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Original Research Article

GENETIC COUNSELING AND ITS IMPACT ON PARENTS WITH CONGENITAL ANOMALY BABIES

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ABSTRACT

Background: Congenital anomalies are a significant cause of neonatal morbidity and mortality, particularly in low-resource settings. Genetic counselling plays a crucial role in educating parents about inherited disorders, associated risk factors, and available preventive and management options for these disorders. This study aimed to assess the types and risk factors of congenital anomalies and to evaluate the impact of genetic counselling on parental awareness.

Materials and Methods: A descriptive observational study was conducted at Mahatma Gandhi Memorial Government Hospital, Tiruchirappalli, from February 2018 to July 2019, involving 182 neonates with congenital anomalies. Maternal history, clinical examinations, and imaging studies were used for the diagnosis. A structured questionnaire was used to assess parental awareness before and after the counselling sessions.

Results: Cardiovascular anomalies were the most common (42.9%), with acyanotic lesions comprising 64.1% of these anomalies. Multiple anomalies were present in 31.4% of neonates, and consanguinity was reported in 31% of the cases. Maternal hypothyroidism and gestational diabetes were observed in 9.9% of pregnancies. Only 13.2% of mothers took folic acid preconceptionally, while 14.3% did not take it at all. Antenatal anomaly scans were performed in 68.1% of the cases. Genetic counselling was attended by 95.6% of the parents. Pre-counselling, 34.5% of parents scored below 7 in awareness, while 81% scored a maximum of 13 post-counselling. The neonatal survival rate was 64.8%, and 55.5% of families adhered to follow-up.

Conclusion: This study highlights the high prevalence of congenital heart defects and multiple risk factors, including consanguinity and maternal metabolic conditions. Genetic counselling significantly improved parental awareness and understanding of congenital anomalies, as evidenced by marked post-intervention score improvements.

Keywords: Congenital anomalies, Genetic counselling, Parental awareness, Consanguinity, Folic acid deficiency.

INTRODUCTION

Congenital anomalies are defined as structural or functional abnormalities that are present at or before birth. They are a major cause of neonatal morbidity and mortality in developing countries. Although these anomalies may result from one or more genetic, infectious, nutritional, or environmental factors, identifying the exact cause is often difficult.[1] Congenital anomalies affect an estimated 3–6% of infants worldwide and are a significant cause of long-term disability, with substantial impacts on individuals, families, healthcare systems, and societies.[2,3] Congenital anomalies are diagnosed in 2–5% of pregnancies, and approximately half of all anomalies are detectable during weeks 18–22 of pregnancy.[4]

Certain risk factors are consistently associated with specific anomalies, including folic acid deficiency, maternal diabetes, advanced maternal age, teratogenic drug exposure, infections during pregnancy (e.g. rubella and Zika virus), genetic predisposition, consanguinity, and alcohol or tobacco use. Early detection through prenatal screening and preimplantation genetic diagnosis can significantly reduce the burden of congenital anomalies in children. However, these measures are often underutilised due to poor awareness, sociocultural beliefs, and limited accessibility.[5]

Congenital anomalies can have a psychological impact on parents. Parental traumatic stress can interfere with parenting practices and lead to adverse family outcomes, such as increased conflict and reduced relationship satisfaction. Consistent with this, high levels of parental conflict may limit parents' ability to meet the potentially substantial demands of caring for a child with a congenital disorder. Importantly, children with malformations may already have compromised health and development. The risk of exposure to negative family dynamics may further threaten the health outcomes of these children.[4] Therefore, it is important to understand the impact of a diagnosis of foetal anomaly on parents' relationships with one another. Understanding more about the impact of congenital malformations on family harmony and parents' psychosocial health may help us improve support for families that could be vulnerable to elevated parental distress and conflict.[6]

Genetic counselling is the process by which individuals or families at risk of inherited disorders are informed about the nature of the condition, its transmission, and the available options for management and reproductive planning.[7] Genetic counselling is one of the most efficient methods for preventing genetic diseases and birth defects. Counselling promotes informed decision-making, especially when combined with prenatal or preimplantation diagnostic techniques.[8] Facilities for such diagnostics, along with the willingness to undergo medical termination in cases of severe anomalies, can greatly reduce the incidence and severity of congenital disorders.[9]

Parental attitudes are crucial, and some may discontinue reproduction despite having no recurrence risk, while others with a high risk continue having children without adequate knowledge of available tests and interventions.[10] Parental education, cultural beliefs, and social stigma significantly influence reproductive decisions.[11] In regions with high consanguinity or poor health literacy, the risk of unrecognised hereditary conditions increases.[12]

Objectives

This study aimed to assess parental awareness of genetic counselling, identify the types of congenital anomalies along with their associated risk factors, and contribute to the reduction of the incidence and severity of genetic disorders and birth defects.

MATERIALS AND METHODS

This descriptive study was conducted on 182 neonates with congenital anomalies and genetic disorders who attended the Department of Paediatrics at Mahatma Gandhi Memorial Government Hospital (MGMGH), Tiruchirappalli, India, over 18 months from February 2018 to July 2019. This study was approved by the institutional ethics committee, and informed consent was obtained before the study initiation.

Inclusion Criteria and Exclusion Criteria

The study included all newborns with a gestational age >24 weeks and congenital anomalies. Stillbirths, neonates born before 24 gestational weeks, those without identifiable congenital anomalies or genetic disorders, those with incomplete clinical data, and those in which informed consent could not be obtained from the parents were also excluded.

Methods

Detailed maternal history and thorough physical examinations were conducted for all neonates suspected of having congenital anomalies. Appropriate imaging studies, including echocardiography, ultrasonography, and chest radiography, along with other relevant investigations, were performed to aid diagnosis. The findings were correlated with potential risk factors to identify those contributing to congenital anomalies. Each anomaly was classified as single or multiple, lethal or non-lethal, genetic or multifactorial, and further categorised system-wise to determine the most prevalent types.

Follow-up of the affected neonates was carried out through a dedicated genetic outpatient clinic conducted weekly in the newborn ward at MGMGH, Tiruchirappalli, India. The proportion of parents attending genetic counselling sessions was documented and analysed. To assess the level of awareness among parents regarding genetic counselling, a structured questionnaire was administered before and after counselling. The questionnaire encompassed three domains: knowledge of genetic diseases (including understanding genetic disorders, genes, congenital anomalies, and their inheritance patterns), access to and acceptability of genetic counselling services, and health care practices and management intentions.

The latter included aspects such as adherence to follow-up, future screening intentions, understanding of disease complications and treatment options, willingness for prenatal diagnosis and possible pregnancy termination, and awareness of the prognosis of lethal anomalies. All categorical data were summarised and presented as frequencies and percentages.

RESULTS

Male neonates constituted the majority (60%), followed by females (37%), and 3% were identified

as having ambiguous genitalia. In terms of parental education, the highest proportion of fathers had completed higher secondary education (37.9%), followed by graduates (34.1%), while only 0.5% were uneducated or had completed only primary school education. Similarly, among mothers, higher secondary education was the most common (34.1%), followed by graduates (23.6%) and those with high school education (22%). A minimal proportion of mothers were uneducated (1.6%) or had primary education (1.1%).

Most patients were Hindu (85.2%), followed by Christians (10.4%) and Muslims (4.4%). The age distribution of mothers showed that most were between 20 and 34 years (89%), while only 6% were under 19 years and 4.9% were 35 years or older. Regarding birth order, first-born (42.3%) and second-born (39.6%) neonates predominated, with fewer third-born (14.3%) and fourth-born (3.8%) neonates. Consanguinity was reported in 31% of the cases [Table 1].

Table 1: Sociodemographic characteristics.

		Frequency N (%)
Father's education	Uneducated	1(0.5%)
	Primary school	1(0.5%)
	Middle school	19(10.4%)
	High school	30(16.5%)
	Higher Secondary	69(37.9%)
	Graduate	62(34.1%)
Mother's education	Uneducated	3(1.6%)
	Primary school	2(1.1%)
	Middle school	32(17.6%)
	High school	40(22%)
	Higher Secondary	62(34.1%)
	Graduate	43(23.6%)
Religion	Hindu	155(85.2%)
	Muslim	8(4.4%)
	Christian	19(10.4%)
Mother's age group (years)	≤ 19	11(6%)
	20–34	162(89%)
	≥ 35	9(4.9%)
Birth order	1st	77(42.3%)
	2nd	72(39.6%)
	3rd	26(14.3%)
	4th	7(3.8%)

The majority of neonates (50%) weighed between 1501 and 2500 g, with 41.8% weighing >2500 g; no birth weights were recorded ≤1000 g. Most deliveries occurred between 34 and 36 weeks of gestation (74.2%), whereas term births (37–41 weeks) were rare (1.1%).

Pregnancy-induced hypertension (PIH) was present in 4.9% of patients, while maternal hypothyroidism and gestational diabetes mellitus (GDM) were present in 9.9% of patients. A total of 99.5% of mothers reported no exposure to teratogenic drugs. Similarly, maternal infections occurred in only 1.6% of cases, and only 95.6% had a similar illness in their family.

Antenatal anomaly scans were conducted in 68.1% of pregnancies, with anomalies diagnosed antenatally in 26.9% of cases. Only a few patients had undergone abortions before, i.e. 13.7% with one abortion and 2.7% with two abortions. Post-conception folic acid supplementation was the most common at 46.2%, followed by irregular intake (26.4%), preconception intake (13.2%), and 14.3% of mothers who did not take folic acid at all [Table 2].

Table 2: Distribution of perinatal and maternal clinical characteristics in the study population.

		Frequency N(%)
Birth weight (gm)	≤ 1000	0
	1001 to 1500	15(8.2%)
	1501 to 2500	91(50%)
	> 2500	76(41.8%)
Gestational age	28 to 31 weeks + 6 days	14(7.7%)
	32 to 33 weeks + 6 days	31(17%)
	34 to 36 weeks + 6 days	135(74.2%)
	37 to 41 weeks + 6 days	2(1.1%)
Pregnancy-induced hypertension (PIH)	No	173(95.1%)
	Yes	9(4.9%)
Gestational diabetes mellitus (GDM)	No	164(90.1%)
	Yes	18(9.9%)
Maternal hypothyroidism	No	164(90.1%)
	Yes	18(9.9%)
Teratogenic drug exposure	No	181(99.5%)
	Yes	1(0.5%)

Antenatal anomaly scan	No	58(31.9%)
	Yes	124(68.1%)
Folic acid intake	Preconception	24(13.2%)
	Post conception	84(46.2%)
	Irregular intake	48(26.4%)
	Not taken	26(14.3%)
Anomalies diagnosed antenatal	No	133(73.1%)
	Yes	49(26.9%)

Cardiovascular system (CVS) malformations were the most common congenital anomalies, present in 42.9% of cases, with acyanotic lesions (64.1%) occurring more frequently than cyanotic lesions (35.9%). Multiple anomalies were high (31.4%), and gastrointestinal (GIT) malformations were reported in 17% of neonates, followed by limb anomalies and syndromic associations (9.3% each). Dysmorphic facies, brain malformations, pulmonary anomalies, and oral cavity anomalies were each identified in 8.2% of patients.

Spinal and genitourinary anomalies were detected in 5.5% of patients, while renal malformations accounted for 4.9%. Sequences were identified in 2.7% of cases, and both eye anomalies and congenital skin defects were observed in 2.2% of cases. The least frequent anomalies were ear anomalies and inborn errors of metabolism (IEM), each reported in only 1.6% of cases [Table 3].

Table 3: Distribution of congenital anomalies and associated conditions in neonates

		Frequency N(%)
Dysmorphic facies	No	167(91.8%)
	Yes	15(8.2%)
Brain malformations	No	167(91.8%)
	Yes	15(8.2%)
Spine malformations	No	172(94.5%)
	Yes	10(5.5%)
CVS malformations	No	104(57.1%)
	Yes	78(42.9%)
Type of CVS malformations	Acyanotic	50(64.1%)
	Cyanotic	28(35.9%)
Pulmonary malformations	No	167(91.8%)
	Yes	15(8.2%)
Git malformations	No	151(83%)
	Yes	31(17%)
Renal malformations	No	173(95.1%)
	Yes	9(4.9%)
Genitourinary anomalies	No	172(94.5%)
	Yes	10(5.5%)
Limb anomalies	No	165(90.7%)
	Yes	17(9.3%)
Eye anomalies	No	178(97.8%)
	Yes	4(2.2%)
Oral cavity anomalies	No	167(91.8%)
	Yes	15(8.2%)
Ear anomalies	No	179(98.4%)
	Yes	3(1.6%)
Congenital skin defects	No	178(97.8%)
	Yes	4(2.2%)
Syndrome	No	165(90.7%)
	Yes	17(9.3%)
Sequence	No	177(97.3%)
	Yes	5(2.7%)
Inborn errors of metabolism (IEM)	No	179(98.4%)
	Yes	3(1.6%)
Multiple anomalies	No	125(68.6%)
	Yes	57(31.4%)

Genetic counselling attendance was the most frequent event, with 95.6% of patients receiving counselling, reflecting excellent accessibility and acceptance of this service. Child survival highlights a concerning mortality rate of 35.2%. Follow-up adherence was recorded in only 55.5% of the cases. Only 4.4% of the population did not attend genetic counselling, making it the least frequent occurrence.

Pre-counselling awareness had the majority scoring in the moderate range (8–12) (53.4%), followed by scores below seven (34.5%). Post-counselling awareness showed a marked improvement, with the majority of parents scoring the maximum awareness score of 13 (81%).

Table 4: Assessment of genetic counselling impact, neonatal survival, and parental awareness

Variable		Frequency N(%)
Genetic counselling attended	Yes	174(95.6%)
	No	8(4.4%)
Child survival	Yes	118(64.8%)
	No	64(35.2%)
Follow-up status	Yes	101(55.5%)
	No	81(44.5%)
Awareness (Pre-counselling) (N=174)	Score < 7	60(34.5%)
	Score 8 to 12	93(53.4%)
	Score 13	21(12.1%)
Awareness (Post-counselling) (N=174)	Score 8 to 12	33(19%)
	Score 13	141(81%)

DISCUSSION

In our study, of the 182 babies delivered, 64 (35%) did not survive, representing nearly one-third of the study population with anomalies. Similar to this, Sachdeva et al. reported that mortality rates were higher among newborns with congenital anomalies (17.35%) compared to those without (0.34%) per 1000 births.[13]

In our study, consanguinity was a common risk factor (31% of cases). The majority of neonates with anomalies were first- or second-born (42.3% and 39.6%), while third- and fourth-born neonates comprised 14.3% and 3.8%, respectively. Similarly, Taksande et al. observed parental consanguinity in 14 congenital malformation cases, with most affected infants being first-born (34.63%) or second-to-third-born (51.95%), and fewer fourth-born (13.40%).[14] Stein et al. reported that consanguinity was found in 26 of 1093 cases (2.38%).[15]

In our study, preconception folic acid intake was low (13.2%), while 46.2% took folic acid post-conception at around 8 weeks, 48 of whom took it irregularly; 14.3% did not take folic acid at all. GDM and hypothyroidism each accounted for approximately 10% of the cases. Similarly, Zhao et al. reported in a meta-analysis that pregestational diabetes significantly increased the risk of major congenital malformations, with a relative risk of 3.83.[16] Grattan et al. found that maternal hypothyroidism increased the risk of congenital heart defects in offspring, with an adjusted odds ratio of 1.68.[17] In contrast, McNally et al. reported 64% preconception and 93% first-trimester folic acid use, highlighting the need for increased awareness and early supplementation education.[18]

The most common anomaly in our study was congenital heart disease (CHD), which constituted almost 43% of all anomalies, with 27.5% (50/78) acyanotic and 15.5% (28/78) cyanotic. The overall CHD incidence was 14 per 1000 live births. Similarly, Saxena et al. documented 8.07 per 1000 live births with significant CHDs, of which 79.9% were acyanotic and 20.1% cyanotic. Supporting that CHDs, particularly acyanotic types, emerge as the most prevalent congenital anomaly with a significant mortality impact, reflecting global and regional variations.[19] Additionally, Bhide et al. reported a prevalence of major congenital anomalies of 230.51 per 10,000 births, with CHDs being the most frequent at 65.86 per 10,000.3 Van der Linde et al. reported that Asia's highest CHD birth prevalence was 9.3 per 1000 live births ($p < 0.001$), followed by Europe at 8.2 per 1000 live births.[20]

In our study, GIT anomalies constituted 17% (31 cases) with an incidence of 5.5 per 1000 live births. Similarly, Narmadha et al. reported GIT anomalies in 15.13% of 152 cases, reinforcing consanguinity and lower birth order as key risk factors. Limb anomalies were the third most common, at 9.3%, with an incidence of 3 per 1000 live births, and genitourinary malformations accounted for 5.5%.[21] Consistent with this, Radojević et al. found that limb anomalies comprised 26.67% of skeletal defects in 580 foetal and neonatal autopsies.[22] Similarly, Shrestha et al. identified genitourinary anomalies as the most frequent (24.2%) among 66 congenital malformations in 2,456 live births, followed by musculoskeletal (21.2%) and cardiovascular anomalies (18.2%).[23]

In our study on awareness, 174 parents completed pre- and post-counselling questionnaires; scores >13 increased from 12% to 81% post-counselling, demonstrating marked improvement. Of all parents, 95.6% attended genetic counselling. Supporting this, Brueckner demonstrated significant patient empowerment post-counselling, with GCOS-24 scores increasing by 10.1 points (effect size d=0.72).[24] Glynn et al. reported a genetic counselling utilisation of 20%, consistent across all categories and regions, but significantly higher among high-need cases, stillbirths, postnatal deaths, and tertiary hospital births, with utilisation rising from 39.7% in 1991 to 48.4% in 2004.[25] Additionally, Alotaibi found that 73.2% had a good understanding of genetic disorders.[26] Fitzgerald-Butt et al. reported a mean genetic knowledge score of 73.8%, with higher education tripling the likelihood of better knowledge.[27]

Genetic counselling significantly enhances parental awareness and empowerment regarding congenital anomalies. Consanguinity and lower birth orders remain important risk factors. The prevalence of CHDs and limb, GIT, and genitourinary anomalies was consistent with regional and global data. Higher education and regular follow-up promote better genetic knowledge and support. These findings highlight the need to integrate routine genetic

counselling and prenatal diagnostic services into maternal and child healthcare frameworks, particularly in resource-limited locations, to reduce the incidence and consequences of congenital disorders.

Limitations

As a hospital-based study, the findings may not reflect the true population-level prevalence of congenital anomalies. Advanced genetic investigations such as FISH, CGH, chromosomal microarrays, and MLPA were not performed in all cases due to cost constraints and limited availability. Karyotyping was conducted only in selected cases. The exclusion of stillbirths may have led to an underestimation of the overall burden of congenital anomalies. Furthermore, follow-up data were not available for families of neonates who did not survive.

CONCLUSION

The study concluded that genetic counselling enhances parental understanding and improves the management of congenital anomalies. While congenital heart defects were the most common anomalies observed, several risk factors, such as consanguinity, maternal health conditions, and inadequate folic acid supplementation, were also associated with these conditions.

These findings highlight the key role of integrating genetic counselling into routine prenatal and perinatal care. It not only provides families with knowledge but also supports better decision-making and adherence to follow-up care. Therefore, the widespread implementation of genetic counselling and prenatal diagnostic services is essential to reduce the burden of congenital and genetic disorders, particularly in resource-limited settings such as ours.

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