Original Research

Congenital Talipes Equinovarus: Prevalence, Characteristics, and Therapeutic Outcomes

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Abstract

Congenital Talipes Equinovarus (CTEV), commonly known as clubfoot, is a complex congenital deformity of the foot characterized by four key abnormalities: plantar flexion, heel inversion, forefoot adduction, and a high arch. Affecting approximately 1-2 in 1,000 live births, CTEV shows significant geographic and ethnic variations in prevalence, with higher rates in some populations. The condition is predominantly observed in males, and both genetic and environmental factors are believed to play a role in its development. CTEV can occur as an isolated deformity or as part of syndromic conditions, complicating management and outcomes.

This cross-sectional study conducted in Kolkata, India, focused on 100 CTEV patients, examining associated musculoskeletal anomalies and treatment outcomes. Key maternal factors, such as twin pregnancies and breech presentation, were noted. A positive family history of CTEV was observed in a significant portion of patients, highlighting the genetic component of the condition. Associated anomalies, including Down's syndrome and arthrogryposis, were prevalent, complicating treatment and prognosis.

The Ponseti method, a non-surgical treatment involving serial casting, emerged as the most effective approach, with a high success rate. However, some patients required additional procedures like tenotomy or surgical correction, especially those with syndromic CTEV. Socioeconomic factors influenced parental awareness of CTEV but did not significantly affect treatment outcomes.

This study underscores the importance of early diagnosis and intervention in CTEV management. While treatment outcomes are generally favorable, the presence of associated anomalies complicates clinical management, necessitating a multidisciplinary approach. Further research into genetic and environmental factors contributing to CTEV is essential to improve long-term patient outcomes.

Key Words: CTEV, musculoskeletal anomalies, ponseti, arthrogryposis, musculoskeletal problem

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Introduction

Congenital Talipes Equinovarus (CTEV), commonly known as clubfoot, is a prevalent congenital deformity affecting the musculoskeletal system. It involves four primary foot abnormalities: plantar flexion (equinus), heel inversion (varus), forefoot adduction, and cavus (high arch). These deformities cause the foot to twist inward and downward, potentially leading to significant disability if untreated.[1] CTEV affects approximately 1-2 out of every 1,000 live births globally, with some populations having rates as high as 6 per 1,000[2]. Males are more commonly affected, with a male-to-female ratio of 2:1 or 3:1 [3]. While the exact causes remain unclear, both genetic and environmental factors are implicated. CTEV can present as an isolated deformity or in association with other congenital abnormalities[4].

Prevalence

CTEV prevalence varies based on geography, race, and genetics. Estimates of the birth prevalence of clubfoot in low and middle income settings range from 0.5 to 2 per 1000 births. However, there is currently no estimate of global birth prevalence of clubfoot as per 2023 studies . The pooled prevalence of clubfoot was 1.18 per 1000 births (95% CI: 1.00–1.36) based on data from 44,818,965 births[4]. The highest prevalence rates were observed in low- and middle-income countries, particularly in the South-East Asia Region (1.80, 95% CI: 1.32–2.28) and the Africa Region (1.31, 95% CI: 0.86–1.77)[5]. We estimate that 176,476 (95% CI: 126,126–227,010) children will be born with clubfoot globally each year[6]. Ethnic and racial differences have also been observed, with Caucasians generally showing higher CTEV rates compared to Asians and Africans. Notably, specific groups, such as the indigenous Polynesians of New Zealand, have unusually high incidence rates [7-8]. Apart from genetic variations, seasonal variations in CTEV births have also been reported, with higher incidence in colder months, potentially linked to environmental factors like vitamin D deficiency during winter [9-11].

Characteristics

CTEV is categorized into idiopathic and syndromic types. Idiopathic CTEV, the most common form, occurs in otherwise healthy children and often has a hereditary component, with siblings of affected individuals at higher risk[12]. Syndromic CTEV, associated with other congenital disorders such as arthrogryposis, spina bifida, or trisomy 18, tends to be more severe and resistant to treatment[13-14]. Clinically, CTEV is easily recognizable at birth, with the foot turned inward and downward. Severity varies, with some cases requiring minimal intervention and others needing more extensive treatments. Early diagnosis and intervention are crucial for favorable outcomes, ideally within the first few weeks of life [15].

Outcomes and Treatment

The Ponseti Method is the current gold standard for treating CTEV. It involves weekly manipulations and casting over 6 to 8 weeks, followed by bracing to maintain correction [16]. The initial casting period takes about 6 to 8 weeks, followed by a period of 3 months during which the baby wears a removable orthotic 23 hours a day. After that, the child continues orthotic treatment for sleep (naps and nighttime) until 5 years of age. The treatment phase should begin as early as possible, optimally within the first week of life. Gentle manipulation and casting are performed on a weekly basis. Each cast holds the foot in the corrected position, allowing it to gradually reshape. Generally, 5 to 6 casts are required to fully correct the alignment of the foot and ankle. At the time of the final cast, the majority of infants (90% or higher) will require an Achilles tendon lengthening procedure. The final cast remains in place for 3 weeks, after which the infant's foot is placed into a removable orthotic device. The orthosis is worn 23 hours per day for 3 months and then during naps and night-time until 5 years of age. Failure to use the orthosis correctly may result in recurrence of the clubfoot deformity. Good results have been demonstrated at multiple centers, and long-term results indicate that foot function is comparable with that of normal feet. This approach has a success rate of up to 90% when initiated early[17]. For more severe or resistant cases, surgery may be required, though it carries higher risks of complications[18]. Even with treatment, some children may experience residual deformities, and recurrence can occur, particularly during growth spurts [18-19]. Long-term follow-up is essential to monitor for potential relapses[20].

The present research aims to study the proportion of musculoskeletal congenital anomalies in congenital talipes equinovarus, any gender predilections associated and hoe much are the families are aware of such anomaly and the outcome of such anomalies related to the awareness.

Aims and Objectives

The primary aim of this study was to examine the proportion of other musculoskeletal anomalies in CTEV patients and assess parental awareness regarding the condition. It also sought to identify the clinical outcomes of these associated anomalies. The specific objectives included:

- Assessing the prevalence of CTEV in relation to other congenital musculoskeletal anomalies.
- Investigating factors such as socioeconomic background, maternal health during pregnancy, and the role of family history in CTEV occurrences.

Methodology

This cross-sectional study was conducted at a medical institution in Kolkata, India, over an 18-month period. The sample comprised 100 CTEV patients ranging from newborns to children up to 5 years of age. These patients were selected from the Orthopedic and Pediatric outpatient departments. Exclusion criteria included patients with severe congenital visceral anomalies such as cardiological or urogenital abnormalities. Data was collected by Systematic random sampling. Power analysis was not done.

Data was collected selectively focused on the child's age and gender, maternal factors (such as age, antenatal history, nutritional deficiencies, and family history), and physical examinations. The study also included imaging studies, such as digital X-rays of various body parts, to assess limb deformities and musculoskeletal anomalies.

Statistical analyses were performed using SPSS software. Chi-square tests were employed to compare proportions, and t-tests were applied to examine differences in continuous variables. P-values below 0.05 were considered statistically significant.

Results

The study population comprised 74% male and 26% female patients, reflecting a male-to-female ratio of 2.8:1. A similar gender distribution has been observed in other studies on congenital musculoskeletal anomalies. The majority of the patients (43%) were in the 0-1 year age group, followed by 27% in the 1-2 year range.

Socioeconomic status varied significantly between the two groups studied: aware and non-aware. The non-aware group had a higher proportion of individuals from lower socioeconomic backgrounds. Furthermore, the majority of patients were from urban areas, though this distribution was not statistically significant between the aware and non-aware groups.

Key maternal factors such as twin pregnancies, breech presentation, and gestational diabetes were noted. Breech presentation was observed in 51% of the cases, while 17% of the pregnancies involved twins. Amniocentesis had been performed on 18% of the mothers, and gestational diabetes was diagnosed in 8% of the cases. (Table 1)

Table 1: Distribution of study subjects according to maternal factors during pregnancy (n=100)

Maternal factors during pregnancy	Frequency	Percent
Twin pregnancy	17	17.0%
Amniocentesis	18	18.0%
Gestational DM	8	8.0%
Pre-eclampsia	6	6.0%
Breech presentation	51	51.0%
Total	100	100.0%

The study revealed a strong association between family history and CTEV occurrence. Among the non-aware group, 81.1% had a positive family history of CTEV, compared to 74.6% in the aware group. However, this difference was not statistically significant. Previous studies also indicate that siblings and offspring of individuals with CTEV are at increased risk of developing the condition.

Associated Anomalies

Several congenital anomalies were observed among the CTEV patients. Notably, 31% of patients

had Down's syndrome, 16% had arthrogryposis multiplex congenita, and 7% had Moebius syndrome. Additionally, 6% had diastrophic dysplasia, and 5% were diagnosed with Freeman-Sheldon syndrome. These syndromic associations complicate the treatment and prognosis of CTEV patients. (Table 2)

Table 2: Distribution CTEV patients as per presence Of other congenital Syndromes. (n=100)

Presence of Syndromes	Frequency	Percent
Absent	14	14.0%
Arthrogryposis congenital multiplex	16	16.0%
Diastrophic dysplasia	6	6.0%
Down syndrome	31	31.0%
Freeman Sheldon syndrome	5	5.0%
Larsen syndrome	6	6.0%
Moebius syndrome	7	7.0%
Spinal dysraphism	15	15.0%
Total	100	100.0%

Co-occurring musculoskeletal anomalies included polydactyly (27%), developmental dysplasia of the hip (21%), congenital knee dislocation (6%), and cleft lip (11%). Absent pectoralis major and proximal radioulnar synostosis were also observed in a small number of patients. (Table 3)

Table 3: Distribution study subjects according to presence of Co-occurring musculoskeletal anomalies (n=100)

Co-occurring musculoskeletal anomalies	Frequency	Percent
Absent pectoralis major	5	5.0%
Cleft lip	11	11.0%
Congenital knee dislocation	6	6.0%
Developmental dysplasia of hip	21	21.0%
Myotonia congenital	6	6.0%
Osteogenesis imperfecta	6	6.0%
Polydactyly	27	27.0%
Proximal radioulnar synostosis	18	18.0%
Total	100	100.0%

Treatment Outcomes

The study evaluated three primary treatment modalities: the Ponseti method, tenotomy, and surgical correction. The Ponseti method was the most widely used, with 65% of patients receiving this treatment. Of these, 20% required additional tenotomy, and 15% underwent surgical correction .

In terms of clinical outcomes, the majority of patients achieved successful correction with the Ponseti method, while a small proportion exhibited residual deformities or required further corrective surgeries. Notably, patients with associated syndromes such as Down's syndrome or arthrogryposis had poorer outcomes compared to those with isolated CTEV.

Discussion

This study highlights key insights into Congenital Talipes Equinovarus (CTEV), also known as clubfoot, focusing on its characteristics, risk factors, and treatment outcomes. CTEV, a congenital musculoskeletal deformity, requires non-surgical or surgical interventions for correction. The research uncovers factors such as gender distribution, socioeconomic disparities, maternal influences, genetic predisposition, and associations with other congenital anomalies, all contributing to the complexity of managing CTEV..

Gender Distribution and Age Group

The study shows that CTEV predominantly affects males, with 74% of the patients being male (male-to-female ratio 2.8:1), consistent with prior studies. This gender bias is noted in other congenital musculoskeletal anomalies, though the biological reasons are unclear, possibly linked to genetic or hormonal factors[5]. The majority of patients (43%) were in the 0–1 year age group, with 27% in the 1–2 year group, emphasizing the importance of early diagnosis. Since CTEV is usually diagnosed at birth, early intervention, especially within the first year, improves treatment outcomes[21] .

Socioeconomic Status and Awareness

Socioeconomic factors significantly impacted patient awareness and healthcare access. Lower socioeconomic groups were more represented in

the non-aware group, consistent with prior studies indicating that limited resources and healthcare access can delay diagnosis and treatment[22] . While most patients were from urban areas, urbanization did not statistically influence awareness, suggesting that even in urban areas, socioeconomic challenges remain barriers to care. Enhancing awareness among lower-income groups could lead to earlier diagnosis and better adherence to treatment[23].

Maternal Factors

Key maternal factors linked to CTEV included twin pregnancies, breech presentations, and gestational diabetes. Breech presentation, observed in 51% of cases, aligns with research suggesting that abnormal fetal positioning increases the risk of musculoskeletal deformities like CTEV[24]. Twin pregnancies, noted in 17% of cases, are associated with higher congenital anomaly risks due to reduced intrauterine space and increased fetal pressure[25]. Gestational diabetes and amniocentesis, though less directly linked to CTEV, have been associated with congenital anomalies in other studies[26-27].

Family History and Genetic Predisposition

The study found a strong genetic component to CTEV, with 81.1% of the non-aware group having a positive family history, compared to 74.6% in the aware group. Although not statistically significant, the high familial cases reinforce previous findings that siblings and offspring of those with CTEV have a heightened risk[28-29] . Genetic counseling could benefit families with a history of CTEV, especially when multiple family members are affected.

Associated Congenital Anomalies

A significant portion of the study population had additional congenital anomalies, complicating treatment and prognosis. Down's syndrome (31%) was the most common, followed by arthrogryposis multiplex congenita (16%) and Moebius syndrome (7%). Syndromic associations like these often complicate CTEV treatment[30]. For instance, Down's syndrome is linked to hypotonia and ligamentous laxity, making treatment less effective and increasing recurrence risks[31]. Additional musculoskeletal anomalies, such as polydactyly (27%) and developmental dysplasia of the hip (21%), further complicate the clinical management of CTEV[32] . These findings suggest that a multidisciplinary approach is crucial for managing CTEV cases with co-occurring anomalies.

Treatment Outcomes

The study evaluated three main treatment methods: the Ponseti method, tenotomy, and surgical correction. The Ponseti method was the most common, used in 65% of cases. It involves serial casting and gentle manipulation, offering a high success rate and minimal invasiveness, and is widely regarded as the gold standard for CTEV treatment[33]. However, 20% of patients required tenotomy, a minor surgical procedure to lengthen the Achilles tendon, while 15% underwent more invasive surgery, typically reserved for severe or syndromic cases where non-surgical treatments were inadequate[34]. Syndromic CTEV patients, such as those with Down's syndrome or arthrogryposis, had worse outcomes and were more likely to require surgery[35], highlighting that while the Ponseti method works well for most, aggressive interventions may be needed in more complex cases.

Conclusion

This study provides significant insights into the epidemiology and outcomes of CTEV in an Indian population. Although overall treatment outcomes were favorable, the presence of associated congenital anomalies complicates management and prognosis. Future research should focus on understanding the genetic and environmental factors contributing to CTEV and improving awareness and early detection, particularly in low-income populations.

CTEV is influenced by genetic predispositions, maternal health, and possibly environmental factors. While parental awareness of the condition varies, it does not significantly affect clinical outcomes when appropriate treatment is provided. Continued research and a multidisciplinary approach to care are essential to further refine treatment strategies and improve long-term outcomes for CTEV patients, especially those with associated anomalies.

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