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CLINICAL

Congenital anomalies in newborns and associated maternal risk factors: A cross sectional study in western India

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ABSTRACT

Background-Data from various Indian studies demonstrated 2.5 % of live newborns have congenital anomaly and it is the 3rd most frequent cause of perinatal mortality in India. However, the detailed spectrum and the changes associated with variable maternal risk factors is inadequately studied Objectives- This cross-sectional analytical study aimed to determine the different patterns of congenital anomalies detected in a tertiary care referral teaching hospital and to analyze the maternal risk factors associated with them. Methods- A total of 162 cases of congenital anomalies were studied in the Neonatal unit and its allies in Sassoon General Hospital and B J Medical College, Pune. Results- Overall, the most commonly affected system was musculoskeletal system. Abnormal maternal antenatal USG findings indicate an increased risk for having congenital anomalies, especially of the central nervous system and genitourinary system. Also, consanguinity was associated risk factor in the causation of musculoskeletal anomalies. Conclusions- The pattern of congenital anomalies in western India and the relationship of various maternal factors in relation to congenital anomalies is depicted. Investing in the care and prevention of birth defects reduces child mortality and disability and therefore this should be an integral part of any comprehensive maternal, newborn and childhood health program.

KEYWORDS

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Key words- Congenital anomalies, Newborn, Maternal risk factors, Birth defects.

INTRODUCTION

A congenital anomaly may be narrowly defined in terms of physical structure as a malformation, an abnormality of physical structure or function usually found at birth or during the first few weeks of life; or defined more widely to include functional disturbance as a defect, any irreversible condition existing in a child before birth, in which there is sufficient deviation in the usual number, size, shape, location or inherent character of any part, organ, cell or cell constituent to warrant its designation as abnormal.(1, 2, 3, 4) The World Health Organization in 1991 suggested the need to evaluate the potential burden of congenital disorders in every country, whatever its stage of development, to introduce appropriate preventive measures at the appropriate time.(6) Various studies from India demonstrated 2.5 % of live newborns have congenital anomaly and it is the 3rd most frequent cause of perinatal mortality in India.(5, 7, 8, 9, 10) However, the detailed spectrum and the changes associated with variable maternal risk factors is inadequately studied. This study was hence proposed to study the clinical spectrum of congenital anomalies and to determine the significant maternal risk factors associated with these anomalies. The pattern of congenital malformations prevalent in Pune, Western Maharashtra, India is described in detail.

METHODS

Study setting: A cross-sectional study was conducted at Byramjee JeeJeeboy Government Medical College (BJGMC) and Sassoon General Hospital(SGH), Pune, India between September 2012 till June 2014, among neonates admitted to Neonatal Intensive care Unit, postanatal ward and pediatric surgery ward. Sassoon General Hospital a public hospital offers services to surrounding rural and urban district of Pune. Yearly 10,000 deliveries are conducted at this center. Study population: All neonates delivered at Sassoon General Hospital from birth till 1 month of age with major and minor congenital anomaly admitted in Neonatal Intensive Care Unit, post natal ward and the pediatric surgery ward were included. Still born babies and extramural babies were excluded. Methodology: The study investigators identified neonates with anomalies from case record sheets of the patients. Written informed consent was provided by the parents of enrolled neonates. The newborns enrolled were subjected to detailed clinical examination, and investigations to identify all the congenital anomalies . A validated case report form was used to document all clinical, laboratory and radiological data. Depending upon the anomaly, relevant investigations like radiographs, ultrasonography, transthoracic echocardiography, computerized tomography, magnetic resonance imaging, radiological dye studies were performed to describe the anomaly more aptly and to rule out other known associated anomalies.

Neonatal variables included, gestational age, birth order, gender, type of congenital anomaly and relevant investigations were performed depending upon antenatal USG report and clinical examination. Relevant clinical photographs were obtained after consent from the parents. A structured questionnaire by the study investigator of the mother or father assessed the maternal and environmental risk factors. Variables recorded were maternal age, registration, immunization, parity, history of previous abortions, maternal anaemia, history of systemic illness in mother, drug ingestion, exposure to X-ray, obstetric complications, family history of congenital anomaly, parental consanguinity and antenatal ultrasonography findings were obtained from reports available with the mother.

Study Outcome: The primary study outcome was major congenital anomaly and minor congenital anomaly. The secondary study outcome included maternal risk factors like hypertension, anaemia, systemic illness in mother, drug ingestion by mother, exposure to X-ray, obstetric complications, parental consanguinity and abnormal findings of antenatal ultrasonography.

Définitions:

Major and minor congenital anomaly- The defect was considered as major if potentially life threatening and/or if not corrected, might impair the child’s development or well being while classed as minor when affected non vital organs, had little or no functional effect and doesn’t cause distress in the neonatal period.(4, 5, 14)

Still birth- The definition recommended by WHO for international comparisonis is a baby born with no signs of life at or after 28 weeks gestation.(34)

Statistical analysis: Data was entered into excel data sheet and STATA version software was used for appropriate statistical analysis. Descriptive data was presented in percentage and same was calculated for systemic wise distribution of anomalies. Bivariate analysis was applied to each of the specified suspected maternal risk factor against each system of involvement in the anomaly, and the association was studied by Pearson- Chi Square method. The level of significance was set at $p < 0.05$.

Ethical : The study was reviewed and approved by the ethical review committee of BJGMC.

RESULTS

A total of 162 neonates with congenital anomalies were enrolled. A high coverage (97.5%) of registration and immunization with tetanus toxoid was noted. One fourth of the mothers had history of a previous abortion and maternal obstetric complications were seen in 16.7% of the cases which may indicate the need to specifically screen such pregnancies for congenital anomalies. Consanguinity was found in 23.45% cases which corroborates with the known implication of consanguinity as a causal factor for congenital anomalies. (Table 1)

Half of the cases were born preterm and among the small for gestational age cases, the commonly involved systems were musculoskeletal system, gastrointestinal system and the central nervous system in decreasing order of frequency. The mortality observed was 23.5%, which was in accordance with similar studies from west which also indicate a mortality of upto 25% observed in congenital anomalies.(1, 35)

Single system anomalies were more common than multiple system anomalies. Details of the anomalies system wise are elicited in Table 2. Overall, the most commonly affected system was musculoskeletal system followed by the central nervous system and cardiovascular system, whereas in anomalies of single system involvement, the most common system involved was central nervous system followed by both musculoskeletal and cardiovascular systems.(Figure1) In anomalies involving multiple systems, the most commonly involved system in our study was musculoskeletal system followed by gastrointestinal system. A statistically significant association between parental consanguinity and anomalies of the musculoskeletal system was found. Also, there was a statistically significant association between abnormal antenatal USG findings and anomalies of the central nervous system and of the genitourinary system. (Figure 2& 3) Correlation of maternal age, increasing parity and maternal anaemia with congenital anomalies in our study did not show any statistical significance.

DISCUSSION

This was a cross-sectional analytical study in which total 162 cases of congenital anomalies were studied. In our study we found that 21% cases of congenital anomalies were born to mothers with parity of three or more. However, near to fifty percent were primigravida (42.6%). Similarly Akruti Parmar et al. revealed more congenital anomaly in primigravida mothers i.e. 42%.(11) This was in contrast to previous studies which found high incidence of congenital anomalies in multipara women. Matloob H Y et al. in Iraq found 76.7% of congenital anomalies in mutipara women and suggested that mutlipara women have 1.4 times risk of developing congenital anomaly than primipara.(3)

It was found that out of all cases, approximately one fourth (24.7 %) mothers had history of a previous abortion. There is paucity of studies in literature that have linked previous history of abortions with congenital anomalies. Coulam C B in 1997 had found a possible association between recurrent spontaneous abortion and congenital anomalies, and in that study, the association was attributed to the presence of maternal antiphospholipid antibodies as cause for the anomalies.(12)

Maternal anaemia was found to be present in 19.1% of the cases. Study done by Gupta et al. found maternal anaemia as a relevant factor for congenital anomaly.(13) Maternal infection was found in 16.04% of the cases. It was confirmed by TORCH titres in two of the cases. In one case, the mother had tested positive for anti-toxoplasma IgM antibody at sixth month of gestation and the newborn presented with hemifacialhemimegalencephaly with hydrocephalous. Maternal toxoplasma infection is known to be associated with hydrocephalus but association with hemifacialhemimegalencephaly to the best of our knowledge has not been found presently. In the other case, the mother had tested positive for anti-rubella IgM antibody in the fourth month of gestation and the newborn was having bilateral congenital hydronephrosis. Congenital toxoplasmosis has been associated with congenital hydrocephalous(14, 15), while congenital rubella infection associated with hydronephrosis is a rare complication not reported in literature.

Maternal obstetric complications were seen in 16.7% of the cases, out of which the common ones were oligohydramnios, polyhydramnios and pregnancy induced hypertension and it may suggest that anomalies in the fetus should be suspected and anomaly scans be performed for pregnancies that present with obstetric complications. Gupta et al. found maternal factor of polyhydraminos and oligohydraminos in around 5-10% cases with malformed babies.(13)

Consanguinity was found in 23.45% cases which corroborates with the known implication of consanguinity as a causal factor for congenital anomalies as shown in various other studies done by Abdalla et al., Stoltenberg et al., Tadmouri et al. and further more.(3, 16, 17, 18, 19, 20, 21, 22)

Abnormal findings on antenatal USG were found in 46.92% of the cases, this low rate of detection of the anomalies might be explained by the fact that most of the USGs were performed by gynaecologists or radiologists not well experienced in fetal anomaly scans. A study by Bawa et al. described that a single third trimester scan for foetal anomalies by an experienced sonologist, in areas where routine antenatal screening program has failed or not feasible can help save the newborn’s life at least in surgically correctable anomalies.(23) Thus, antenatal diagnosis of congenital anomalies may be improved if pregnancies are routinely screened by an experienced radiologist for congenital anomalies especially those pregnancies in which some abnormal findings were noted on an earlier sonography.

Male predominance was noted which has been found in several other studies by Taksande et al., Ali et al., Singh et al. and further more.(1, 2, 3, 5, 24, 16, 25, 26) 22.8% newborns with congenital anomalies were small for gestation age, while 3% were large for gestational age. Half the cases enrolled in the study were premature and

most of the newborns(59.3%) were below 2500 gms of weight. The finding is similar to a study done by Marwah S et al. and Matloob et al. where congenital anomalies were significantly higher in preterms than in full term babies.(3, 5) Among the small for gestational age cases, the commonly involved systems were musculoskeletal system, gastrointestinal system and the central nervous system in decreasing order of frequency. The mortality observed was 23.5%. Similarly, studies from west also indicate a mortality of upto 25%.

Among the cases studied, 72.2% cases had single system involvement, while 27.8% cases had multiple system involvement and this finding of predominant single system involvement was also found in a study conducted by Marwah S et al. in 2014.(5) In our study, overall, including both single system and multisystem anomalies, the most commonly affected system was musculoskeletal system(29.6%), followed by the central nervous system(27.8%) and cardiovascular system(20.4%). Musculoskeletal system has been found to be the most commonly affected system in congenital anomalies in other studies done by Ali et al., Singh et al., Chinara et al. and Bhat et al.(1, 2, 27, 16) Other studies done by Marwah S et al., Verma M et al. and further more have found central nervous system to be the most commonly affected.(5, 6, 24, 28, 29, 25, 17, 18, 30) In another study conducted from 2005 to 2007, Taksande et al. found cardiovascular anomalies to be most common followed by musculoskeletal anomalies.(31) FakhriJamil Al-Dalla Ali et al. in 2009found genitourinary system to be most commonly involved followed by central nervous system.(32)

In this study, with regard to anomalies of single system involvement, the most common system involved was central nervous system(28.2%) followed by both musculoskeletal and cardiovascular systems, both being involved individually in 17.9% cases. With regard to anomalies involving multiple systems, the most commonly involved system in our study was musculoskeletal system(60%) followed by gastrointestinal system(31.1%).

In this study, bivariate analysis to check the association of each of the maternal risk factors against the involved systems in the anomalies revealed a positive association between parental consanguinity and anomalies of the musculoskeletal system, thereby suggesting that consanguinity may be a risk factor in the causation of musculoskeletal anomalies. A similar finding was seen in a study done by Sreenivas T et al. in 2012 where they found a positive association between consanguinity and congenital talipesequinovarus.(33) Also, in our study, a statistically significant positive association was found between abnormal antenatal USG findings and anomalies of the central nervous system and of the genitourinary system, which may be interpreted as abnormal antenatal USG findings indicate a significantly increased risk for having congenital anomalies, especially of the central nervous system and genitourinary system. The diversity of causes and determinants of congenital disorder requires a wide range of preventive and interventional health care system for their management and further research is required to evaluate the role of diet and environmental factors in their causation. Large multi-centric studies are needed to determine the exact causes and risk factors for these anomalies and to know their distribution and burden in our country and region.

CONCLUSIONS

The study helps to know the pattern of congenital anomalies and the relationship of various maternal factors in relation to congenital anomalies. Abnormal findings on antenatal USG were found in less than half of the cases, this low rate of detection of the anomalies indicates a need for routine antenatal USG screening by an experienced radiologist for congenital anomalies atleast once especially for those pregnancies in which some abnormal findings were noted at an earlier sonography. Overall, the most commonly affected system was musculoskeletal system. Abnormal maternal antenatal USG findings indicate an increased risk for having congenital anomalies, especially of the central nervous system and genitourinary system. Musculoskeletal system anomalies are specifically associated with consanguinity of parents as a risk factor in this study. Investing in the care and prevention of birth defects reduces child mortality and disability and therefore this should be an integral part of any comprehensive maternal, newborn and childhood health program.

Conflict of Interest: None

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Table 2: DETAILS OF ANOMALIES- SYSTEM WISE

Figure 1 : Frequency of specific system involvement in overall cases

DETAILS OF MATERNAL RISK FACTORS WITH STATISTICAL SIGNIFICANCE-
Figure 3: Association of abnormal antenatal USG findings with anomalies of central nervous system and genitourinary system-

Abnormal antenatal USG findings include polyhydramnios, oligohydramnios, IUGR, single umblical artery, fetal hydrocephalus, cystic lesion in fetal brain, dilated fetal renal pelvis, fetal renal malformations, cleft lip, polydactyly, fetal cardiac septal defects, fetal cardiomegaly and pericardial effusion, hypoplastic left heart, rhizo / mesomelic shortening of fetal limbs, omphalocoele, tracheo-esophagial fistula, situs inversus, duodenal atresia, diaphragmatic hernia etc.

Table 1: Maternal and neonatal demographics

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