STUDY OF CONGENITAL MUSCLES AND SKELETON DEFORMITIES EXAMINATION

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ABSTRACT

Background: In India congenital malformations have emerged as the third commonest cause ofperinatalmorbidityandmortality,nextonly to malnutrition andinfections.

Aim: Toknow thefrequency of congenitalmusculoskeletal anomaliesin neonates.

Methods: The present study was undertaken by collecting the data from the Department of Anatomy, PES institute of Medical Sciences and Research, Kuppam, Andra Pradesh, India, for a period of one year fromOctober2021 to October 2022, during which 650new born neonates were examined which included live birthsand still births. All birth safter 28 completed weeks were examined forcong enitalmusculosk eletalanomalies with in 24-48hours after birth with a written consentfromparents/ relatives,opinion of concerned Paediatrician was soughtin confirming the diagnosis.

Results: Total births during the study period were 650. The total number of cases withmusculoskeletal malformations were 25 with an incidence of 2.9/1000 births and the totalnumber of malformations were 25. 80% of cases were born alive and 20% of caseswere still born. MSK malformations were more common in male babies (64%) than infemale babies (36%). 56% of cases had low birth weight and 44% had normal birthweight (meanbirth weight of allneonates = 2.28 kg). 60% of cases attained the termand 40% were

premature babies. Malformations were more common in first and second para(44% each) and declined with increased parity.

Conclusion: The current study found a lower incidence of congenital MSK abnormalities than in India and internationally. CTEV incidence was higher than other anomalies in our sample, but lower than other studies. Males had more congenital MSK abnormalities than females. Most newborns with MSK abnormalities were full-term and healthy. Younger moms have more kids with MSK abnormalities, and parity reduces them. Antenatal USG detected most abnormalities. Most patients were from upper-low SES families with poor parental education. Consanguinity seldom caused MSK malformations.

Keywords: Musculoskeletal; Congenital anomalies; Pre-natal diagnosis,Ultrasonography; Birth weight; Consanguinity

INTRODUCTION:

Congenital abnormalities have drawn attention since the beginning of time. William Harvey made an effort to investigate the causes of deformities, and his research led him to conclude that teratological abnormalities were the outcome of developmental disorders [1, 2]. Congenital malformations are structural anomalies that are apparent at birth and are caused by improper development [3, 4]. Genetic and environmental factors interact in a complicated way to cause malformations. Most frequent abnormalities are thought to be the result of multifactorial inheritance. When used during intrauterine life, certain teratogens may have an impact on the developing foetus, resulting in persistent postnatal harm, changes in morphology, or changes in function [5]. These agents can include chromosomal problems, radiation, medications like thalidomide, infections like rubella, and dietary inadequacies [6]. Numerous maternal variables, such as maternal age, parity, prenatal sickness, and medications, have been linked to these congenital abnormalities. Mothers of deformed newborns were more likely to have a history of past abortions, concurrent illnesses, or pre-eclampsia during the present pregnancy. When a research on numerous congenital deformities was published in the 1960s using information gathered from many maternity facilities in Mumbai, India, interest in congenital malformations began to grow in that country [7-10].

One of the most often reported neonatal diseases is a flail upper extremity caused by a brachial plexus injury during childbirth or a lower limb weakness caused by myelodysplasia. The condition known as developmental dysplasia of the hip (DDH) is another one that

frequently affects newborns [11]. Flexion contractures are frequent in newborns, however they typically go away over time. Even though they are less frequent, other diseases including congenital muscle torticollis and congenital knee dislocation have been frequently documented [12]. It is crucial for both pediatricians and orthopaedic surgeons to recognised and classify these diseases. A newborn's future health can be ensured as well as the prevention of later complications with prompt identification and treatment [13]. The purpose of this study was to evaluate the risk variables associated with orthopaedic congenital abnormalities in newborns in our context, as well as to determine the prevalence of these abnormalities and compare our findings to those from earlier surveys [14]. We wanted to make clear the actions that may be taken at the right moment to avoid and treat these abnormalities and, in turn, ensure a safer future. To do this, we sought to analyse these risk factors [15]. It is now much simpler to investigate and identify different congenital defects, intervene, and take the appropriate measures thanks to the development of interventions like ultrasound and amniotic fluid analysis.

METHODOLOGY

Sourceofdata: The data for the present study was collected from Department of Anatomy, PES institute of Medical Sciences and Research, Kuppam, Andra Pradesh, India. Thestudywasdoneforaperiodofoneyear, from October 2021 to October 2022. All birth safter 28 completed weeks were examined forcong enitalmusculosk eletal anomalies with in 24-48 hours after birth with a written consentfromparents/relatives. The detail so feach baby with congenitalmusculoskeletalanomaly,regardingbirthweight,sex,periodofgestationandfamilyhistoryincludingmother'sage,antenatalhistory,parentaleducation,socio-economicstatusandparentalconsanguinitywererecordedaspertheproforma.
Afterbirthbabieswithmusculoskeletalanomalieswereexaminedclinically.PhotographsandRadiographsweretakeninnecessarysituation.

INCLUSIONCRITERIA

- Allbirthsafter28completedweeks(livebirthandstillbirths)withmusculoskeletalanomalies wereincluded.
- Babieswhohaveundergonesurgeryforthecorrectionofmusculoskeletaldefectsduringearlyneonatalperiodwereincluded.
- Babieswithothersystemanomaliesalongwithmusculoskeletalanomalieswereincluded

EXCLUSIONCRITERIA

- Babiesbornbeforetwentyeightcompletedweekswereexcludedfromthestudy.

RESULTS:

The present studywas undertakenbycollecting the data which includedLive births, Still births (>28 weeks of gestation) duringwhichperiod650newbornneonateswereexamined. Out of 650 births, 25 cases of congenital musculoskeletal mal formations were observed with anincidence of 2.9/1000 births,16cases(64%)weremale,9 cases (36%) female. In these 25 cases, a total of 20 malformations were notedwithanincidenceof3.2/1000births.

The distribution of congenital MSKmal formation shighamongmale children with a male to female ratio of 1.31:1.(Table-1)

Genderwisedistributionofmusculoskeletalmalformations

TABLE1:GENDERWISEDISTRIBUTIONOFMUSCULOSKELETALMALFORMATIONS

Sexofthechild	**TotalNo.**	**%**
Male	16	64.00
Female	09	36.00
Total	**25**	**100**

Birthweightofthebabies: Out of 25 cases ,significantnum berof babies (14babies–64.00%)were found in the birth weigh trange of 2,460 gmand aboveconstituting 64.00%(p=0.006).The numberof cases in creased with in crease in the birth weight of babies .Only 2 babies had the birth weight<1000gm.(Table 2)

TABLE2:BIRTHWEIGHTOFCASES

Weight(grams)	**No.ofcases**	**%**
<1000	1	4.0
1001-1500	3	12.0
1501-2000	5	20.0
2001-2500	5	20.0
2501andabove	11	44.00
Total	**25**	**100**

RelationofBirthweighttoMSKanomalies: Babies weighing less than 2.5 kg are considered as low birth weight babies.Most of the babies born with congenital MSKmalformations had normal birthweight.Outof25cases,14 cases (56%) hadlowbirthweight,weighinglessthan 2.5

kg. whereas 11cases(44.00%) weighedmore than 2.5kg. But themeanbirthweightofallneonateswas2.28kgshowingrelationshipbetweentheoccurrenceofcongenitalmalformationsandlowbirthweight.Themeanbirthweight of female babies (2.35 kg) was more than male babies (2.22 kg) but thedifferencewasnotstatisticallysignificant.(Table3and4)

TABLE3:MEANBIRTHWEIGHTOFCASES(N=25)

Sex	**MeanWeightinkg**	**SD**
Male	1.99	0.83
Female	2.70	0.71

P=0.6

TABLE4:CONGENITALMSKCASESINRELATIONTOBIRTHWEIGHT

Birthweight(kg)	**No.ofcases**	**%**
<2.5	09	36.00
>2.5	16	64.00
Total	**25**	**100**

Periodofgestation: In our study out of total 25 cases, 15 cases (60.00%) of congenital MSKmalformation were seen in full term neonatesand 10 cases (40.00%)inpreterm babies and the difference wass tatistically significant (p=0.033)(Table-5). When birth weight and gestational age was analysed, it showed 7 cases (28%) of full term and6(24%) of preterm neonates were of low birthweight. 3 case (12%) of pre-term and 9 cases (36%) of term neonates had normalbirthweight (Table 6). Out of 25 cases,8cases were belonging to first birth order,in that 6 cases (24%) were termneonatesand 7cases (28%)were preterm.There maining 17 cases were of second birth order and above, of which 9 cases (36%)were termand 3cases (12%)werepreterm (Table7).

TABLE5:PERIODOFGESTATION(N=25)

Gestationperiod	**TotalNo.**	**%**
Pre-term	10	40.00
Term	15	60.00
Total	**25**	100

P=0.033

TABLE6:BIRTHWEIGHTV/SGESTATIONALAGE(N=25)

Birthweight	Pre-term	Term	Total
<2.5kg	7(28%)	6(24%)	13
>2.5kg	3(12%)	9(36%)	12
Total	10	15	25

TABLE7:ORDEROFBIRTHV/SPERIODOFGESTATION(N=25)

Order	Pre-term	Term	Total
G1	3(12%)	5(20%)	8
G2andabove	7(28%)	10(40%)	17

Maternalage:

Outof25cases,10cases(40%)ofMSKmalformationsweredistributedintheagegroupof21-25years,whereas6cases(24 %)eachweredistributedinthe agegroup of15-20years and26-30years. Only3 cases wereborntoanelderlyprimi(12%). (Table8). Thehighestoccurrenceofcaseswasdistributedintheagegroupof21-25years(40 %)andthedifferencewasstatisticallysignificant(P=0.006).

TABLE8:DISTRIBUTIONOFCASESACCORDINGTOMATERNALAGE

MaternalAgeGroup(years)	No.ofcases	%
15-20	6	24
21-25	10	40
26-30	6	24
31andabove	3	12
Total	25	100

P=0.006

Parity: Outof25casesweobserved11 cases(44%)eachbelongedtoparaIandII,2cases(8%)toparaIIIand1case(4%)toparaIV.Thedifferen ceinthedistributionofcasesindifferentparitywasstatisticallysignificant(Table 9).

TABLE9:DISTRIBUTIONOFCASESACCORDINGTOPARITY

Parity	**No.ofcases**	**%**
P1	11	44
P2	11	44
P3	2	8
P4	1	4
Total	25	100

Chi-squaretestforgoodnessoffit,P=0.001

Socioeconomicstatusofparents:

ThesocioeconomicstatusofparentsofneonateswithcongenitalMSKanomalies was classified according to Prasad's classification, based on per capitapermonthincome.11cases(44%)belongedtoupperlowerclassandthedifferencewasstatistic allysignificant.Ourstudyisbasedonagovernment Institutionwhereinpatientsbelongingtopoorerse ctionofsocietyattendonalargerscaleandformasizablepopulationvisitingthehospital(Table 10).

TABLE 10: NUMBER OF CASES ACCORDING TO PRASAD'SCLASSIFICATIONOFSOCIO-ECONOMICSTATUS

Socio-economicclass	**No.ofcases**	**%**
Upperclass	2	8
Uppermiddleclass	3	12
Lowermiddleclass	4	16
Upperlowerclass	11	44
Lowerclass	5	20
Total	25	100

p=0.047

DistributionofanomaliesinrelationwithGeographicarea:

Inourstudy11cases(44%)belongedtoHyderabad, India**.**proper,14cases(56%) belonged to rural areas within a radius of 30 kms from Hyderabad, India.Thus most ofthe casesbelongtoruralpopulation.

Maternalriskfactors:

Outof25casesofcongenitalMSKanomalies,13cases(52%)wereassociatedwithvarioustypesofris

kfactorsseenduringpregnancywhereasin12cases(48 %)therewasnoriskfactor.However,chromosomalstudycouldnotbedoneduetolackoffacilityinour institutionandunaffordabilityofparentsduetohighcost. Altogether, 13 cases (52%) were associated with various risk factors ofwhich,3 cases (12%) were associated witholigohydromnios, 3 cases (12%)withpolyhydromnios,2cases(8%)gaveH/Ofeverduringfirsttrimester and during20weeksof pregnancyandwasonregularinsulinmedication,2cases(8%)withH/Oprevious abortions, 2 case (8%) of PIH patient developed intrapartum eclampsia (Table 11).

TABLE11:MATERNALRISKFACTORS

MaternalRiskfactors	**No.ofcases**	**%**
Oligohydramnios	3	12
Polyhydromnios	3	12
Eclampsia	2	8
Infections/fever	1	4
Diabetesmellitus	2	8
Previousabortions	2	8
Total	13	52

Consanguinity: Out of 25 cases, 6 cases (24%) were born to consanguineous couplesand19 cases(76 %)tonon-consanguineouscouples.Seconddegreeconsanguinitymeansmarriagesbetweenuncle-nieceoraunt-nephew.Thedifferencewasstatisticallysignificant (Table 12).

TABLE 12: RELATION OF CONSANGUINITY TO MSKMALFORMATIONS

Consanguinity	**No.ofcases**	**%**
Consanguineous	6	24
Non-consanguineous	19	76
Total	25	100

Chi-squaretestforgoodnessoffit,p<0.0001

Percentageofcasesamonglivesbornandstillborn: Out of25cases, 20 cases(88.2%)were bornaliveand5cases(20%)were stillborn and the difference was statistically significant. (p=0.000) (Table 13)Among 2 still births, 2 case (no.22 in MC) had B/L CTEV, hydrocephalus, spinabifida of lumbosacral spine. 1 Case (no.11) had spina bifida, anencephaly, and thoracicmeningocele. 1 Case (no.8) had omphalocele with liver, spleen, small and largeintest ineprotruding ou tof the abdomenassociated with Pentalogy of Cantrell and it was atermbaby. 1Case (no.10)hadlargeomphalocele with liver,small and largeintest ineprotruding out of the abdomenand it was alsoassociated with severe shortening of spine and large meningocele. Only one baby was still born atterm gestationandotherswerepre-term babiesandthesebabieshadassociated CNSanomaliesandinmostofthestudiesithasbeendocumentedthatbabieshavingCNSanomalieswill dieatanearlygestationperiod.

TABLE13:PERCENTAGEOFCASESAMONGLIVEBORNANDSTILLBORN

Typeofbirth	**No.ofcases**	**%**
Liveborn	20	80
Stillborn	5	20
Total	25	100

Chi-squaretestforgoodnessoffit,p<0.0001

Percentageofcasesaccordingtotypeofdelivery: Out of 25 cases,16 cases (64%) were born by normal vaginal delivery, where as 9 cases (36%) were extracted by LSCS. The difference was not statistically significant (Table-14).

TABLE14:PERCENTAGEOFCASESACCORDINGTOTYPEOFDELIVERY

Typeofdelivery	**No.ofcases**	**%**
Vaginal	16	64
LSCS	09	36
TOTAL	25	100

Chi-squaretestforgoodnessoffit,p=0.5

Casesdetectedbyultrasonography: Out of 25 cases 15 cases (60%) of musculoskeletal malformations weredetected by ultrasonography. Rest of the 10 cases (40%) were not detected by ultrasonography (Table 15) the difference was not significant statistically.All25 cases hadantenatal screening foranomaliesat least onc ebutinmostof the cases anomalies were

not detected.

TABLE 15: NUMBER OF MSK ANOMALIES DETECTED BY ULTRASONOGRAPHY

Anomaliesdetectedbyultrasound	**No.ofcases**	**%**
Yes	15	60
No	10	40
Total	25	100

DISCUSSION:

Congenital abnormalities have become a significant contributor to prenatal mortality in developed nations due to improved infection and nutritional deficiency illness control, and they will likely soon play a significant role in perinatal mortality in India. The current study found that there are 4.4/1000, or 0.44%, musculoskeletal abnormalities overall. In our analysis, the most prevalent abnormalities, accounting for 22.4% of cases, were musculoskeletal. According to several workers, defects of the musculoskeletal system are the most common anomalies in the majority of India. The incidence in various studies can differ based on the population sampled, the choice of research material, the clinical judgement of the treating physician, and the accessibility of lab assistance [16]. Talipes and polydactyly were the most frequent malformations of this system that were found in the current investigation (8cases). According to Stevenson et al., the incidence of talipes varied from 3.42 per 1000 people in Kolkata to 10.95 per 1000 people in Panama City in a global research sponsored by the WHO [17]. This variance may have occurred primarily because in some institutions, any foot malposition was labelled as a talipes. Perhaps because they are outwardly evident and hence easily recognised at birth, musculoskeletal anomalies rank near the top of most series' lists of malformations. Sometimes they could be overdiagnosed and merely be foetal postural restrictions [18]. The prevalence of deformed newborns did not appear to be influenced by the mother's age. Numerous other researchers have reported having similar results. Additionally, it was found that the prevalence of congenital abnormalities is mostly unaffected by better maternal care and rising levels of life.

Similar to what Parmar et al. reported, there is a higher incidence of abnormalities in the primipara group as compared to multipara. However, a number of studies have identified a

greater frequency of multipara, with Naoom et al. reporting 76.7% and Jehangir et al. reporting 88.89%, respectively, whereas Anand et al. found no significant correlation. We discovered a strong relationship between congenital anomalies and maternal age. Taksande et al. reported similar outcomes. Other research found no association between maternal age and congenital abnormalities. Foot abnormalities made up a significant portion of all deformities (43%) with the highest CTEV frequency of 5.62/1000 [19]. In contrast to the 0.66/1000 calcaneovalgus in our study, Chotigavanichaya et al. reported CTEV as 4/1000, polydactyly as 2.6/1000, and calcaneovalgus as 60/1000 live infants. They also reported 7.6/1000 live newborns having metatarsus adductus. In their investigation, Widhe found that 3.1% of people had adductus, and 0.5% had calcaneovalgus. 4.78 per 1000 people had a hand deformity, with poly- and syndactyly being the most common types. In 0.19/1000 cases, a radial club hand was observed. Upper limb abnormalities were reported to occur 1 in 506 times by Giele et al. and 21.5 times per 10,000 live births by Ekblom et al. Anencephaly (0.19/1000 live births), spina bifida (0.11/1000 live births), and encephalocoele (0.07/1000 live births), which are far less common than those in our study [17, 19], are the most prevalent neural tube abnormalities (NTDs) according to the Malaysian registry. But Bhide et alsystemic .'s review, which reported an overall birth prevalence of 4.1/1000 and a live birth prevalence of NTDs at 1.3/1000 births, was quite similar to our findings.

Birth-related trauma is not unusual. The incidence may rise as a result of the use of forceps or caesarean sections. Although the clavicle has been identified as the bone that fractures most frequently during birth, other bones, including the femur, humerus, and ribs, have also been implicated. 73 occurrences of clavicular fracture were found by Ozdener et al., with a prevalence of 0.75 percent (73/9700). A number of humerus diaphyseal fractures have been documented by Husain et al., whereas six neonatal distal humeral epiphysis separations have been documented by Jacobsen et al. With a combined frequency of 1.12/1000, we discovered three occurrences of humeral and femoral fractures, as well as one clavicle fracture. Our study's incidence of Erb's palsy matches Jaggat et al., perfectly. Study's In their study, Ouzounian et al. reported a 3/1000 incidence rate. There are between 1.5 and 2.5 cases of DDH recorded per 1000 live births. In contrast, we discovered substantially fewer DDH in our study. Our study's primary drawback is that it was conducted exclusively in hospitals, meaning that the results might not be generalizable to other populations [20]. Additionally, because OPD patients were not included in the study, the study group, which consisted primarily of neonatal unit patients, may have been biased. The most crucial step in lowering

the prevalence of congenital abnormalities, which are a significant source of illness and mortality in newborns, is raising awareness among parents and medical professionals. Prenatal screening should be a standard practise for all patients as part of preventive efforts, as well as genetic counselling for the parents.

CONCLUSION

As contrast to India and other Asian countries, our current research showed a decreased incidence of congenital MSK abnormalities. In our population, CTEV incidence was high than some other anomalies although less than in other research. Congenital MSK anomalies were more common in males than females. The majority of neonates with MSK anomalies were healthy full-term babies. Parity decreases MSK abnormalities, which are more common in children of younger mothers. Most problems were found during antenatal USG. The majority of patients came from lower-middle class backgrounds with uneducated parents. It is uncommon for MSK abnormalities to be congenital.

Conflict of Interest

None

Funding

None

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