The prevalence of abnormal lab results in pediatric patients at a hospital outpatient clinic: A cross-sectional study

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Abstract---Objective: Examining the prevalence of different forms of birth defects and identifying their causes. Study design: Study Design: A cross-sectional study with retrospective analysis for causes. Place and duration of study: Department of Neurosurgery MMC hospital Mardan from 05 Jan 2021 to October 2021. Methods: All gender-neutral newborns seeking well-child care department of Neurosurgery MMC Mardan. Participating subjects were enlisted, and data were
input and analyzed using SPSS v 22.0, regardless of whether they had received any prenatal care, or vaccinations, or were born with any observable abnormalities. Results: In our analysis of 497 individuals, 23% exhibited CNS congenital abnormalities. 47 children (9%) had gastrointestinal malformations, 111 had musculoskeletal anomalies, 42 had cardio-vascular anomalies, and 181 had genito-urinary anomalies. Extreme maternal age (less than 74 and more than 42 at conception), consanguinity, preterm deliveries, maternal smoking, and family history of birth abnormalities were similarly distributed across patients with diverse malformations. Genito-urinary abnormalities are the most common, followed by CNS and musculoskeletal anomalies.

Keywords---birth abnormalities, maternal risks, GUT, CNS.

Introduction

A person is considered to be dysmorphic or have a congenital deformity if they have abnormal facial or bodily characteristics. In the past, people often blamed supernatural powers for the occurrence of birth defects. About one in every 33 babies suffer from a congenital anomaly, and as a consequence, 3.2 million are left with impairments and 270 thousand die in their first 28 days of life every year, as reported by the World Health Organization. Malformation, genetic factors, environmental variables, and teratogenesis are all possible reasons for a birth abnormality. A distinction may be made between large and minor malformations. These are often fixed by surgical means. Examples of frequent significant abnormalities include cleft lip and palate, often known as orofacial cleft, and meningomyelocele, a deformity of the neural tube. Mild deformities are mostly aesthetic. Ear tags, clinodactyly, and single transverse palmar creases are all examples of malformations that are considered to be rather modest.

Both genetic and non-genetic factors contribute to the development of congenital abnormalities. Chromosome disorders (such as Down syndrome), single gene (monogenic) disorders such as autosomal recessive cystic fibrosis, autosomal dominant Marfan syndrome, and X-linked (such as hemophilia), and multifactorial disorders resulting from the interaction of multiple genes and environmental factors are all examples of genetic abnormalities. Cleft lip/palate, heart defects, and birth defects affecting the neural tube are examples of the latter defects. Environmental causes, such as maternal phenylketonuria (PKU) or diabetes, teratogens (e.g., alcohol, oral isotretinoin), infections (e.g., CMV, rubella), and twinning, are examples of nongenetic etiologies. Fertilization is the typical starting point for teratogenesis. Alterations in cellular development and proliferation, migration, apoptosis (for instance, fetal alcohol syndrome), and interactions between cells or between cells and tissues are only a few examples. Toxic mutagenesis may occur if an organism is exposed to a substance before conception. It has been linked to birth abnormalities if exposed to pregnant women. Even though the FDA mandates testing for all prescription medications, it is sometimes very difficult to identify whether or not a chemical is teratogenic. While research on animals may be instructive, it's possible that the findings won't always apply to people. The teratogenic effects of thalidomide in humans are far
stronger than those in animals, but the same cannot be said of aspirin. These aren’t usually sufficient to prove teratogenicity and instead need confirmation from epidemiological research. Several environmental teratogens have been linked to birth defects. Toxoplasmosis, rubella, CMV, herpes, and syphilis (the so-called TORCH illnesses), as well as varicella and parvovirus B19, are all examples of infectious agents that may cause infection.

Multiple birth abnormalities have been linked to maternal diseases such as insulin-dependent diabetes mellitus, which has been shown to increase the risk of having an abnormally-shaped baby by a factor of two to three. Congenital heart disease and spina bifida risk rise. Maternal phenylketonuria causes microcephaly, intellectual impairment, and cardiac defects. Maternal antibodies may pass the placenta and damage the fetus. Physical factors may also harm the fetus. Elevated maternal core temperature in the first trimester of pregnancy may increase the risk of neural tube defects. Excessive exposure to ionizing radiation may cause fetal mortality, development problems, somatic abnormalities, mutation, chromosomal fragmentation, and cancer.

Medical and recreational maternal drug use may harm fetuses and newborns. Thalidomide, retinoic acid, misoprostol, penicillamine, fluconazole, and lithium are frequent teratogenic medicines. Genetic and environmental causes of birth defects:

1. Extreme maternal age at conception, 21 and >41
2. Consanguinity
3. Early births
4. Smoking mother
5. Birth defect inheritance.

The majority of moms with anomaly infants, 54.92%, were 21 years and >41 years whereas in the control group 47.74% belonged to the extreme age category. Similarly, consanguineous marriage and preterm delivery were higher in anomalous babies at 57.74% and 54.46% as compared to normal babies at 49.66% and 48.64%. Maternal smoking, family history of birth abnormalities, and co-morbid condition in pregnancy are additional important risks; their prevalence was 18.1%, 19.4%, and 15.7% in a Pakistani study. No data on normal newborns is available to establish a suitable link. Local data on baby congenital abnormalities and their causes is sparse. This prompted the current investigation.

Methods

A method called Consecutive Sampling was used, which does not rely on chance. All male and female children with congenital abnormalities meeting the study’s operational criteria who presented to the Neurosurgery Unit MMC Mardan for regular examination or immunization were included. Conditions for exclusion: 1. Clinical evaluations of infants with congenital defects that are part of a syndrome, such as Down syndrome. 2. All Children Under One Year Old Whose Parents Cannot Accompany Them. A proforma with space for each variable served as the primary data collecting instrument. To gather data, informed consent was obtained from guardians first. Every parent whose child met the inclusion
requirements upon presentation to the pediatrics department was contacted. The giving of one’s complete permission obtained consent from parents to participate in the research. To collect information for the research variables, the moms filled out a proforma with their information. The information’s privacy was protected. Echocardiography, a CT scan of the brain and skull, and a full clinical evaluation were performed.

**Data analysis**

SPSS v 22.0 was used for data entry and analysis. Mean and standard deviation was used to summarise the numerical variable (mother’s and baby’s ages). Quantitative variables were presented as frequencies and percentages, including the sex of the infant, the presence and type of congenital anomalies (central nervous system anomalies, cardiovascular system anomalies, muscular system anomalies, and gastrointestinal and genitourinary anomalies), and the presence of contributing factors such as advanced maternal age at conception, consanguineous marriage, maternal active smoking, and the presence of a co-morbid condition during pregnancy. The information was broken down by gender. The Chi-square test and the p-value of 0.05 were used to determine whether or not the gender post-stratification impact could be eliminated The significance level was set at 0.05.

**Results**

Our study population included 497 individuals, with ages ranging from 1 year down to 1 month. There were 197 female patients (34%) and 300 male patients (60%). As a percentage, 16 people had a congenital abnormality in their central nervous system, 47 people had one in their digestive system, 111 people had one in their musculoskeletal system, 42 people had one in their cardiovascular system, and the remaining 181 people had one in their genitourinary system. Premature birth affected 363 (73%) of the cases. Mothers aged 20–39 accounted for 32.8% of all cases (163 total). Among the patients in the study, 56 (11.3%) were born to first or second cousins. Mothers who smoked accounted for 21 (4.2% of the total) of the patients. However, just 52 individuals (10.5%) had a known family history of abnormalities, whereas 445 patients (89.5%) did not. Comorbidities were present in the mothers of 33 patients (6.6% of the total population). Plag

Using a cross-tabulation of congenital anomaly types by gender and the Pearson chi-square test, we found that the distribution of these abnormalities was the same for both sexes (p=0.723). Using a cross-tabulation of congenital anomaly types and a Pearson chi-square test, we found no statistically significant association between prematurity and any of the anomaly types (p=0.999). Our data demonstrated a similar incidence of anomaly types for premature and term births. Using a Pearson chi-square test, we found no statistically significant relationship between the different kinds of congenital abnormalities and the age of the mother, which we found to be between 21 and 41 years old (p=0.165). In this study, we found no evidence that maternal age was related to the occurrence of abnormalities. Data from a cross-tabulation of congenital malformations and cousin marriage showed no statistically significant difference (p = 0.234),
indicating that cousin marriage does not affect the prevalence of congenital abnormalities. Cross-tabulating the different kinds of birth defects with the number of pregnancies in which the mother smoked yielded no statistically significant difference (p=0.108). Patients with maternal smoking history were distributed similarly to those with a maternal nonsmoking history. Applying the Pearson chi-square test to a cross-tabulation of congenital anomaly categories and familial history findings that failed to reach statistical significance (p=0.603). Our findings made it evident that a patient’s family history of abnormalities did not have a major role in the development of a congenital defect. The p-value of 0.167 indicates that the cross-tabulation of congenital abnormality type and maternal comorbidities is not statistically significant.

Table No: 01 Gender Distribution Rate Of Sample Population

<table>
<thead>
<tr>
<th>Gender</th>
<th>Count</th>
</tr>
</thead>
<tbody>
<tr>
<td>Male</td>
<td>301</td>
</tr>
<tr>
<td>Female</td>
<td>197</td>
</tr>
</tbody>
</table>

Table No: 02: family maternal abnormally and morbidity rate

<table>
<thead>
<tr>
<th>Condition</th>
<th>Count</th>
</tr>
</thead>
<tbody>
<tr>
<td>Extreme age of mother</td>
<td>165</td>
</tr>
<tr>
<td>Cousin marriage</td>
<td>56</td>
</tr>
<tr>
<td>Maternal</td>
<td>21</td>
</tr>
<tr>
<td>Family</td>
<td>52</td>
</tr>
<tr>
<td>Maternal anomaly</td>
<td>33</td>
</tr>
</tbody>
</table>

Table 03: Sample population rate distribution by kind of congenital anomaly

<table>
<thead>
<tr>
<th>System</th>
<th>Count</th>
</tr>
</thead>
<tbody>
<tr>
<td>Gastro-intestinal system</td>
<td>181</td>
</tr>
<tr>
<td>Central nervous system</td>
<td>116</td>
</tr>
<tr>
<td>Musculoskeletal system</td>
<td>141</td>
</tr>
<tr>
<td>Genitourinary system</td>
<td>147</td>
</tr>
<tr>
<td>Cardiovascular system</td>
<td>42</td>
</tr>
</tbody>
</table>
Discussion

16 (23%) of our persons had spina bifida, hydrocephalus, or microcephaly\(^\text{11}\). This finding contradicts studies 6, 7, and 8. CNS abnormalities were observed in 51.6% of patients by Masood SN et al.\(^7\), 86.02% by Gul F et al.\(^8\), and 45.94% by Babu RS et al. The observed disparity might be explained by the sample population or procedure. Fewer children with CNS issues have sought treatment due to a lack of a pediatric neurosurgery department\(^12\). 47 children (9%) had digestive system defects, 111 had musculoskeletal system defects, 42 had heart and vascular system defects, and 181 had genitourinary system defects. Our study confirmed the findings of others. Congenital ocular abnormalities were the second most prevalent (19%) following genitourinary defects in a local study\(^13\). In our group, genitourinary anomalies were the most prevalent. According to Raza MZ et al., the most common system was the musculoskeletal system (12.9%), followed by the faciomaxillary system (12.1%), the CNS (10.9%), the gastrointestinal system (3.2%), and the cardiovascular system (2.9%). Depending on the sample size, the indicated difference may not exist. Smoking during pregnancy and advanced maternal age (defined as 21 years or more) are risk factors for congenital abnormalities. Congenital abnormalities are risk factors for advanced mother age during conception, consanguinity, early births, smoking during pregnancy, and family history. Smoking by the mother, a family history of birth defects, and a concurrent pregnancy sickness may all play a role. Our sample size is insufficient to conclude all five criteria. There were 197 women (34.6%) and 300 males (60.4%). The distribution of congenital anomaly categories by gender was found to be the same for both sexes (p=0.723). 363 cases (73%)
were preterm. We found no statistically significant link between preterm birth and any form of the congenital defect (p=0.999). The incidence of abnormalities was similar in premature and term babies. Mothers aged 21 to 41 made up 32.8% of the cases (163 total). We detected no statistically significant connection between congenital abnormalities and mother age using a Pearson chi-square test (p=0.165). Maternal age was not connected to abnormalities in this study. There were 56 patients (11.3%) who were first or second cousins. Cross-tabulated data on congenital malformations and cousin marriage indicated no statistically significant difference (p = 0.234), indicating that cousin marriage did not affect the incidence of congenital abnormalities\textsuperscript{14}.

There were no statistically significant links discovered between congenital abnormalities and maternal smoking (p=0.108). Maternal smokers were dispersed in the same proportion as nonsmokers. In a study that compared the prevalence of congenital malformations by family history, 52 patients (10.5%) had a family history of abnormalities, whereas 445 (89.5%) did not. Using an anomaly database and Pearson chi-square (p=0.603), we found no statistically significant differences. Our findings demonstrated that a patient's family history of abnormalities did not result in congenital malformations. Comorbidities were present in 33 (6.6%) of the patients' mothers. The p-value of 0.167 indicates that both the kind of congenital abnormalities and the maternal comorbidities are not statistically significant\textsuperscript{15}.

**Conclusion**

Most patients had genito-urinary abnormalities, followed by CNS, musculoskeletal, gastrointestinal, and cardiovascular defects. Extreme maternal age (21 and >42 at conception), consanguinity, preterm deliveries, maternal smoking, and family history of birth abnormalities were similarly distributed across patients with diverse malformations. Interessenkonflikt: No conflict-of-interest declarations

**References**