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ORIGINAL ARTICLE

An Inquiry into Fetal Congenital Anomalies: Exploring Termination Methods and Ethical Considerations

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ABSTRACT:
Background:In the current global shift from communicable to noncommunicable and chronic genetic diseases, there is a noticeable increase in congenital malformations. These malformations rank as the fifth major cause of neonatal deaths in India. It's important to note that the prevalence may be underestimated, as existing data primarily stems from hospital-based studies. The advent and widespread adoption of prenatal ultrasound (USG) as a diagnostic tool in obstetrics have significantly improved the detection of fetal anomalies.The study's objectives are multifaceted. Firstly, it aims to estimate the prevalence and patterns of congenital malformations, exploring their distribution and associated risk factors. Additionally, the study seeks to compare various methods employed for terminating pregnancies in cases of fetal anomalies during both the first and second trimesters.**Methods**:The research described herein is a cross-sectional study carried out at a tertiary care hospital spanning a duration of four years.**Results**:Among 3,200 deliveries observed, a total of 50 cases involving major congenital anomalies were identified, resulting in a prevalence rate of 1.26%. Notably, central nervous system (CNS) anomalies were the most frequently recorded. The majority of these cases were detected during the second trimester of pregnancy. The primary method of termination employed was medical, utilizing a combination of Mifepristone and Misoprostol.**Conclusion**:In conclusion, the prevalence of congenital anomalies is found to be 1.26%. Among these anomalies, the central nervous system (CNS) is the most commonly affected, with anencephaly being the most frequently reported condition. The termination of pregnancy using a combination of Mifepristone and Misoprostol appears to yield the most favorable outcomes. It is noteworthy that the incidence of congenital anomalies could potentially be mitigated through heightened awareness, effective counseling, and vigilant surveillance measures.
Keywords: trimester, congenital, malformations, Mifepristone.

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INTRODUCTION

In the context of recent decades, where strides in genetic science and medical technologies have allowed for the early detection of fetal anomalies during intrauterine life, the profound decision to terminate a pregnancy due to a loss of hope for a healthy child emerges as an emotionally challenging and complex issue. Navigating this traumatic event requires a delicate and supportive approach from healthcare professionals.[1]In this scenario, the pivotal role of healthcare providers is to not only offer clear explanations of the current situation but also to provide comprehensive information and non-judgmental counseling. The aim is to empower couples with the knowledge and emotional support necessary for making informed decisions. A study conducted by Asplin underscores the ongoing need for information and counseling at various points in time for women facing these challenging circumstances.However, a concerning trend illuminated by various studies is that women undergoing pregnancies, especially those contemplating the decision to terminate, often grapple with enduring psychological outcomes. Post-traumatic stress, severe grief, and depression are among the long-term challenges faced by these individuals. Complicating matters further is the time-sensitive nature of deciding to terminate a pregnancy, intensifying the emotional burden on parents.Amidst the urgency, women find themselves caught in severe internal conflicts, with emotions ranging from self-blame to denial, anger, and depression.[2] The gravity of the situation is heightened by the realization that, in many instances, women do not receive the necessary information and support essential for navigating this complex terrain.In light of these challenges, healthcare providers bear a significant responsibility to ensure that women receive comprehensive support, including the vital information needed to make decisions during this emotionally charged process. This approach not only acknowledges the gravity of the situation but also underscores the importance of fostering an environment that prioritizes empathy, understanding, and the well-being of those facing these difficult choices.

Congenital anomalies, also commonly known as birth defects or congenital malformations, encompass a wide spectrum of structural and functional

irregularities that arise during the intricate process of intrauterine development.[3] As defined by the World Health Organization (WHO), these anomalies extend beyond mere physical deviations, often affecting the functionality of various organs and systems. Their significance is underscored by the considerable and lasting impact they exert on individuals, families, and societies.According to the WHO, congenital anomalies contribute substantially to long-term disabilities, posing challenges that extend beyond the affected individuals to influence the dynamics of families and societies at large. The global burden of disease study recognizes congenital anomalies as a significant factor, ranking them among the top 10 causes of perinatal mortality. This acknowledgment emphasizes the urgency and importance of addressing the complexities associated with these anomalies on a global scale.On an epidemiological level, the incidence of congenital anomalies worldwide is estimated to be approximately 2-3%. However, it is crucial to note that a noteworthy percentage of these anomalies are incompatible with sustaining life, presenting both medical and emotional challenges for affected families.Breaking down the prevalence by organ systems, congenital anomalies involving the Central Nervous System (CNS) stand out as the largest group, affecting 10 per 1000 live births. Following closely, Cardiovascular System (CVS) anomalies are reported at 6 per 1000 live births, Renal anomalies at 3 per 1000 live births, and limb anomalies at 1 per 1000 live births.[4,5] This categorization provides a nuanced understanding of the distribution and impact of congenital anomalies across different physiological domains.Despite advances in medical knowledge, the etiology of congenital malformations remains unknown in a significant 60% of cases. This highlights the intricate and multifaceted nature of these anomalies, necessitating ongoing research efforts to unravel the complex interplay of genetic, environmental, and other contributing factors. Approximately 20-25% of congenital anomalies are attributed to identifiable causes, including genetic factors, infections, metabolic diseases, exposure to certain drugs, and environmental influences. Recognizing these diverse influences is paramount in developing comprehensive strategies for prevention, early detection, and management.In essence, the study and understanding of congenital anomalies require a holistic and interdisciplinary approach that combines medical, genetic, environmental, and public health perspectives.[6] By delving into the intricate factors contributing to these anomalies, the goal is to enhance preventive measures, support affected individuals and families, and ultimately alleviate the global burden associated with congenital anomalies.

The genetic underpinnings of congenital anomalies are diverse, encompassing numerical abnormalities, structural defects, and mosaicism. Structural chromosomal anomalies involve alterations such as deletions, translocations, and inversions. Notable sex-linked chromosomal abnormalities include Turner's syndrome and Klinefelter's syndrome.Infections during pregnancy pose a significant risk for congenital anomalies. Pathogens like Rubella, Herpes, Cytomegalovirus, Toxoplasmosis, and Syphilis are known culprits. Additionally, exposure to various environmental factors can contribute to congenital anomalies.[7] The increased use of irradiation, alkylating agents, antimetabolites, as well as habits like smoking, alcohol consumption, and exposure to pesticides, are recognized contributors to these anomalies.Certain medical disorders also play a role in causing congenital anomalies. Uncontrolled diabetes and hypothyroidism are among the conditions associated with an increased risk. Exposure to specific drugs, such as anti-epileptics, warfarin, thalidomide, and retinoic acid, during pregnancy may elevate the risk of congenital anomalies.Implementing preventive strategies is crucial in mitigating the incidence of congenital anomalies. This involves avoiding exposure to radiation, antimetabolites, smoking, and alcohol.[8] Pre-conception and antenatal folic acid supplementation have been identified as effective measures in reducing the risk of congenital anomalies.In cases where congenital anomalies are diagnosed, termination of pregnancy becomes a significant consideration. This approach can lead to a marked decrease in perinatal mortality and morbidity. Advances in prenatal diagnostic testing have played a pivotal role in enhancing the early detection of anomalies during pregnancy. This early detection facilitates timely intervention, contributing to improved outcomes and the overall management of congenital anomalies.

MATERIALS AND METHODS

This research initiative was conducted within the premises of a prominent tertiary care hospital located in India, spanning a duration of four years. The primary focus of the study was to investigate and analyze antenatal women whose unborn children were diagnosed with congenital anomalies. The participants were either registered with or referred to the Obstetrics and Gynecology department at NRIGH, the designated hospital for this research endeavor.

The inclusion criteria were carefully delineated to encompass a specific cohort—pregnant women grappling with the complexities of congenital anomalies in their developing fetuses. The data collection process was meticulous, targeting a range of variables to create a comprehensive profile of each participant. These variables included age, parity (number of previous live births), gestational age, consanguinity (relatedness between parents), history of recurrent abortions, maternal infections, folic acid intake, obesity, prior instances of congenital anomalies in the family, and any pre-existing medical conditions such as diabetes or thyroid disease.

By incorporating such a diverse array of variables into the data collection framework, the study aimed to paint a detailed and multifaceted portrait of the demographic and medical characteristics of pregnant women facing the challenging prospect of congenital anomalies in their unborn children. This thorough exploration sought to provide valuable insights into potential patterns, associations, and risk factors associated with congenital anomalies. Such comprehensive information is crucial for advancing our understanding of these complex conditions and could potentially inform more effective strategies for diagnosis, prevention, and the overall management of congenital anomalies in the antenatal population.

RESULTS

In the extensive pool of 3,200 deliveries scrutinized during the course of this study, a distinctive finding emerged with the identification of 50 cases featuring major congenital anomalies. This revelation serves as a crucial insight into the prevalence of congenital anomalies within the context of the observed deliveries. The calculated prevalence rate of 1.26% encapsulates the proportion of deliveries wherein significant congenital anomalies were diagnosed.This prevalence rate becomes a pivotal metric in understanding the magnitude of the issue within the studied population. It signals the imperative for heightened awareness, thorough antenatal screening, and comprehensive healthcare strategies to address the challenges posed by major congenital anomalies.This data not only highlights the prevalence of congenital anomalies but also underscores the potential impact on maternal and child health. It beckons the need for continued research to delve into the underlying causes, risk factors, and potential interventions to mitigate the occurrence and impact of these anomalies. By expanding our understanding of congenital anomalies, healthcare practitioners can refine their approaches to antenatal care, offering better support and management for expectant parents facing such circumstances.In essence, the prevalence rate of 1.26% serves as a numerical representation of the scope of major congenital anomalies in the context of the observed deliveries. This figure catalyzes a deeper exploration into the intricacies of these anomalies, paving the way for informed strategies aimed at prevention, early detection, and improved outcomes for both mothers and infants.

Table 1: Age Wise Distribution

Maternal age in years	No. of cases
< 20 years	14
21- 25 years	20
26 – 35 years	13
36-40 years	3

Table 2: Distribution According To Parity of The Women

Parity Of Women	No. Of Cases
Primigravida	23
Multigravida	27

Figure 1: Distribution According To Parity of The Women

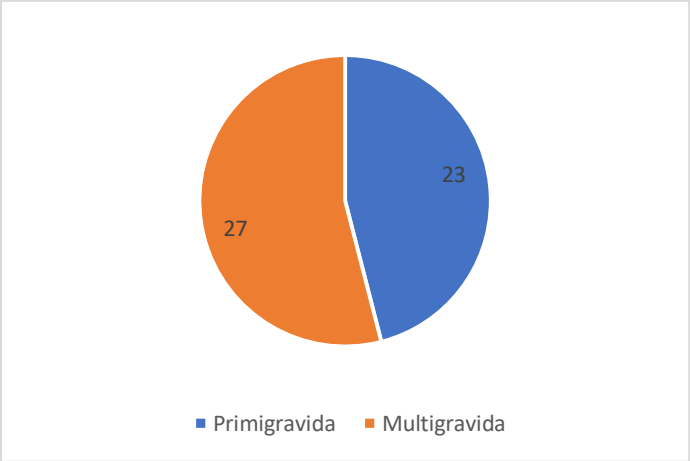

Table 3: Gender of The Fetus

Gender Of The Fetus	No. Of Cases
Male	18
Female	23
Ambiguous	6

Figure2: Gender of fetus

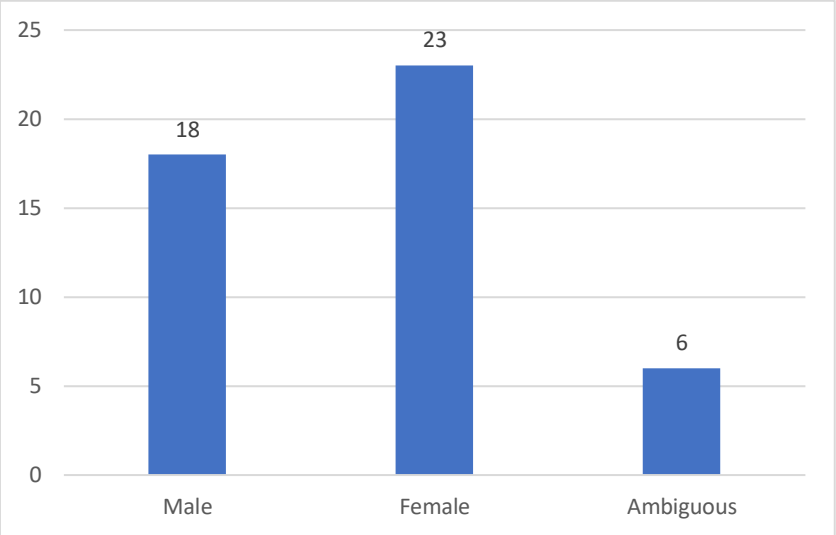

Figure 3: Risk Factors

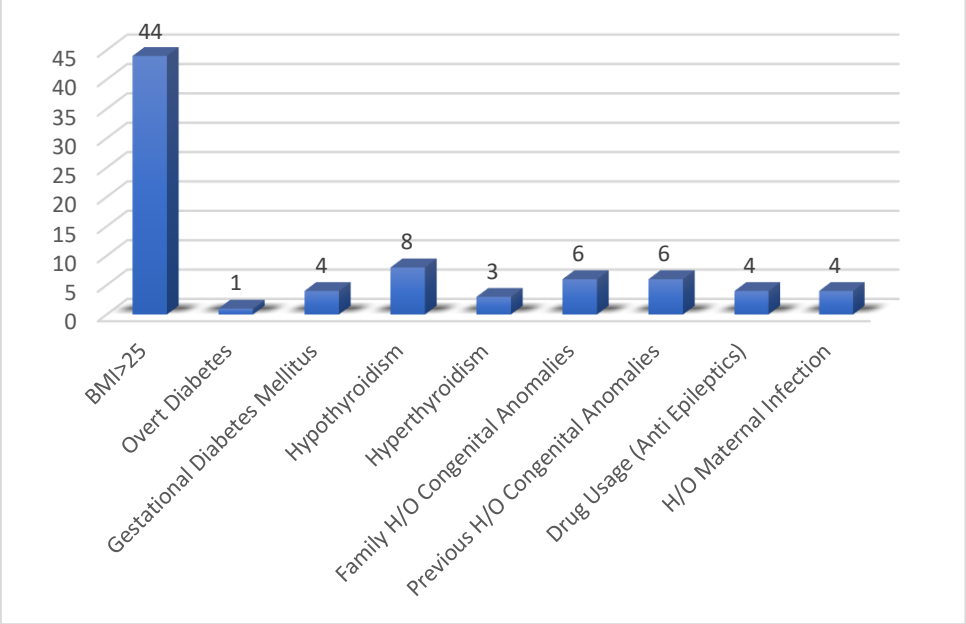

Table 4: Method of Termination

Method Of Termination	NO. OF CASES
Medical (Mifepristone + Misoprostol)	33
Mifepristone Alone	1
Misoprostal Alone	1
Mechanical Dilation With Foley's Catheter	3
Extra-Amniotic Ethacridine Lactate	7
Dinoprostone Gel	2
Extra Amniotic Saline Infusion	1
Hysterotomy	1

DISCUSSION

Congenital anomalies represent a critical factor contributing to adverse outcomes in pregnancy, encompassing the spectrum of stillbirths, perinatal mortality, and childhood morbidity. The prevalence of congenital anomalies, a focal point in our study, was documented at 1.26%, aligning with the established global prevalence range of 2-3%. Acknowledging the inherent challenges in preventing congenital anomalies outright, the study emphasizes the pivotal role of advancing technology, particularly the development of high-resolution ultrasound, and the growing expertise of medical professionals, both of which have markedly improved detection

rates.[9,10]Early detection of anomalies during gestation is a pivotal breakthrough, creating avenues for timely interventions and management. The study also identified an increased risk of congenital anomalies in women of advanced maternal age. While the study did not yield a substantial number of pregnant women above 35 years for definitive conclusions, the majority of registered patients were in the 21-25 age group, with a higher incidence of anomalies noted in this demographic.In consonance with prior research, the study underscored a higher incidence of congenital anomalies in multigravida compared to primigravida, aligning with findings from a study conducted by Mohanty C et al. This correlation raises intriguing questions about the potential association between multiple pregnancies and the occurrence of congenital anomalies.

Further analysis of the termination of pregnancy group shed light on the distribution of congenital anomalies within this subset.[11] Central Nervous System (CNS) anomalies emerged as the predominant category at 51%, followed by cardiovascular anomalies at 26%, musculoskeletal anomalies at 13%, renal anomalies at 11%, facial anomalies at 6%, gastrointestinal tract anomalies at 4%, respiratory anomalies at 2%, and hydrops at 5%. This detailed breakdown offers valuable insights into the prevalence and diversity of anomalies in cases where termination of pregnancy was pursued.A notable contrast with the 45th annual report of the Indian Council of Medical Research (2002-03) lies in the emphasis on cardiovascular anomalies as the most common cause. This discrepancy is contextualized by the specific inclusion criteria of the present study, focusing on patients registered for the termination of pregnancy, thereby influencing the observed distribution of anomalies.In summary, this study contributes significantly to the comprehension of congenital anomalies by elucidating their prevalence, demographic associations, and distribution among those choosing termination of pregnancy. The insights gleaned from such research are instrumental in refining healthcare practices, enhancing counseling strategies, and advancing interventions to ultimately improve outcomes for both mothers and infants affected by congenital anomalies.

The findings of this study underscored the paramount importance of tailoring information to the specific circumstances of individuals who have experienced fetal loss.[12] Participants expressed a strong need for information that is personalized to their unique situations, encompassing aspects such as coping strategies and preparation for subsequent pregnancies. This aligns with the broader literature on fetal loss, where similar studies focusing on intrauterine fetal death and abortion have revealed that mothers consistently articulate the importance of addressing their informational and educational needs to facilitate a smoother emotional recovery.A significant revelation from this study was the prevalence of unanswered questions among participants regarding the causes of fetal anomalies. This knowledge gap highlighted a critical area that requires attention in post-fetal loss care. Similar sentiments were echoed in the study by Maguire et al., which found that pregnancy termination due to fetal anomalies is associated with perceived stigma.[13] This underscores the need for targeted counseling to dispel misinterpretations or misunderstandings surrounding the causes of anomalies. Addressing the perceived stigma can play a pivotal role in the emotional healing and well-being of these individuals.Furthermore, the study emphasized the potential disruption in the mourning process when healthcare providers fail to provide sufficient explanations about the causes of anomalies, as noted in Facchinetti's research. Consequently, there is a compelling case for educating parents about the causes of fetal anomalies and associated risk factors. Armed with this knowledge, parents are better equipped to navigate the complexities of subsequent pregnancies, thereby enhancing the management of these pregnancies and reducing the risk of recurrence of such incidents.In essence, the study illuminates the critical need for a comprehensive and personalized approach to post-fetal loss care. This includes addressing informational and educational needs, dispelling stigma, and providing clear explanations about the causes of fetal anomalies.[14] Such interventions can contribute significantly to the emotional well-being of those who have experienced fetal loss, empowering them to navigate subsequent pregnancies with a greater understanding and resilience.

In our comprehensive study, a significant and noteworthy positive association emerged between congenital anomalies and maternal thyroid abnormalities. This finding echoes the results of a study conducted by Khoury MJ et al., reinforcing the potential link between the health of the maternal thyroid and the incidence of congenital anomalies.[15] This association prompts a deeper exploration into the intricate relationship between maternal thyroid function and fetal development, emphasizing the importance of considering thyroid health in the broader context of prenatal care.A pivotal aspect of our study involved an investigation into the methods employed for the termination of pregnancies in cases of congenital anomalies. The predominant and most frequently utilized method was a combination of Mifepristone and Misoprostol. This particular approach was applied in 20 cases and demonstrated notable advantages, including fewer post-abortion complications and reduced hospital stays. These favorable outcomes closely align with findings from a study conducted by J E Dickinson et al., affirming the efficacy and safety associated with the use of the Mifepristone and Misoprostol combination in termination procedures for pregnancies affected by congenital anomalies.The success of medical termination was underscored by the high rate of

complete terminations observed in the majority of cases. Remarkably, only one patient required hysterotomy, further emphasizing the overall effectiveness and acceptability of medical methods in managing cases of congenital anomalies.Our study systematically explored various medical methods employed for termination, offering a diverse spectrum of approaches.[16] These included the combination of Mifepristone and Misoprostol, Mifepristone alone, Misoprostol alone, Dinoprostone gel, Extra-amniotic Ethacridine lactate solution combined with Mifepristone, Extra-amniotic saline infusion, and Mechanical dilatation using Foley's catheter. This diversity underscores the need for a tailored and individualized approach, recognizing the unique circumstances and preferences of each patient.

The findings from our study contribute not only to the understanding of the association between maternal thyroid abnormalities and congenital anomalies but also shed light on the effectiveness of various medical termination methods.[17] This knowledge is pivotal for healthcare professionals in making informed decisions regarding the most suitable approaches for managing pregnancies affected by congenital anomalies. Ultimately, these insights enhance the quality of care provided to patients, ensuring a more nuanced and personalized approach to the challenging circumstances surrounding congenital anomalies.

CONCLUSION

Morbidity stemming from congenital anomalies, while challenging to eliminate entirely, can be significantly minimized through early diagnosis using ultrasound (USG) and timely intervention. The proactive approach of early detection allows for strategic planning, particularly in the context of pregnancy termination, leading to reduced morbidity associated with congenital anomalies.The incidence of congenital anomalies can be effectively curtailed through pre-conception counseling, a crucial component of reproductive healthcare. This counseling serves to educate prospective parents about potential risk factors, encouraging lifestyle modifications and strategies to optimize maternal health before conception. Folic acid supplementation, for instance, is a recognized preventive measure that contributes to reducing the risk of certain congenital anomalies.Early detection of congenital anomalies not only facilitates timely medical and surgical interventions but also provides valuable information for informed decision-making, including the option of pregnancy termination when appropriate. Planning for the termination of pregnancy at an early stage is associated with fewer complications and reduced morbidity, underlining the importance of timely interventions in the management of congenital anomalies.In essence, the multifaceted approach outlined—comprising early diagnosis, pre-conception counseling, risk factor avoidance, folic acid supplementation, management of medical disorders, and strategic planning for timely interventions—serves as a comprehensive strategy to minimize morbidity associated with congenital anomalies. This holistic approach reflects the evolving landscape of prenatal care, emphasizing the importance of proactive measures to optimize maternal and fetal health.

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