frequency of cardiovascular anomalies when compared with those born to nonsmokers. This cardiovascular category included infants with a patent ductus arteriosus, ventricular septal defect, atrial septal defect, congenital stenosis of any valve, teratology of Fallot, and transposition of great vessels.

In this study Neural tube defects were the most common congenital anomalies (55.3%). This abnormality can be simply prevented by folic acid supplementation during pregnancy, for which public awareness should be increased. Again the incidence of congenital abnormalities as expressed in this study represent only those which were visible on gross examination. The postmortem study of stillborn and neonatal deaths is not a routine feature and therefore has not been taken into consideration.

In conclusion, and in the light of our data, each governorate in upper Egypt should develop a strategy to reduce the impact of birth defects, a framework of activities by which this can be accomplished, in the form of introducing genetic services, training of the personnel at all levels of health care to build a strong foundation for primary care system, and finally, monitoring of health are delivery and outcomes. Therefore, national policy should support the collection

analysis and dissemination of information on health care outcomes.

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