ORIGINAL ARTICLE

Types of Congenital Anomalies among Families of Medical Students

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ABSTRACT.

Aim: To confirm the types of congenital anomalies and find out the relationship of dietary factors, addiction pattern, radiation exposure, genetic causes, infections, toxic substances, family history of any anomaly and consangunous marriages and to find the most common and most rare anomaly, their ratio in male and female babies and child mortality due to that anomaly among families of medical students of a Private Medical College Lahore.
Study design: Cross-sectional descriptive.
Study population and settings: Study was conducted among MBBS Medical students of all five years, of CMC Lahore. Duration was of three 3 months from June 2015 to August 2015.
Results: This study was based on the Types of Congenital Anomalies in families of Medical Students of all 5 years. A sample of 100 students was selected with positive family history of anomalies in their families. According to the results obtained, the percentage of the diseases which were mentioned by students in decreasing order were Cleft Palate 16%, CVS Disorder 12%, Thalasemia 12%, Polydactyly 10%, Mental Retardation 10%, Down Syndrome and Turner Syndrome 9%, Limb Defects 6%, Congenital Deafness 5%, Macrocephaly 5% and Microcephaly 4%,Cleft lip 4%, Night Blindness 3% Congenital Cataract 2%, Anencephaly 1% ,Micrognathia 1%. The rate of consanguinous marriages was 67% in 1st cousins, 23% in 2nd cousins and 10% out of family.
Conclusion: From the results it is concluded that the students of CMC have a complete awareness about Congenital Anomalies. We got 100 positive cases of congenital anomalies in the families of medical students out of 500 students. The most common diseases seen were Thalassemia, CVS disorder and Cleft Palate. The most rare diseases are Micrognathia, Congenital Cataract and Anencephaly.
Key words: Types of Congenital Anomalies, Families of medical student

INTRODUCTION

A congenital disorder, or congenital disease, is a disorder present at birth and sometimes before birth, or that develops during the first month of life (neonatal disease), regardless of causation. Of these disorders, those characterized by structural deformities are termed "congenital anomalies" and involve damage to a developing fetus. A congenital disorder is mostly the result of genetic abnormalities, the intrauterine causes, defects in morphogenesis, infection, or a chromosomal abnormality. The outcome of the congenital disorder will depend on complex interactions between the pre-natal deficit and the post-natal environment [1]. Animal studies indicate that the mother's (and possibly but less probably the father's) diet, vitamin intake, and levels of glucose prior to ovulation and conception have long-term effects on fetal growth and adolescent and adult disease[2]. Any substance that causes birth defects is known as a teratogen. Some disorders can be detected before birth by prenatal diagnosis (screening).

A congenital physical anomaly; is an abnormality of the body part structure. An anomaly may or may not be perceived as a problem condition. Many people have one or more minor physical anomalies if examined carefully. Examples of minor anomalies include curvature of the 5th finger (clinodactyly), a third nipple, tiny indentations of the skin near the ears ,4th metacarpal or metatarsal bones shortness, or dimples over the lower spine (sacral dimples). Sometimes minor anomalies are clues to more significant internal abnormalities[1]. A congenital malformation is a congenital physical anomaly that is deleterious, i.e., a structural defect perceived as a problem[4].

Examples of primary structural are limb anomaly is called a dysmelia. These include all forms of limbs anomalies, such as ectrodactyly, Amelia, phocomelia, polymelia, polydactyly, syndactyly, polysyndactyly, brachydactyly, achondroplasia, oligodactyly, congenital aplasia or hypoplasia, amniotic band syndrome, and cleidocranialdysostosis. Congenital anomalies of the heart include patent

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ductusarteriosus, ventricular septal defect, atrial septal defect and tetralogy of fallot[5].

Chromosomal: Down Syndrome and Turner Syndrome which result due to some mutations in genetic recombination[6].

A review published in 2010 identified 6 main teratogenic mechanisms associated with medication use: folate antagonism, neural crest cell disruption, endocrine disruption, oxidative stress, vascular disruption and specific receptor- or enzyme-mediated teratogenesis[7].

A vertically transmitted infection is an infection caused by bacteria, viruses, parasites transmitted directly from the mother to an embryo, fetus or baby during pregnancy or childbirth. It can occur when the mother gets an infection as an intercurrent disease in pregnancy[8]. Lack of nutrients; e.g., a lack of folic acid in the mother diet can cause cellular neural tube deformities that result in spinabifida[9].

Genetic causes of congenital anomalies are inheritance of abnormal genes from the parents, as well as new mutations in one of the germ cells that gave rise to the fetus .Genetic disorders or diseases are all congenital, though they may not be recognized until later in life[10]. Genetic diseases may be divided into single-gene defects, multiple-gene or chromosomal defects. Single-gene defects may arise from abnormalities of both copies of an autosomal gene (a recessive disorder) or of only one of the two copies (a dominant disorder). Some conditions result from deletions or abnormalities of a few genes located contiguously on a chromosome. Chromosomal disorders involve the loss or duplication of larger portions of a chromosome containing bundles of genes. Large chromosomal abnormalities always produce effects on body parts or organ systems[11].

Unknown or multifactorial; despite significant progress has been made in identifying the etiology of some birth defects, approximately 65% have no known or identifiable cause. These are sporadic, a term that implies an unknown cause, random occurrence regardless of maternal living conditions, and a low recurrence risk for future children[12]. For 20-25% of anomalies there is a "multifactorial" cause, meaning a complex interaction of multiple minor genetic anomalies with environmental risk factors. Another 10-13% of anomalies have a purely environmental cause (e.g., infections, illness, or drug abuse in the mother). Only 12-25% of anomalies have a genetic cause, the majority are chromosomal anomalies[13].

Role of radiation. For the survivors of the atomic bombing of Hiroshima and Nagasaki, who are known as the Hibakusha, there was statistically demonstrable increase of birth defects/congenital malformations. Anomalies were found among their later conceived children, or found in the later conceived children of cancer survivors who had previously received radiotherapy[14,15,16,17]. The surviving women of Hiroshima and Nagasaki who conceived, though exposed to substantial amounts of radiation, went on and had children with higher incidence of abnormalities/birth defects than in the Japanese population[18,19].

Pre and peri-conceptional care includes basic reproductive health practices as well as medical genetic screening and counseling. Screening can be conducted during the following three periods[20]. Preconception screening can identify persons at risk for specific disorders or at risk for passing a disorder on to their children. Screening includes obtaining family histories and carrier screening, and is very beneficial in countries where consanguineous marriage is common[21,22].

Antenatal screening includes screening for advanced maternal age, incompatibility of Rhesus blood group, carrier screening and screening for alcohol, tobacco and other psychoactive substance use[23].

Newborn screening includes clinical examination and screening for haematological, hormonal, andmetabolicl disorders. Screening for heart and deafness defects as well as early detection of birth defects can facilitate life-saving treatments and prevent the more serious physical, intellectual, visual or auditory disabilities[24].

In countries with developed health services, many structural birth defects can be corrected with paediatric surgery and early treatment can be provided to children with functional problems such as thalassaemia (inherited recessive blood disorders), sickle cell disorders and congenital hypothyroidism[25].

RESEARCH METHODOLOGY

This cross-sectional descriptive was carried out from July,2014 to September,2014. The population for study chosen by us was the MBBS students of all years of Continental Medical College, Lahore who consented to be included in the study and they had any type of congenital anomaly in the family. So 100 students gave the family history of having congenital anomalies and they were included in the study as a purposive sampling technique. The instrument for data collection was a Structured, Pre-tested Questionnaire. Following protocols were followed:

- Explanation of the purpose of study: Students were informed about our research project and study.
- Interpretation: The technique of face to face interview was used to draw:

- Socio-demographic Data of families with anomaly.
- Family history of any congenital anomalies
- Cosanguinous Marriages
- Maternal history of any disease during pregnancy.
- Respondent's awareness about Congenital Anomalies

Data was entered and analyzed through SPSS version 16. Frequencies and the proportions were calculated for selected variable. All results were presented in the form of tables and graphs.

RESULTS

According to the results obtained, the percentage of the diseases which were mentioned by students in decreasing order were Cleft Palata 16%, CVS Disorder 12%, Thalassemia 12%, Polydactyly 10%, Mental Retardation 10%, Down Syndrome and Turner Syndrome 9%, Limb Defects 6%, Congenital Deafness 5%,Macrocephaly 5%, Microcephaly 4%,Cleft lip 4%, Night Blindness 3% and CongenitaL Cataract 2%,Anencephaly 1%, Micrognathia 1%

The rate of consanguis marriages was 67% in 1st cousins, 23% in 2nd cousins and 10% out of family. All these marriages resulted in some congenital anomalies. The anomalies noticed by parents were 37% and the anomalies diagnosed by doctors were 63%. Live birth were 89% and the still births 11% and the parents were well aware of the cause of death. Some anomalies started to appear after some months. 60% of children are still alive. 2% died after 2 months, 2% died at the age of 2 years, 1% died when they turned 4 and 2% died at the age of 20 and 24 years. Only 2% of the mothers have history of alcohol consumption while 98% of the mothers are alcohol free. During pregnancy, the percentage of mothers who suffered from different diseases were

Anemia 19%, Gestational Diabetes 16%, Hypertension 58%, Thalassemia 1% and the mother who remained healthy during pregnancy were 6%. The mothers who had exposure to radiations in different trimesters were 1st Trimester 17%, 2nd Trimester 11%, 3rd Trimester 4%. Fifty seven (57%) of the mothers have undergone vaccination during pregnancy and 42% have no history of vaccination during pregnancy. 93% of the students said that it is important for the mother to have complete antenatal, natal and postnatal care while 7% said that it is not that much necessary. 88% of the children were delivered at hospital and only 12% were delivered at home. 59% of the students said that if there are informed that the growth of the fetus is retarded they would prefer to terminate the pregnancy but 41% said that they will continue the pregnancy. With the help of this survey we estimated that 29% of the mothers used herbal or homeopathic medicine during pregnancy while 71% have no history of such drugs. The anomalies appeared in 1st child was 42% while the anomalies in 2nd or other children was 58%. 14% of the children with congenital anomalies had some defects in their siblings too while there was no such defects in 86% of the siblings.

From the results it is concluded that the students of CMC have a complete awareness about Congenital Anomalies. Out of 500 MBBS students we got 100 positive cases of congenital anomalies in the families of Medical students. The most common diseases seen are Thalassemia, CVS disorder and Cleft Palate. The rarest diseases are Micrognathia, Congenital Cataract and Anencephaly.

DISCUSSION

We did our research on 100 medical students with positve family history of congenital anomalies in their families. Among those 100 cases the most common anomalies were Cleft Palate, Cleft Lip, CVS Abnormalities, Thalassemia and Polydactyly, Mental Retardation. The diseases with low incidence rate were Congenital Deafness, Microcephaly and Limb Disorder, Micrognathia

Congenital anomaly is a worldwide escalating problem caused by a complex interaction of genetic, sociodemographic, behavioural and mainly environmental factors. Societal and behaviour changes over the last decades are held responsible for the considerable increase in inappropriate dietary patterns including smoked food, alcohol consumption, carcinogenic food e.g., nitrosamines, benzo pyrene etc[10,11,12]. There has been only limited success in identifying environmental chemicals that may be responsible for human congenital malformations. Attention in this discussion is directed to major environmental causes (occupational hazards, chemicals of predilection or habituation, pharmaceutical drugs, anticonvulsant drugs, cytotoxic drugs, vaginal spermicides, and nutrition); and prevention of congenital malformations. . Personal habits and preferences involving a variety of substances have been suspected of causing prenatal maldevelopment. They are coffee, tea, and alcohol drinking; cigarette smoking; tobacco chewing; toluene and gasoline sniffing; and drug addiction. . The causes of the majority of congenital defects are still unknown. In time some of these defects and some of those now included in the multifactorial group may be controllable. It is to the understanding and control of such conditions that efforts and resources should be turned[13,14].

More than 20% of all marriages worldwide (more than a billion people) and both Albert Einstein and Charles Darwin were married to first cousins[6,7]. Some cases were also seen in far of family relatives and marriages out of family too. This fact had already been proved worldwide. A complete research was done on the condition of mother i.e., if she had any disease before or during pregnancy. For example some mothers get gestational diabetes, anemia, hypertension, psychosis etc during pregnancy. A question was asked that if parents or grandparents have any anomaly like their child? This question was meant to show the genetic predisposition to some anomalies. According to modern research on consanguity marriages, it is proved that in cousin marriages there is a high chance that the anomaly will pass to the child. Sometimes parents or one of parent has recessive trait for any diseases but this disease pattern becomes dominant in their child e.g., thalassemia[17].

An analysis of a large number of reports alleging that increased rates of congenital malformation and spontaneous abortion were induced by exposure of pregnant women to anesthetic gases concluded only that some evidence indicated an increased rate of spontaneous abortion[11]. Of all substances and situations persons are exposed to at work, only anesthetics have aroused more than a mere suspicion of prenatal harmfulness. Recent findings emphasize that spontaneous abortion is a "soft" sign[13].

None of the drugs most frequently prescribed or self administered has been clearly or consistently implicated as being teratogenic, including aspirin, which has come under suspicion[11]. Many such medications have been either called into questioner accused outright. Those for which such charges are considered by current medical opinion to be dubious or unproved. Recent studies indicate a possible role of a deficiency of folate in the development of neural tube defects, but acceptance of this association should be reserved until expanded trials are completed[14].

Addictive drugs and agents have not been convincingly associated with congenital malformations, although increased stillbirth rates, reduced birth weight, and neonatal withdrawal symptoms have consistently been found in studies of the most commonly used drugs--heroin and methadone[24,25].

Recent studies would seem to have laid to rest the accusation of teratogenesis that had been charged against coffee and by inference other products containing caffeine. Only reduced birth weight has been found to be associated with heavy coffee drinking[26,27]. Like coffee drinking and cigarette smoking the drinking of alcoholic beverages during pregnancy has been reported to have a great variety of effects on the unborn--abortion, growth retardation, defects of the central nervous system, mental retardation, and congenital structural anomalies(29). Alcohol consumption and tobacco smoking is also one of the major concerns regarding the congenital birth defects. So the students were asked if the mother had any tobacco or alcohol consumption during pregnancy because it can lead to serious problems in the fetus. The alcohol consumption rate was very low in the mothers of affected child.

Radiation exposure during pregnancy can lead to congenital anomalies. For the survivors of the atomic bombing of Hiroshima and Nagasaki, who are known as the Hibakusha, there was statistically demonstrable increase of birth defects/congenital malformations. Anomallies were found among their later conceived children, or found in the later conceived children of cancer survivors who had previously received radiotherapy[14,15]. The surviving women of Hiroshima and Nagasaki who were able to conceive, though exposed to substantial amounts of radiation, went on and had children with higher incidence of abnormalities/birth defects than in the Japanese population as a whole[18,19].

A vertically transmitted infection is an infection caused by bacteria, viruses or, in rare cases, parasites transmitted directly from the mother to an embryo, fetus or baby during pregnancy or childbirth. It can occur when the mother gets an infection as an intercurrent disease in pregnancy[31,32]. Any infectious disease during pregnancy can affect fetus growth e.g., rubella infection can cause congenital blindness, syphilis infection can cause corneal opacities, Hutchinson tooth and Saddle Nose Deformity. So it was asked if mother had undergone any vaccination during her pregnancy for any infectious disease. But the vaccination rate was not very high. The choice of termination or to continue the pregnancy in case of diagnosed anomaly during pregnancy, it was proved through research that the reason behind this was that they have religious belief related to it in case of refusal for termination of pregnancy. The age of the mother had a great effect on the fetus. If the mother was above 40 years, there is a high chance of chromosomal abnormality in the fetus e.g., Turner Syndrome or Klinefelter Syndrome.

RECOMMENDATIONS

1. To improve folate status with supplementation of folic acid.
2. To advise women taking medications to seek medical advice before trying to get pregnant

3. To ensure that guidelines are, or are going to be made available for physicians regarding risk-benefits balance for use of medications in pregnancy, particularly those medications used for treating chronic diseases
4. To promote overdosing of vitamins and essential trace elements in women at child bearing age.
5. To ensure pre-conceptional care for women with diabetes, epilepsy and other chronic diseases.
6. To include consideration of specific pregnancy related actions in public health action plans on all the major health determinants.
7. The implementation of policies to minimize the exposure to pollutants identified as teratogens.
8. To ensure a suitable surveillance system where environmental risks can be identified through the integration of congenital anomaly registers with developments in bio-monitoring.
9. Mother should take healthy diet and avoid hazardous conditions which can lead to adverse effects on fetus.
10. The trend of cousin marriages should be discouraged.
11. Take complete family history and if there is a risk of congenital anomalies avoid such marriages.
12. The couples should run complete panel of test which can confirm if they have any chromosomal abnormalities or any other defect.

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