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Original Research Article

Magnitude, risk factors and immediate outcome of external congenital anomalies in neonates in government Cuddalore medical college and hospital: an observational study

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ABSTRACT

Background: Despite the enormous incidence of congenital malformations in developing countries, there are presently few thorough data on these disorders because there are no birth defect registries. This study was conducted with objectives to determine the magnitude, risk factors and outcomes of external congenital anomalies in neonates born in government Cuddalore medical college and hospital.
Methods: The present study is an observational study. All the neonates born during the study period were included in our study and risk factors and outcome of 201 babies born with external congenital anomalies were analyzed in detail.
Results: The incidence of external congenital anomalies is 5.68% with 33% having major and 63% having minor anomalies. Among the major anomalies cleft lip and/or palate is the most common anomaly (5%) in our study. Overall sacral dimple is the most commonly observed external congenital anomaly (9.50%). Four-fifths of the newborns with external congenital anomalies were discharged. About 13% of the newborns with congenital anomalies expired.
Conclusions: A comprehensive package that includes preventive services, diagnostic, surgical or medical intervention, financial assistance, counselling, and psychosocial support, as well as follow-up treatments like rehabilitation, is required in combating the incidence of congenital anomalies.

Keywords: External congenital anomalies, Magnitude, Risk factors, Outcome

INTRODUCTION

Congenital abnormalities (CA) are anatomical, behavioral, functional, and metabolic conditions that develop in utero and can be detected during pregnancy, at birth, or later in infancy. Congenital defects increase the risk of morbidity and mortality in newborns. Major CA are thought to affect 7.9 million newborns annually.[1] Global newborn mortality as a percentage caused by CA increased from 3% in 2008 to 4.4% in 2013.[2]

Major CAs, which occur in 2-3% of live births and 20-30% of stillbirths, are anomalies that have a considerable impact on life expectancy.[3] Due to complex interactions between genetic and environmental factors, incidence varies over time and geographic regions.[4] They represent between 15 and 30 percent of pediatric hospitalizations and roughly 3 percent of live births in the United States.[5]

In studies that have concentrated on externally anomalies, the musculoskeletal system is most frequently afflicted.[4,7] The digestive and cardiovascular systems have hitherto dominated investigations.[8] The gastrointestinal system has been the most commonly reported in earlier research in low-income countries.[8]

Birth deformities have a multifactorial etiology. These elements may have genetic (10-30%), environmental (5-

10%), or multifactorial inheritance (2035%) origins; the remaining (30-45%) origins are unknown.[3] In LMICs, infectious pathogens appear to be the most significant environmental component. Age, lifestyle, pregnancy-related disorders, medication use, and peri-conceptional folic acid use are all implicated maternal variables.[3,7] Other crucial elements in the etiology of CAs include parental consanguinity, prior miscarriages and stillbirths, and inheritable congenital illness.[7]

Major CA in LMICs have mortality rates of 20-85% (compared to fewer than 10% in high-income countries), and typically, newborns with CA have higher mortality rates than infants born normally.[9]Among CA fatalities, 95% occur in LMIC countries. Birth abnormalities contribute to 25.3 to 35.8 million disability-adjusted life years globally.[1]

Despite enormous incidence of congenital malformations in LMICs, there are presently few thorough data on these disorders because there are no birth defect registries. Due to a lack of proper surveillance at medical institutions, underreporting, inadequate diagnostic capability, and low awareness, the prevalence of CA is significantly underestimated in LMICs.[10]

Prevalence studies are required to establish baseline rates, show trends over time, and provide etiological hints.[6] To determine the relative morbidity and mortality of neonatal admissions with congenital defects against those with other acute conditions, outcomes of neonates with congenital anomalies should be studied. Thus, this study can help policymakers strengthen surveillance of these abnormalities and perhaps boost public understanding of how these anomalies affect total infant death.

METHODS

Study design

Cross-sectional study design was used.

Study duration

Study carried out from September 2022 to August 2023.

Study place

Study conducted at government Cuddalore medical college and hospital, department of paediatrics.

Target population

Neonates born during the study period in government Cuddalore medical college and hospital.

Inclusion criteria

All neonates who are born in government Cuddalore medical college and hospital are included for determining the magnitude. All neonates with external congenital anomalies are included for consideration of the risk factors and immediate outcomes were included in study.

Exclusion criteria

Still born and terminated pregnancies were excluded from the study.

Sampling procedure

Total population sampling procedure was used.

Ethics approval

Study was approved by the institutional ethics committee (IEC) of government Cuddalore medical college and hospital. Parents were informed about the purpose of the study. Written informed consent was obtained from parents. Participants were assured that the information obtained would be for research purposes and would therefore be anonymous and kept strictly confidential.

Data collection method

A questionnaire was administered to determine the risk factors in neonates with congenital anomalies. Careful methodical head to toe examinations of the neonates was then done. Follow-up of the above neonates was done till discharge/death.

Data collection instruments

Physical instruments

An infantometer for measuring baby length, a digital baby weighing machine to measure weight, inch tape for head circumference were used.

Pretested semi-structured questionnaire

It was used to collect data on socio-demographic details, risk factors, and natal histories.

Examinations

Head-to-toe examination for all external anomalies were done.

Statistical analysis

Data collected entered in MS excel and analyzed using SPSS 22 version. Prevalence is presented as proportions. Continuous values were given with mean and SD.

RESULTS

During the study period, a total of 3537 live births were delivered in government Cuddalore medical college and

hospital and all neonates were examined to detect external congenital anomalies.

Magnitude of the problem

Out of the total live births, 201 newborns had external congenital anomalies. The children born with external congenital anomalies among all the live births delivered for one year is 5.68%.

Among major anomalies, cleft lip and/or palate is most common anomaly (5%) and overall sacral dimple is the most commonly observed external congenital anomaly (9.50%).

Table 1: Proportion based on type of anomalies.

Type of anomalies	N	Percentage (%)
Major	67	33.3
Minor	127	63.2
Both	7	3.5

Table 2: Distribution of major external congenital anomalies.

Name of the anomaly	N	Percentage (%)
Cleft lip and cleft palate	10	5
Congenital talipes equinovarus	7	3.50
Macrocephaly	6	3
Congenital diaphragmatic hernia	5	2.50
Meningocele/meningomyelocele	5	2.50
Microcephaly	4	2
Pierre robbins syndrome	3	1.50
Ambiguous genitalia/congenital adrenal hyperplasia	3	1.50
Collodion baby	3	1.50
Downs syndrome	3	1.50
Imperforate anus/ high anorectal malformation	3	1.50
Hypospadiasis	2	1
Craniosynostosis	2	1
Edwards syndrome	2	1
Inguinal hernia	2	1
Non immune hydrops fetalis	2	1
Vacterl anomaly (Vertebral anomalies, anorectal anomalies (analatresia), cardiac anomalies, tracheoesophageal fistula, renal anomalies and limb anomalies)	2	1
Spina bifida	2	1
Corneal opacity	2	1
Buphthalmos	1	0.50
Anencephaly	1	0.50
Anovestibular fistula	1	0.50
Congenital cataract	1	0.50
Left macrostomia	1	0.50
Cystic hygroma	1	0.50
LAX abdomen	1	0.50
Epidermolysis bullosa	1	0.50
Genu recurvatum	1	0.50
Potter's syndrome	1	0.50
Choanal atresia	1	0.50
Omphalocele	1	0.50
Hypoplastic pectoralis major, Sprengel deformity of shoulder with spine deformity	1	0.50
Developmental dysplasia of hip	1	0.50
Skeletal dysplasia	1	0.50
Cutis Laxa, sutural diastasis	1	0.50
Joint laxity	1	0.50

Table 3: Distribution of minor external congenital anomalies.

Name of the anomaly	N	Percentage (%)
Sacral dimple	19	9.50
Preauricular tag	17	8.50
Single umbilical artery	16	8

Continued.

Name of the anomaly	N	Percentage (%)
Undescended testis	13	6.50
Polydactyly	9	4.50
Natal teeth	8	4
Pre auricular pit	7	3.50
Capillary hemangioma	6	3
Pigmented /hairy naevus	5	2.50
Low set ears	4	2
Single palmar crease	4	2
Midface hypoplasia	3	1.50
Amniotic band syndrome	2	1
Clinodactyly	2	1
Congenital hydrocele	2	1
Overlapping of digits	2	1
Plagiocephaly	2	1
Rocker bottom foot	2	1
Sacral tuft of hair	2	1
Tongue tie	2	1
Retrognathia	2	1
Short limbs	1	0.50
3 umbilical artery+ 3 umbilical vein	1	0.50
Brachydactyly	1	0.50
Coloboma iris	1	0.50
Congenital torticollis	1	0.50
Hypertelorism	1	0.50
Left eye micro-ophthalmic	1	0.50
Micro penis	1	0.50
Portwine stain	1	0.50
Syndactyly	1	0.50

Distribution of risk factors among babies with external congenital anomalies

Lower segment caesarean section is the commonest mode of delivery among newborns with congenital anomalies (50.7%) closely followed by normal vaginal delivery (46.3%).

Table 4: Intra-natal history.

Distribution of intra-natal factors		N	Percentage (%)
Mode of delivery	Normal vaginal delivery	93	46.3
	Assisted vaginal delivery	6	3
	Lower segment caesarean section	102	50.7
Complications during delivery	No complications	159	79
	Birth asphyxia	26	13
	Meconium stained liquor	8	4
	Prolonged 2nd stage of labour	2	1
	Breech presentation	6	3

More than 79% of the newborns did not encounter any complications in the immediate postpartum period. Birth asphyxia was the commonly observed complication in 13% of the neonates.

The average birth weight of the neonates with congenital anomalies was 2.51 kg (SD=0.59 kg), whereas the mean length at birth was 49.38 cm (SD=0.1, 95) and mean head circumference was 32.90 cm (SD=1.69).

The distributions of the birthweight, length at birth, and head circumference follow a normal distribution.

The majority of the newborns had a birth length of 50 cm.

The APGAR score of the newborns was measured at 1 and 5 minutes of the birth and the minimum and maximum scores were 1 and 8 at the 1st minute respectively and 4 and 9 at the 5th minute respectively.

Number of neonates with APGAR score less than 5 at 1 minute was 25 (12.4%) and APGAR score less than 7 at 5 minutes was 17 (8.5%).

Among the neonates with external congenital anomalies, 40 (20%) neonates were preterm of which 7 (3.5%) were born with gestational age of 28 weeks or less amounting to extreme prematurity. The rest 161 (80%) were born at term and none post-term.

More than half of the newborns were born in a first pregnancy and O positive is the most common blood group among the mothers.

Table 5: Distribution of maternal factors.

Distribution of maternal factors		N	Percentage (%)
Obstetric h/of mother	Primigravida	114	56.7
	Multigravida	87	43.3
The blood group of mothers	A positive	20	10
	A negative	1	0.5
	B Positive	72	35.8
	B negative	2	2.5
	O positive	81	40.3
	AB positive	21	10.4
	AB negative	1	0.5

The age of the mother during the current pregnancy and body weight were found to be distributed normally.

Table 6: Distribution of maternal age and body weight of the mother.

Maternal factors	Min	Max	Mean	SD
Age of mother at current child's birth (In years)	18	38	24.97	4.04
Age of mother at marriage (In years)	17	36	21.72	2.95

Distribution of maternal age of mother is normally distributed.

Table 7: Distribution of maternal complications.

Category	Maternal complications	N	Percentage (%)
Systemic diseases	No complications	94	46.8
	Anaemia	24	11.9
	Hypothyroidism	30	14.9
	Gestational DM and type 2 DM	17	8.5
	Pregnancy hypertension and pre-eclampsia	18	9.0
	Obesity	6	3.0
	Cardiac complications	2	1.0
	Bronchial asthma	1	0.5
Pregnancy-related complications	Oligohydramnios	13	6.5
	Polyhydramnios	8	4.0
	Anhydramnios	1	0.5
	Previous bad obstetric history	6	3.0
	Short primi	1	0.5
Miscellaneous	Parotid cyst	1	0.5
	HBsAg positive	2	1
	Vulvovaginitis	1	0.5

Hypothyroidism was the common complication encountered followed by anemia, and systemic diseases like diabetes, hypertension, and obesity. Among the pregnancy-related complications, oligohydramnios was the commonly found complication in the antenatal period. Around 3% had previous bad obstetric history namely recurrent pregnancy loss in the past.

Table 8: Antenatal risk factors of the new-born.

Distribution of antenatal risk factors		N	Percentage (%)
History of previous abortion	Yes	24	11.9
	No	63	31.3
	Not applicable (Primigravida)	114	56.7
Previous live child present	Yes	78	38.8
	No	9	4.5
	Not applicable (Primigravida)	114	56.7
Consanguinity among the parents	No consanguinity	165	82.1
	Second-degree consanguinity	6	3
	Third-degree consanguinity	30	14.9
Family history of congenital anomalies	Positive	7	3.5
	No history	194	96.5

Outcome of the newborns with external congenital anomalies

More than four-fifths of the newborns with external congenital anomalies were discharged. About 13% of the newborns with congenital anomalies expired.

Table 9: Distribution of type of interventions among newborns with major external congenital anomalies.

Intervention	N	Percentage (%)
No intervention	1	1
Medical	8	11
Surgical	39	53
Expired	26	35

Among the major external anomalies, 26 (35%) of the newborns expired, 8 (11%) required medical intervention, 39 (53%) required surgical intervention and 1 (1%) required nil intervention. Among those who underwent medical intervention, 2 neonates were treated with pharmacological intervention namely steroid administration.12 neonates underwent surgical correction whereas 21 waiting for surgery.

Six underwent manipulation and fixation by splinting or plaster cast application.

Table 10: Distribution of type of interventions among the newborns with minor external congenital anomalies.

Intervention	N	Percentage (%)
No intervention	74	58.4
Medical	36	28.3
Surgical	16	12.6
Expired	1	0.7

Among the minor external anomalies, 1 (0.7%) of the newborns expired (due to extreme prematurity), 36 (28.3%) required medical intervention, 16 (12.6%) required surgical intervention and 74 (58.4%) required nil intervention. 16 neonates were waiting for surgery (hydrocele, tongue tie, amniotic band syndrome).

DISCUSSION

A cross-sectional study was conducted to assess the magnitude of external congenital anomalies in babies born in government Cuddalore medical college and hospital.

The magnitude of the problem

The 201 newborns had external congenital anomalies and the proportion is 5.68% which is around 568 per 10,000 live births. Among these 33.3% had major anomalies, 63.2% had minor anomalies and 3.5% had both anomalies. Among the major anomalies cleft lip and/or palate is the most common anomaly (5%) in our study. Overall sacral dimple is the most commonly observed external congenital anomaly (9.50%). Among the external congenital anomalies 3% had macrocephaly, 2.5% had congenital diaphragmatic hernia, 1.5% had ambiguous genitalia, 1.5% had Pierre Robin syndrome, 1.5% had collodion and Down's syndrome was 1.5% in our study.

According to Bhide et al cohort study, there were 1822 births overall, with a prevalence of 230.51 serious CA per 10,000 births.[13]

The incidence was higher in our study (568 per 10000 live births) compared to the above Cohort study. This may be because the above cohort study included only serious congenital anomalies whereas we conducted a study to assess both major and minor anomalies.

Sinha et al conducted an observational study on congenital anomalies in North India. The musculoskeletal system (52.2%), central nervous system (28.3%), and gastrointestinal system (26.1%) were the systems most commonly affected.[11] Cleft palate followed by CTEV is the most common congenital anomaly in our study whereas CTEV is the most common in Sinha et al study. But the incidence of cleft palate (6.4%) in Sinha et al study is similar to our study report.

Taye et al, conducted a study among children during 2010-2014 where they reported results similar to our study with oro-facial defects being highest congenital anomaly detected.[14]

Risk factors of the problem

This current study found no correlation between gender and a higher frequency of CBDs. In the Sinha et al study, male newborns showed a somewhat greater prevalence of deformity (p=0.064) Numerous Indian studies back up the results.[11] The fact that there are still some areas of India where female newborns are deemed undesired pregnancies and are terminated may help to explain the rising male preponderance. Sachdeva et al observed a higher frequency of deformity in female infants, which is contrary to our findings.[9] This study found no correlation between gender and a higher frequency of CBDs.

Around 50.7% were LSCS-delivered babies and 12.9% of babies had birth asphyxia, 3.9% had meconium-stained liquor, 1% had prolonged second stage of labor. Around 12.4 % had low APGAR at 1 min and 8.5% had low APGAR at 5 mins in the current study.

The mean maternal age of childbirth is 24.9±4.04 years and the mean mother's weight is 60.17±10.6 kg. Around 53.2% had maternal complications with hypothyroidism (14.9%) followed by anemia, GDM and PIH. Oligohydramnios was present in 6.5%, polyhydramnios was present in 4% and BOH was present in 3% in our study The present study shows 11.9% had previous h/o abortion, 17.9% had consanguineous marriage and 3.5% had a positive family history of congenital anomalies.

Sinha et al have shown that no antenatal visits (p=0.041), TORCH and/or VDRL positive (p=0.023), gestational diabetes mellitus (GDM) (p=0.007), hypertension (p=0.090), consanguinity (p=0.03), and no folic acid consumption (p=0.015) were significant variables linked to CBD.[11]

In our study 20% neonates were preterm of which extremely premature babies were 3.5%. Patel et al observed that the majority of newborns with CBD were above 2500 g (59%) and that their mean weights of 2352.49 g were adequate for gestational age.[15] Padmanabhan et al discovered that 77% of CBD neonates were born at term.[16] In contrast to Sarkar et al, Marwah et al showed a strong connection between abnormalities and prematurity and low birth weight.[17,18] Fetuses having various CA have a higher risk of preterm birth, according to research by Doddabasappa et al.[19]

Outcome

Among the major external anomalies (74), 26 (35%) of the newborns expired, 8 (11%) required medical intervention, whereas 39 (53%) required surgical

intervention and 1 (1%) required nil intervention in our study.

Among the minor external anomalies (127), 1 (0.7%) of the newborns expired (due to extreme prematurity), 36 (28.3%) required medical intervention, 16 (12.6%) required surgical intervention and 74 required nil intervention (58.4%) in our study.

Anane-Fenin et al conducted a study, where 236 newborns with congenital anomalies were admitted to the facility in South Africa in which 33.2% of newborns with congenital anomalies expired which is similar to the mortality among major external congenital anomalies in our study.[12]

Limitations

The odds ratio was not calculated as the risk factors on babies without congenital anomalies were not studied

Also, this is a single-centric study conducted in one tertiary care center so, this result can't be generalized to all socio-economic populations**.**

CONCLUSION

The magnitude of congenital anomalies and risk factors in our study highlights the necessity for a birth defect surveillance system. A comprehensive package that includes preventive services, diagnostic, surgical or medical intervention, financial assistance, counselling, and psychosocial support, as well as follow-up treatments like rehabilitation, is required in combating the incidence of congenital anomalies.

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