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A case series on congenital anomaly of fetus and its outcome

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Abstract
Aim: To study the congenital anomalies of foetuses over a period of 1 year (2023-2024) at ESI-PGIMSR & ESIC Medical College, Joka.
Materials and Methods: This prospective study followed a series of 13 pregnancies. Case ascertainment was done through radiological and clinical examinations. Appropriate radio-diagnostic investigations were done to confirm the internal anomalies of MTPs.
Results and Analysis: The study included 2057 pregnancies were admitted in our institution, out of which 13 had congenital malformations, with a prevalence of 0.63%. Among the pregnancies there were 2 vaginal deliveries, 4 Caesarean sections and 7 MTPs. 4 anomalies were identified after birth. The other 9 anomalies were identified by ultrasonography during the antenatal period.
Conclusion: Good health education, proper antenatal care, indicated prenatal tests and strong preventive care will decrease the incidence of congenital anomalies.

Keywords: Congenital anomaly, pregnancy, caesarean section, anencephaly, termination of pregnancy.

Introduction

According to WHO, congenital malformations can be defined as structural or functional anomalies that occur during intrauterine life. They are also known as birth defects, congenital anomalies or congenital malformations. These conditions develop prenatally and may be identified before or at birth, or later in life. Roughly 6% of all neonates globally are born with congenital malformations, leading to hundreds of thousands of related fatalities [1]. However, these figures might be underestimated as they often exclude terminated pregnancies and stillbirths.

Some congenital anomalies are lethal and are incompatible with life while some like cleft lip and cleft palate, clubfoot or hernias can be addressed through surgical or non-surgical interventions. However, others such as heart defects, neural tube defects, and Down's syndrome can lead to lifelong challenges. Treatment options and outcomes vary greatly depending on the specific disorder and individual circumstances.

According to WHO, an estimated 240,000 newborns die worldwide within 28 days of birth every year due to congenital malformations. Congenital disorders cause a further 170,000 deaths between the ages of 1 month and 5 years [2].

Congenital anomalies can lead to long-term disabilities, which can have profound effects on individuals, families, healthcare systems, and societies as a whole. The impact extends beyond just the affected individuals, influencing various aspects of life and requiring comprehensive support systems to address the diverse needs arising from these conditions.

Identifying the exact causes of congenital disorders can be challenging because they may result from a combination of genetic, infectious, nutritional, or environmental factors. Pinpointing the precise cause often requires extensive medical evaluation and research, and even then, it's not always possible to determine with certainty. This complexity underscores the importance of ongoing research and preventive measures to mitigate the risks associated with these disorders.

Some congenital anomalies are preventable by pre conceptional counselling, ensuring adequate intake of essential nutrients like folic acid and optimum maintenance of blood glucose levels. Providing proper care before and during pregnancy, including regular prenatal check-ups and avoiding harmful substances, also plays a crucial role in preventing congenital malformations. These preventive measures are essential components of public health efforts aimed at reducing the incidence of these conditions.

Materials and Methods

A prospective observational study was carried out in the Department of Obstetrics and Gynecology at ESI-PGIMSR & ESIC Medical College, Joka for a period of 1 year from February 2023 to March 2024. The hospital provides medical care to all ESI employees who were booked cases at ESIC Joka or referred from other ESIC centers.

After anomaly scans, detailed patient history was obtained and guidelines set by the government were strictly followed for terminating the pregnancy. MTP was done with Mifepristone and Misoprostol. Detailed antenatal and maternal histories, including factors like age, parity, consanguinity, familial history, and gestational factors were obtained through prenatal interviews. The newborns were examined by a paediatrician and diagnosis was confirmed by radiography and ultrasonography.

Results

During the study, 2057 pregnancies were admitted in our institution, out of which 13 had congenital malformations, with a prevalence of 0.63%. Among these pregnancies, 2 were vaginal deliveries, 4 Caesarean sections and 7 medical terminations of pregnancy. 4 anomalies were identified after birth. The other 9 anomalies were identified by ultrasonography.

The systems involved in our study were Central nervous system (CNS) (23.08%), gastrointestinal system (GI) 15.38%, musculoskeletal system (15.38%), cardiovascular system (CVS) (7.69%), Multiple congenital anomalies were seen in 23.08% of the cases which included CNS, CVS, respiratory, genitourinary systems. Others (15.38%) included cleft palate and a case of non-immune hydrops fetalis for which the exact cause couldn't be assessed.

8 patients were primigravida, 2 were second gravida (15.38%), 1 had previous history of MTP (7.69%), the other 2 had previous history of spontaneous abortions (15.38%). In this study, congenital anomalies were found more in primiparas (84.62%). The prevalence of congenital anomalies was found more in the maternal age group of 21-30 years (84.61%), and (7.69%) for those aged more than 30 years. Out of 6 babies born, 33.33% had low birth weight with an equal male to female ratio.

Table 1: General characteristics of pregnant women whose foetuses had congenital malformations.

Age (years)	Frequency	Percentage %
21-30	11	61.54
31-35	1	7.69
>35	1	7.69
Socioeconomic Status		
Low	1	7.69
Middle	11	61.54
High	1	7.69
Residence		
Urban	11	61.54
Rural	2	15.38

Table 2: Characteristics of pregnant women whose foetuses had congenital malformations.

Variables			Percentage %
Maternal age	20-30years	11	84.62
	>30 years	2	15.38
Parity	Primipara	11	84.62
	Multipara	2	15.38
Antenatal care	Booked cases	11	84.62
	Unbooked cases	2	15.38
Routine IFA intake	Yes	10	76.92
	No	3	23.08
Mode of delivery	Vaginal delivery at term	2	15.38
	Caesarean section	4	30.77
	MTP delivered vaginally	7	53.85
Antepartum conditions	No comorbidity	8	61.54
	Hypertension	1	7.69
	Diabetes mellitus	1	7.69
	GDM	2	15.38
	Pre-eclampsia	1	7.69
Birth weight	Low birth weight	2	33.33
	Normal	4	66.67

Table 3: General characteristics of baby born with congenital anomaly

Variables		Frequency	Percentage %
Gender	Male	3	50
	Female	3	50
Maturity	Preterm	1	16.67
	Term	5	83.33
Mode of delivery	Caesarean	4	66.67
	Vaginal delivery	2	33.33
Outcome	Discharged	5	83.33
	Died after discharge	1	16.67

Table 4: System distribution of congenital Anomaly

System	Number	Percentage %
Central nervous system	3	23.08
Gastrointestinal system	2	15.38
Musculoskeletal system	2	15.38
Cardiovascular system	1	7.69
Multiple	3	23.08
Others (cleft palate, non-immune hydrops)	2	15.38

Discussion

The prevalence of congenital anomalies found in our study was 0.63% (including live births and abortions), which is lower in comparison to most of the other studies done in India. Prajkta Bhide *et al*., documented the prevalence of prenatal diagnosis of congenital anomaly was 10.98 per 1000 births and the congenital anomaly termination of pregnancy rate was 4.39 per 1000 births [3]. Chaturvedi P *et al*., and Taksande A *et al*., reported an incidence of 2.72% [4]. and 1.9% [5]. respectively.

Different studies show different frequencies of congenital malformations [6, 7, 8].

In our study, the systems involved were central nervous system (CNS) (23.08%), gastrointestinal system (GI) 15.38%, musculoskeletal system (15.38%) and cardiovascular system (CVS) (7.69%). Multiple congenital anomalies were seen in 23.08% of the cases which included CNS, CVS, respiratory and genitourinary system. Others (15.38%) include cleft palate and a case of non-immune hydrops fetalis for which the exact cause couldn't be assessed. Some studies had a higher incidence of CNS malformation followed by GIT and musculoskeletal system [8, 9]. In our study, CNS and multiple system involvement were more common, followed by musculoskeletal system, GI system and CVS. Other studies like Anuja Bhalerao *et al*., reported musculoskeletal system as the most common system affected [6], Sugna *et al*., [10]. reported GI malformation as the most common one.

In our study, the 3 cases of CNS malformation were anencephaly, hydrocephalus and hydranencephaly, each having a history of inadequate intake of folic acid. Another CNS anomaly was alobar holoprosencephaly with proboscis (Figure 1) with respiratory and cardiac malformation was diagnosed and terminated in the second trimester when the mother had LRTI with fever. Anencephaly and alobar holoprosencephaly was diagnosed in the second trimester anomaly scan and was terminated as it was incompatible with life.

Anencephaly is a severe malformation of the central nervous system (CNS), being the most common type of neural tube defect. It represents the total or partial absence of the calvarium with absence of the brain. The brainstem, cerebellum and diencephalon are usually present [11]. Overall estimate of the prevalence, incidence and attenuation of anencephaly worldwide were 5.1 per ten thousand births (95% confidence interval 4.7-5.5 per ten thousand births), 8.3 per ten thousand births (95% confidence interval 5.5-9.9 per ten thousand births), 5.5 per ten thousand births (95% confidence interval 1.8-15 per ten thousand births) respectively [12].

It has been classified as one of the most lethal congenital defects, with a first-year mortality rate of 100% [13]. Male: Female ratio being 3:1. Irrespective of POG, termination of pregnancy is recommended.

The male baby with hydrocephalus (head circumference >97th centile) who was delivered by LSCS due to CPD was diagnosed as Aqueduct of Sylvius stenosis and was planned for ventriculoperitoneal shunt procedure. Hydrocephalus, first described by Hippocrates as early as the fifth century BCE, is an abnormal accumulation of cerebrospinal fluid (CSF) within the ventricles of the brain either due to insufficient CSF reabsorption or CSF overproduction [14]. It is a significant public health concern estimated to affect 380,000 new individuals annually [15].

Hydranencephaly is a rare congenital post-neurulation disorder that occurs during the second trimester characterized by the destruction of the cerebral hemispheres, which are replaced with a membranous sac filled with cerebrospinal fluid (CSF) [16, 17]. The cranial vault and meninges are intact. It is most commonly caused by a vascular insult involving the anterior circulation. Midbrain structures such as the basal ganglia, brainstem, and posterior fossa structures are present [18].

The male baby with hydranencephaly (Figure 2,3) was delivered vaginally after CSF drainage (per vagina). The baby was referred to a higher centre but due to poor prognosis no further intervention was done. It is a rare condition and is rarely encountered nowadays due to therapeutic abortions. The incidence may vary from 1 in 10,000 to 1 in 5,000 (0.01%-0.02%) of pregnancies [19, 20, 21]. There is no difference in the incidence between males and females.

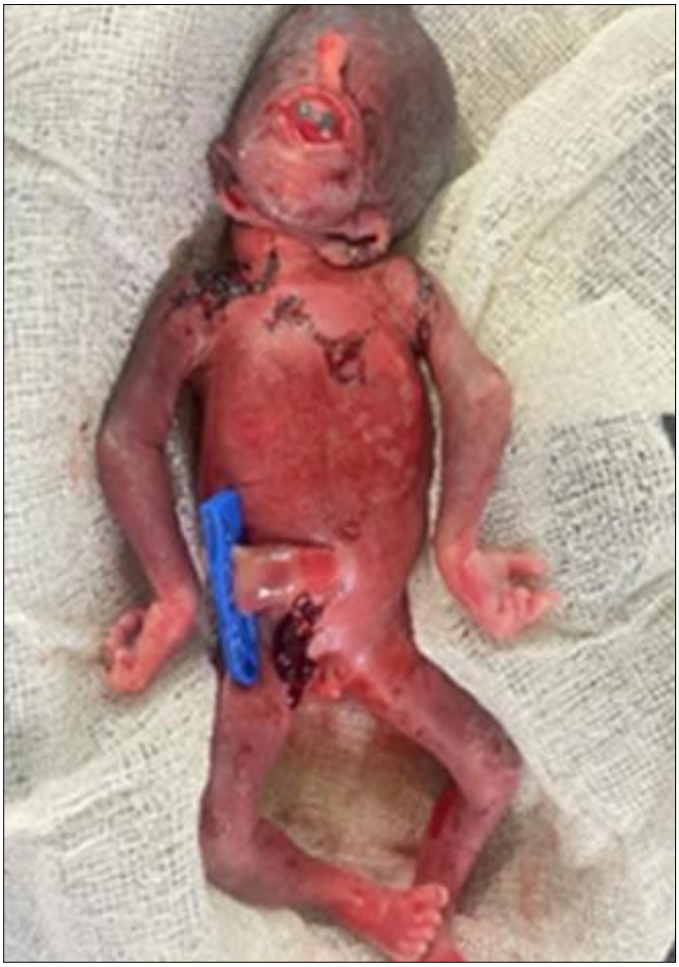

Fig 1: Alobar holoprosencephaly with proboscis.

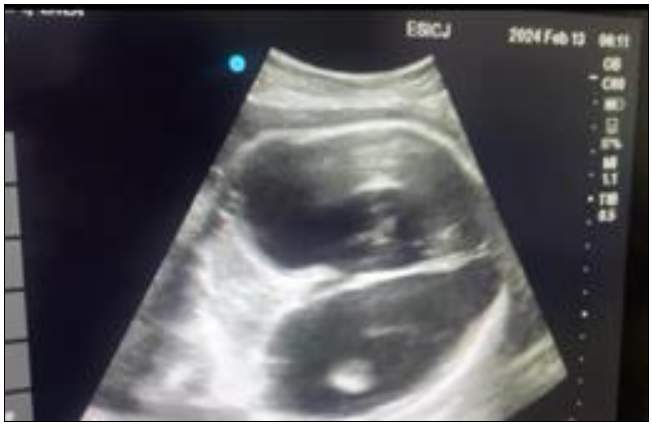

Fig 2: Maternal USG showing hydranencephaly.

Fig 3: Baby born with hydranencephaly.

One baby was noted with cleft palate which was diagnosed after birth in a primigravida with IUGR who was delivered by LSCS. It occurs due to the failure of fusion of the palatal shelves of the maxillary processes, resulting in a cleft of the hard and/or soft palates [22]. Clefts arise during the fourth developmental stage. Exactly where they appear is determined by locations at which fusion of various facial processes failed to occur, this in turn is influenced by the time in embryologic life when some interference with development occurred [23]. Overall incidence of cleft lip and palate is approximately 1 in 600 to 800 live births (1.42 in 1000) and isolated cleft palate occurs approximately in 1 in 2000 live births [24].

The 2 cases of GIT malformations were duodenal atresia and rectal duplication. The female baby who was born with duodenal atresia (Figure 4) was diagnosed after birth in a post CS mother who presented with polyhydramnios. Her anomaly scan was normal. LSCS was done in view of IUGR and polyhydramnios. The baby underwent surgical correction but unfortunately succumbed to dyselectrolytemia and sepsis 2 weeks later.

Duodenal atresia occurs in 1 in 5000 to 10,000 live births. It is often associated with other anomalies, including trisomy 21/Downs syndrome and cardiac malformations. Approximately 30-40% of children with duodenal atresia have Down's syndrome. There is a 3% prevalence of congenital duodenal atresia among patients with trisomy 21/Down's syndrome. There is no difference in prevalence between the genders. There is an association with VACTERL, annular pancreas, and other bowel atresias like jejunal atresia, ileal atresia, and rectal atresia [25]. Prognosis after successful surgical treatment of duodenal atresia is excellent [26]. A study following infants from 1972 to 2001 demonstrated late complications in up to 12% of patients and late mortality of 6% [27].

A girl baby was born with rectal duplication to a primipara mother. This was diagnosed after birth, the baby underwent sigmoid loop colostomy (Figure 5). Duplicate rectal cysts are the least common among gastrointestinal congenital cysts, forming only 4% of them [28].

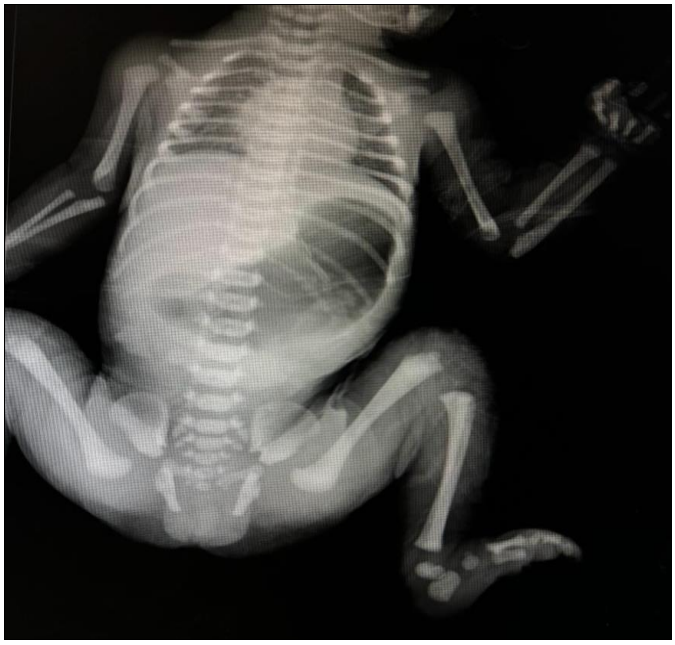

Fig 4: X Ray of the baby born with duodenal atresia.

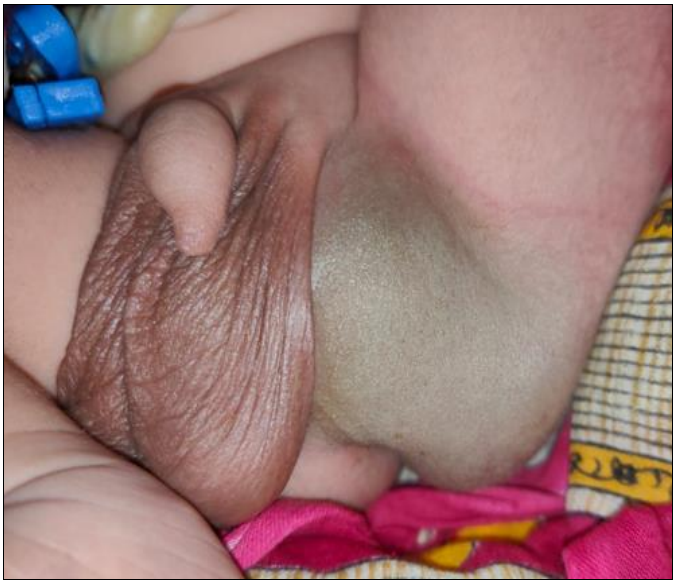

Fig 5: Baby born with rectal duplication.

The 2 cases that involved the musculoskeletal system were fetal skeletal dysplasia (Figure 6) which were diagnosed during anomaly scans and MTP was done in the second trimester. It is caused by an autosomal dominant mutation in FGFR 3 gene [29, 30]. The other female baby was delivered through LSCS due to thick MSL and oligohydramnios with congenital bilateral dislocation of knee (Figure 7). The condition was conservatively managed by cast application which gradually corrected. Congenital dislocation of the knee (CDK) is a rare condition with an approximate incidence of 1 in 100,000 live births [31]. Factors such as lack of intrauterine space, oligohydramnios or breech presentation, alone or in combination with knee abnormalities have been considered as a possible cause of CDK [32, 33].

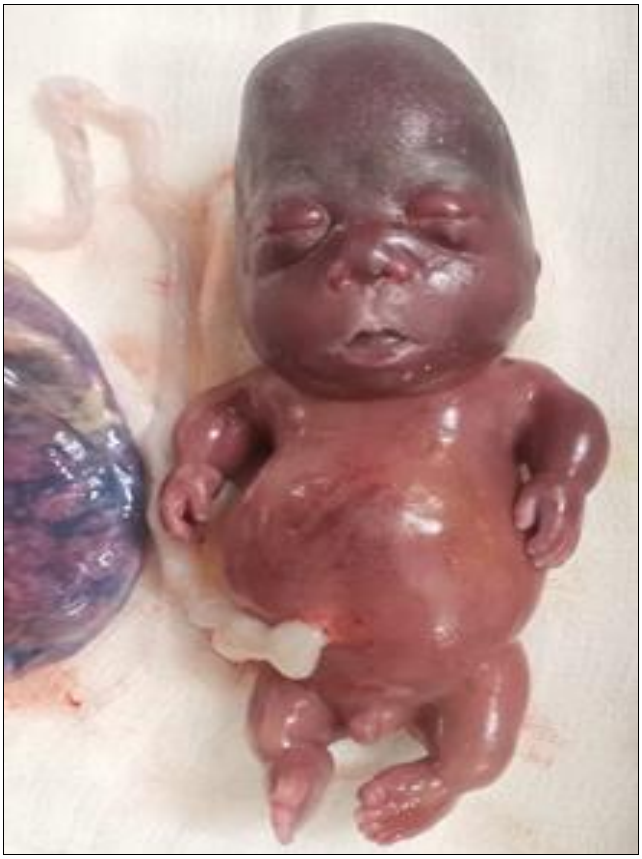

Fig 6: MTP done in second trimester due to anomaly scan showing skeletal dysplasia.

Fig 7: Baby born with Congenital bilateral dislocation of knee.

The MTPs done in second trimester were in a G2P0A1 overt diabetic patient with anomaly scan suggestive of atrioventricular septal defect with partial anomalous pulmonary venous connection, the other was a non-immune hydrops in a Rh positive mother where the fetus karyotyping was found to be normal. 2 other MTP were done which included multiple system involvement (CVS, genitourinary and musculoskeletal) in primigravida.

In our study, we found a higher incidence of congenital anomalies with maternal age of 21-30 years which is similar to a study conducted by Anuja Bhalerao *et al*. [6] Suguna Bai *et al*. [10] had a higher incidence of congenital malformations in mothers aged >35years. Dutta *et al*. [9] suggested that there is an insignificant association of increased maternal age with congenital anomalies. Among the babies born live, equal number of males and females was seen in our study whereas, male preponderance was seen more in other studies [4, 6, 34]. In our study, there was a higher incidence in primiparas, and the association of low birth weight babies with increased risk of congenital malformation was not appreciable whereas, the study conducted by Mohanty C *et al*. [34] suggested otherwise. There were no consanguineous marriages in our study. Good health education, proper antenatal care, indicated prenatal tests and strong preventive care will decrease the incidence of congenital anomalies.

Conclusion

The study highlights the prevalence of CNS anomalies, highlighting the involvement of multiple systems. It also highlights a higher association of congenital anomalies with primiparous mothers and those aged between 21 to 30 years. Socioeconomic status, comorbidity of the mother, history of consanguinity, elderly mothers, family history of congenital anomaly can contribute to increased risk of congenital malformations in the foetus. Pre-pregnancy scheduled dose of folic acid supplementation, proper pre-conceptional counselling, regular antenatal visits and prenatal investigations are advisable for prevention and early detection of anomalies. Adopting new modalities like pre-implantation genetic counselling, maternal cell-free DNA testing and the adopting inverted pyramid of care35 can prevent morbidity and mortality of the mother.

Abbreviations

MTP: Medical termination of pregnancy, LRTI: Lower respiratory tract infection, POG: period of gestation, LSCS: Lower segment cesarean section, CPD: Cephalo pelvic disproportion, IUGR: intrauterine growth retardation, CS: Cesarean section, MSL: meconium-stained liquor, Vacterl: Vertebral, anorectal, cardiac, trachea esophageal, renal anomaly, limb anomaly

Conflict of Interest:
Not available

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Not available

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