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CONGENITAL VISIBLE BIRTH DEFECTS AT 31 HOSPITALS, DELHI

Paediatrics

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ABSTRACT

Background and Aim: Insufficient data exists on the prevalence and types of individual visible birth defects (VBDs) in live-born babies in India. This initiative addressed this gap by reporting on VBDs in live births in Delhi. **Material and Methods:** Data on VBDs for live born babies was collected from November 2021 to July 2023, across 31 hospitals under Mission NEEV (Neonatal Early Evaluation Vision). Pediatricians confirmed diagnoses using ICD10 coding, and prevalence rate was calculated. **Results** showed 818 babies with VBDs among 193,587 screened live births, resulting in an overall prevalence of 0.43% (840 VBDs among 818 live babies). Of these, 310 were minor, and 530 were major, with a higher incidence in male babies (56% vs. 41%). Consanguinity was observed in 7.15% of parents of malformed babies. Among the 818 babies, 798 (97.5%) had isolated and 20 (2.44%) had multiple anomalies. Musculoskeletal VBDs were the most common (663 per 1000 births). The most common anomaly among all VBDs was CTEV (Congenital talipes equinovarus) (48.8%). **In conclusion,** the prevalence of VBDs among live births was 0.43%, with musculoskeletal system deformities being the most common, especially in male babies. Early newborn surveillance for VBDs is essential for identifying associated anomalies and early intervention.

KEYWORDS

Visible birth defects.

BACKGROUND AND AIM:

Visible structural birth defects, whether isolated or multiple, major or minor, can be associated with other internal defects, leading to adverse outcomes. Previous literature has extensively reported on the combined data of visible and internal structural defects, which constitute a significant category among congenital birth defects (CBDs). [1] Both visible and internal defects are a major contributor to neonatal and infant mortality and disability worldwide. According to the 2012 World Health Statistics, serious birth defects affect approximately 6% of newborns annually. [2] The prevalence of CBDs varies across low-income, developing, and developed countries. [3] The 2013 Global Burden of Disease study identified CBDs as one of the top ten causes of under-5 child mortality. [4] In India, CBDs were the fifth significant cause of neonatal deaths in 2010 [5], responsible for 8-10% of perinatal deaths [6] and 13-16% of neonatal deaths [7].

Accurate country-specific data on CBD prevalence and detailed evaluations are crucial for effective planning of surveillance and preventive measures. [8] In the Southeast Asia Region (SEAR), countries have established a regional framework for birth defect prevention and control, supported by the World Health Assembly. In India, although some hospital-based studies have reported varying CBD prevalence [9, 11], there is no comprehensive data on individual and associated VBDs.

This initiative offers comprehensive data on the prevalence, spectrum and associated defects related to VBDs among live babies screened under the NEEV mission supported by Rashtriya Bal Swasthya Karyakram program [12].

MATERIAL AND METHODS:

Data on antenatal history, baby's gender, birth weight, and visible birth defects (VBDs) were collected for live-born babies in 31 Delhi hospitals as part of Mission NEEV between November 2021 and July 2023. Information was recorded within 24 hours of birth with informed consent. Excluded were intrauterine deaths, stillborns, and babies who left against medical advice within 24 hours of birth. Pediatricians confirmed diagnoses using ICD-10 coding and further evaluations done to identify associated visible anomalies. Babies with multiple VBDs were counted once. The VBD birth prevalence was calculated by dividing detected cases by total live births during the same period. All surviving babies were managed according to the mission's protocol.

RESULTS:

Out of 204,973 total deliveries, 199,205 were live births, 193,587 were screened and 818 babies had malformations, resulting in a 0.43% of overall VBDs prevalence (840 VBDs in 818 live babies). Of the 818 babies, 65 died before examination, and 35 died after examination by a pediatrician. The prevalence was higher in males (57%) than in females (41%) and 2% were an unknown gender. Consanguinity was observed in 7.15% of parents of malformed babies. Among the 818 babies, 97.5% had isolated anomalies, and 2.44% had multiple anomalies. Most mothers (61%) were in the 26-30 years age group. 25% babies were preterm, and 23% had low birth weight. Out of the 840 VBDs, 310 were minor, and 530 were major (Table 1).

Table 1 - Major and Minor VBDs

Major			
Male	**Female**	**Unknown**	**TOTAL**
275	235	20	530
Minor			
Male	**Female**	**Unknown**	**TOTAL**
164	146		310

System-wise major anomalies as per International Classification of Diseases, tenth revision classification (ICD-10) – Figure -1. Among the elven syndromes, five were Down syndrome, one was Orofacial Digital syndrome1, Potocki-Shaffer syndrome and Kleeblattschädel syndrome each and three syndromes were non-specific.

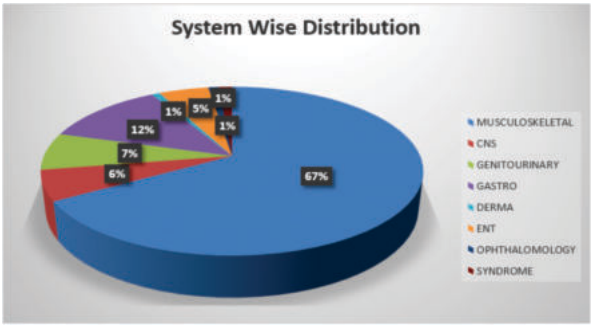

Figure -1 -System-wise major anomalies as per ICD-10. CNS: Central Nervous System

Percentage of individual congenital anomalies based upon commonest malformation in Figure -2.

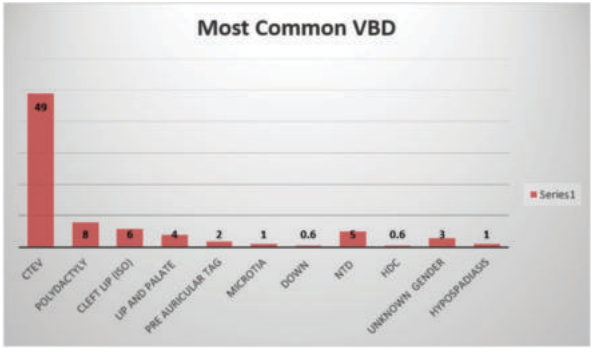

Figure 2- Individual congenital anomalies based upon the commonest malformation (%).

CTEV : Congenital Talipes Equinovarus, NTD : Neural Tube Defect, HDC: Hydrocephalus,

DISCUSSION:

VBDs, a type of congenital structural defect, were identified in newborns during Mission NEEV, with a prevalence of 0.43% among live births. This rate differs globally 1.91% [11] 1.75% [13] in Indian studies and 2.5% [14] in studied from Egypt and 5.1% [15] in Wales. It's higher than studies in Northeast India 0.08% [16] and a North Indian tertiary care center 0.018% [17], possibly due to regional, risk factor, racial, social, ecological, and economic variations, as well as differences in case definition, methodology, and diagnostic protocols.

Our study showed a male preponderance, consistent with other reports [9]. Maternal age between 26-30 years was linked to congenital malformations, but Jenita Barua et al [9] found significance for ages over 30. Our study did not find any significant association of congenital malformations in low birth weight (LBW) or preterm babies, though other studies have reported[9]. Consanguinity didn't show significance in our study, but Patra C et al. [18] found a connection between congenital anomalies and consanguineous relationships.

In our study, musculoskeletal system malformations were the most common (663/1000), aligning with findings in Egypt [14] and Indian studies [9]. In Iran [19] and Tanzania [20], CNS (Central Nervous System) abnormalities were prevalent. While some literature highlights cardiovascular malformations as the most common [11], we focused on gross anomalies data, so cardiac malformations weren't reported here.

Understanding VBDs' prevalence, spectrum, and impact on mortality and long-term disability is crucial for reducing the overall burden and improving prenatal diagnosis and prevention, especially in high-risk groups. With a large, representative sample from North India, this study underscores the need for a national birth defect surveillance program and services that could significantly enhance the achievements in national healthcare system.

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Conflict of Interest: None

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