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Assessment of prevalence of different congenital anomalies in newborn admitted in ICU of Nalanda medical college, Patna, Bihar

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Abstract

Congenital anomalies affect approximately 1 in 33 infants leading to 6.6% deaths in infants and causing significant morbidity in children. Ever since the discovery of Penicillin by Ian Fleming a lot of antibiotics have been introduced along with development in immunology and medicine also the understanding of the preventive aspect of the infective diseases has lead to significant reduction in the morbidity and mortality of infectious diseases. But these have little impact on congenital anomalies but with the advancement of antenatal ultrasonography and availability of trained paediatric surgeons the congenital anomalies are being identified and treated well. Hence based on above findings the present study was planned for Assessment of Prevalence of Different Congenital Anomalies in Newborn Admitted in ICU of Nalanda Medical College, Patna, Bihar.

The present study was planned in Department of Pediatrics, Nalanda Medical College, Patna, Bihar, India. The present study was conducted from May 2012 to Nov 2012. The study included 50 neonates being admitted to the neonatal nursery for evaluation, observation, investigation and management. The detailed general and systemic examinations of the babies were carried out. As per the proforma made, complete medical, family, antenatal and personal history was taken. Thorough physical examinations of newborn babies were done.

The data generated from the present study concluded that the major congenital malformations observed were from Musculoskeletal, Central nervous System and Cardiovascular systems. Incidence of congenital malformed babies appears more now days as compared to past because of advanced diagnostic facilities and availability of neonatal intensive care unit which leads to increase chances of survival of malformed babies. The life-threatening congenital malformations must be identified by thorough clinical examination because early diagnosis and surgical correction or palliation of these infants offer the best chance for survival.

Keywords: congenital anomalies, newborn, congenital malformations, etc

Introduction

Congenital anomalies are also known as birth defects, congenital disorders or congenital malformations. Congenital anomalies can be defined as structural or functional anomalies (for example, metabolic disorders) that occur during intrauterine life and can be identified prenatally, at birth, or sometimes may only be detected later in infancy, such as hearing defects.

A birth defect, also known as a congenital disorder, is a condition present at birth regardless of its cause. Birth defects may result in disabilities that may be physical, intellectual, or developmental [3]. The disabilities can range from mild to severe. Birth defects are divided into two main types: structural disorders in which problems are seen with the shape of a body part and functional disorders in which problems exist with how a body part works. Functional disorders include metabolic and degenerative disorders. Some birth defects include both structural and functional disorders [1].

Birth defects may result from genetic or chromosomal disorders, exposure to certain medications or chemicals, or certain infections during pregnancy. Risk factors include folate deficiency, drinking alcohol or smoking during pregnancy, poorly controlled diabetes, and a mother over the age of 35 years old. Many are believed to involve multiple factors. Birth defects may be visible at birth or diagnosed by screening tests. A number of defects can be detected before birth by different prenatal tests [2].

Much of the language used for describing congenital conditions antedates genome mapping, and structural conditions are often considered separately from other congenital conditions. Many metabolic conditions are now known to have subtle structural expression, and structural conditions often have genetic links. Still, congenital conditions are often classified in a structural basis, organized when possible by primary organ system affected. Several terms are used to describe congenital abnormalities. (Some of these are also used to describe noncongenital conditions, and more than one term may apply in an individual condition.)

A congenital physical anomaly is an abnormality of the structure of a body part. It may or may not be perceived as a problem condition. Many, if not most, people have one or more minor physical anomalies if examined carefully. Examples of minor anomalies can include curvature of the fifth finger (clinodactyly), a third nipple, tiny indentations of the skin near the ears (preauricular pits), shortness of the fourth metacarpal or metatarsal bones, or dimples over the lower spine (sacral dimples). Some minor anomalies may be clues to more significant internal abnormalities.

Birth defect is a widely used term for a congenital malformation, i.e. a congenital, physical anomaly that is recognizable at birth, and which is significant enough to be considered a problem. According to the Centers for Disease Control and Prevention (CDC), most birth defects are believed to be caused by a complex mix of factors including genetics, environment, and behaviors, though many birth defects have no known cause. An example of a birth defect is cleft palate, which occurs during the fourth through seventh weeks of gestation. Body tissue and special cells from each side of the head grow toward the center of the face. They join together to make the face. A cleft means a split or separation; the "roof" of the mouth is called the palate [3].

A congenital malformation is a physical anomaly that is deleterious, i.e. a structural defect perceived as a problem. A typical combination of malformations affecting more than one body part is referred to as a malformation syndrome.

Some conditions are due to abnormal tissue development: A malformation is associated with a disorder of tissue development. Malformations often occur in the first trimester. A dysplasia is a disorder at the organ level that is due to problems with tissue development.

Conditions also can arise after tissue is formed: A deformation is a condition arising from mechanical stress to normal tissue. Deformations often occur in the second or third trimester and can be due to oligohydramnios. A disruption involves breakdown of normal tissues. When multiple effects occur in a specified order, they are known as a sequence. When the order is not known, it is a syndrome [4].

A limb anomaly is called a dysmelia. These include all forms of limbs anomalies, such as amelia, ectrodactyly, phocomelia, polymelia, polydactyly, syndactyly, polysyndactyly, oligodactyly, brachydactyly, achondroplasia, congenital aplasia or hypoplasia, amniotic band syndrome, and cleidocranial dysostosis. Congenital heart defects include patent ductus arteriosus, atrial septal defect, ventricular septal defect, and tetralogy of Fallot. Congenital anomalies of the nervous system include neural tube defects such as spina bifida, encephalocele, and anencephaly. Other congenital anomalies of the nervous system include the Arnold–Chiari malformation, the Dandy–Walker malformation, hydrocephalus, microencephaly, megalencephaly, lissencephaly, polymicrogyria, holoprosencephaly, and agenesis of the corpus callosum.

Congenital anomalies of the gastrointestinal system include numerous forms of stenosis and atresia, and perforation, such as gastroschisis. Congenital anomalies of the kidney and urinary tract include renal parenchyma, kidneys, and urinary collecting system [5]. Defects can be bilateral or unilateral, and different defects often coexist in an individual child.

Substances whose toxicity can cause congenital disorders are called teratogens, and include certain pharmaceutical and recreational drugs in pregnancy, as well as many environmental toxins in pregnancy [26].

A review published in 2010 identified six 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 [6].

An estimated 10% of all birth defects are caused by prenatal exposure to a teratogenic agent. These exposures include medication or drug exposures, maternal infections and diseases, and environmental and occupational exposures. Paternal smoking use has also been linked to an increased risk of birth defects and childhood cancer for the offspring, where the paternal germline undergoes oxidative damage due to cigarette use. Teratogen-caused birth defects are potentially preventable. Nearly 50% of pregnant women have been exposed to at least one medication during gestation. During pregnancy, a woman can also be exposed to teratogens from the contaminated clothing or toxins within the seminal fluid of a partner. An additional study found that of 200 individuals referred for genetic counseling for a teratogenic exposure, 52% were exposed to more than one potential teratogen [7].

Probably, the most well-known teratogenic drug is thalidomide. It was developed near the end of the 1950s by Chemie Grünenthal as a sleep-inducing aid and antiemetic. Because of its ability to prevent nausea, it was prescribed for pregnant women in almost 50 countries worldwide between 1956 and 1962. Until William McBride published the study leading to its withdrawal from the market at 1961, about 8 to 10,000 severely malformed children were born. The most typical disorder induced by thalidomide were reductional deformities of the long bones of the extremities. Phocomelia, otherwise a rare deformity, therefore helped to recognise the teratogenic effect of the new drug. Among other malformations caused by thalidomide were those of ears, eyes, brain, kidney, heart, and digestive and respiratory tracts; 40% of the prenatally affected children died soon after birth. [8] As thalidomide is used today as a treatment for multiple myeloma and leprosy, several births of affected children were described in spite of the strictly required use of contraception among female patients treated by it.

Vitamin A is the sole vitamin that is embryotoxic even in a therapeutic dose, for example in multivitamins, because its metabolite, retinoic acid, plays an important role as a signal molecule in the development of several tisues and organs. Its natural precursor, β-carotene, is considered safe, whereas the consumption of animal liver can lead to malformation, as the liver stores lipophilic vitamins, including retinol. Isotretinoin (13-cis-retinoic-acid; brand name Roaccutane), vitamine A analog, which is often used to treat severe acne, is such a strong teratogen that just a single dose taken by a pregnant woman (even transdermally) may result in serious birth defects. Because of this effect, most countries have systems in place to ensure that it is not given to pregnant women, and that the patient is aware of how important it is to prevent pregnancy during and at least one month after treatment. Medical guidelines also suggest that pregnant women should limit vitamin A intake to about 700 μg/day, as it has teratogenic potential when consumed in excess. Vitamin A and similar substances can induce spontaneous abortions, premature births, defects of eyes (microphthalmia), ears, thymus, face deformities, and neurological (hydrocephalus, microcephalia) and cardiovascular defects, as well as mental retardation [8].

Tetracycline, an antibiotic, should never be prescribed to women of reproductive age or to children, because of its negative impact on bone mineralization and teeth mineralization. The "tetracycline teeth" have brown or grey colour as a result of a defective development of both the dentine and the enamel of teeth [8].

Several anticonvulsants are known to be highly teratogenic.

Phenytoin, also known as diphenylhydantoin, along with carbamazepine, is responsible for the fetal hydantoin syndrome, which may typically include broad nose base, cleft lip and/or palate, microcephalia, nails and fingers hypoplasia, intrauterine growth restriction, and mental retardation. Trimethadione taken during pregnancy is responsible for the fetal trimethadione syndrome, characterized by craniofacial, cardiovascular, renal, and spine malformations, along with a delay in mental and physical development. Valproate has antifolate effects, leading to neural tube closure-related defects such as spina bifida. Lower IQ and autism have recently also been reported as a result of intrauterine valproate exposure [8].

Hormonal contraception is considered as harmless for the embryo. Peterka and Novotná [8] do, however, state that syntethic progestines used to prevent miscarriage in the past frequently caused masculinization of the outer reproductive organs of female newborns due to their androgenic activity. Diethylstilbestrol is a synthetic estrogen used from the 1940s to 1971, when the prenatal exposition has been linked to the clear-cell adenocarcinoma of the vagina. Following studies showed elevated risks for other tumors and congenital malformations of the sex organs for both sexes.

All cytostatics are strong teratogens; abortion is usually recommended when pregnancy is discovered during or before chemotherapy. Aminopterin, a cytostatic drug with antifolate effect, was used during the 1950s and 1960s to induce therapeutic abortions. In some cases, the abortion did not happen, but the newborns suffered a fetal aminopterin syndrome consisting of growth retardation, craniosynostosis, hydrocephalus, facial dismorphysms, mental retardation, and/or leg deformities [8, 9].

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Methodology

The present study was planned in Department of Pediatrics, Nalanda Medical College, Patna, Bihar, India. The present study was conducted from May 2012 to Nov 2012. The study included 50 neonates being admitted to the neonatal nursery for evaluation, observation, investigation and management. The detailed general and systemic examinations of the babies were carried out. As per the proforma made, complete medical, family, antenatal and personal history was taken. Thorough physical examinations of newborn babies were done.

All the cases with major anomalies were enrolled and minor anomalies like polydactyly, CTEV (congenital talipes equino varus) etc were excluded. Detailed history and thorough physical examination was done. Various imaging modalities like radiography, ultrasound and CT Scan/MRI were done as per requirement. The anomalies diagnosed on pre-natal ultrasonography were confirmed clinically or by appropriate radio-diagnostic methods soon after birth.

All the patients were informed consents. The aim and the objective of the present study were conveyed to them. Approval of the institutional ethical committee was taken prior to conduct of this study.

Results & Discussion

According to WHO, congenital anomalies can be defined as a structural or functional anomaly (eg metabolic disorders) that occur during intrauterine life and can be identified prenatally, at birth or later in life [10]. Congenital malformations occur all over the world and are becoming the most important cause of perinatal mortality. In developing countries like India, with the decline of other causes of perinatal mortality, congenital malformations are acquiring importance. Each year eight million children are born worldwide with congenital anomalies of which 3.3 million die before the age of five; 3.2 million of the survivors may be mentally or physically disabled [11]. whereas in India it accounts for 8-15% of perinatal deaths [12] and 13-16% of neonatal deaths [13]. The significance of congenital malformations lies not only in their contribution to mortality but also in causing disability and handicaps. Beyond the direct impact on affected children and their families, they impose a tremendous financial burden on medical treatment, educational and support services [14]. The inability to discover the causes of congenital malformations is one of the most miserable failures of modern medicine. while much progress has been made in early detection of congenital malformations, the area of etiopathogenesis is still shrouded with darkness. Genetics, drugs, viruses and environment all have a role in causation but for vast majority of malformations the exact cause is not known.

In a developing country like India due to high incidence of infectious diseases, nutritional disorders and social stress, the developmental defects are often overshadowed, but the present scenario is changing rapidly. A recent study shows that congenital anomalies contribute to 9% of perinatal deaths as compared to 8% a decade ago. About 2% newborn infants have major anomalies. The incidence is as high as 5% if one includes anomalies detected later in childhood such as abnormalities of heart, kidney, lungs and spine. Anomalies are more common among spontaneous abortuses. Many anomalies are severe and cause abortion. Congenital anomalies represent defective morphogenesis during early fetal life. A broader definition includes metabolic or microscopic defects at a cellular level.

Table 1: Demographic & General Variables

Variable	Number of Cases
No. of Cases	50
Mother age at birth	
Below 20 years	34
Above 22 years	16

Mother BMI	
Non-Underweight	41
Underweight	9
No. of Children's	
1-2	42
More than 3	8
Gender	
Male	38
Female	12
Birth Weight	
Less than 2 kg	40
More than 2 kg	10

Table 2: Distribution of Congenital Malformations in Newborns

Congenital Malformations	Number of Cases
Musculoskeletal	19
Central nervous System	11
Cardiovascular	9
Genitourinary tract	6
Gastrointestinal	3
Miscellaneous	1
Multiple	1
Total	50

Goravalingappa & Nashi [15] and Guha AK [16] also found high incidence of central nervous system malformations. While Mishra PC & Baveja R [24] found high incidence of multiple congenital anomalies. Ghose et al. [18] and Mohanty et al. [17] found higher incidence of musculoskeletal system malformations.

Other investigators namely Hegazy I S et al., [19] Mir N A et al. [20] found that different congenital anomalies were significantly related to male gender. Racial variations, geographical location and socioeconomic status all play an important role in determining the incidence of congenital anomaly in a given population eg., most of the studies from Middle East countries refer consanguinity as one of the leading cause of congenital malformations. Various researchers Kulkarni and Kurian, [21] Tayebi et al. [22] has established the fact that consanguinity has a deleterious effect on fetal growth, and it increases the risk of congenital malformations and fetal loss. The present study found three cases of consanguineous marriage in this region. Although elderly age group and higher parity are considered as risk factors for congenital anomaly.

The incidence of congenital malformations was significantly higher among the low birth weight babies (less than 2000 g) in comparison to normal weight babies. This association of low birth weight and malformations has been well documented [23, 24]. Many studies have documented male preponderance amongst congenital malformed babies. [23] However, in the present series, like some of the earlier studies, we could not observe any sex predilection of malformation. A significantly higher incidence of malformation observed among the stillbirths in the present study is consistent with earlier reports. Aiyar and Agrawal [25, 26] observed that the highest incidence of malformations was among fullterm normal weight babies.

Differences in reported birth prevalence rates of congenital malformations over time and among countries or even within the same country among regions, may be attributed to one or more factors such as design of the study (hospital based or population based), prospective or retrospective, inclusion criteria used, type of surveillance system, etiological heterogeneity of malformations, accuracy of diagnosis, gestational age at which these are included in monitoring reports etc. These factors make comparison of studies difficult. The true incidence of congenital malformation can only be determined if all livebirths, fetal deaths and spontaneous and induced abortions are examined. However, in this study, spontaneous abortions were not studied.

It had been shown that, the frequency of structure abnormalities increase in the first year of life as what calls, silent abnormalities which might not detected at birth, such as heart defects, anomalies of the urinary tract, and bowel malrotation, which were identified from diagnostic studies encouraged by signs and symptoms in the affected infant, However, it was documented that, role of environmental pollutants, drugs and infectious agents in causing congenital anomalies are a major global concern. Furthermore, the underlying causes for the most congenital malformations remain obscure and multifactorial inheritance is believed to be the underlying etiology of most common abnormality

Medical fraternity and society attach lot of value to congenital anomalies for multifarious reasons. One, congenital anomalies are a significant challenge to bring further reductions in neonatal mortality. Two, long-term care of survivors demands huge resources and societal support. Therefore, it is important that the burden of congenital anomalies is reflected appropriately and accurately.

Congenital anomalies contribute to a significant proportion of fetal and infant mortality. Various sources estimate the prevalence of congenital anomalies to be in the range of 1%–3% of all live-born infants and the estimates are considerably higher for the infants that are stillborn or spontaneously aborted. Unfortunately, we do not have complete data about congenital anomalies. Older women, women with medical conditions such as hypothyroidism, uncontrolled diabetes, placental insufficiency, multiple pregnancy, and oligohydramnios have a higher risk of major congenital anomalies than that of the general population.

There are many challenges that exist in the Indian scenario. Till recently, India lacked dedicated national surveillance systems for birth defects. Though the Rashtriya Bal Swasthya Karyakram is still in the fledgling stage, it is envisioned that national data will be generated by early identification of certain birth defects. A major challenge that will emerge with increased detection and referral is being able to provide timely corrective surgery when warranted. Currently the ratio of skilled personnel to population is extremely low. Out of the 1% live newborns with congenital heart disease that result in 10% infant mortality, less than 2% receive life-saving surgery [27]. Hence developing a parallel program aimed at capacity building is essential. Till tertiary level fetal screening becomes easily available,

affordable and accessible to all pregnant mothers at risk, the only option is relying on more basic community-based preventive health measures like preconception care and improving the health of women of reproductive age group.

Conclusion

The data generated from the present study concluded that the major congenital malformations observed were from Musculoskeletal, Central nervous System and Cardiovascular systems. Incidence of congenital malformed babies appears more now days as compared to past because of advanced diagnostic facilities and availability of neonatal intensive care unit which leads to increase chances of survival of malformed babies. The life-threatening congenital malformations must be identified by thorough clinical examination because early diagnosis and surgical correction or palliation of these infants offer the best chance for survival.

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