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Original Article

Prevalence and Risk Factors of Clubfoot in Al-Baha Region - Saudi Arabia

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Abstract

Background: Clubfoot, or Congenital talipes equinovarus (CTEV), is a structural defect that occurs early in pregnancy. It affects 1 in 1000 live births and can lead to permanent disability and severe social and economic consequences if untreated. It is more common in boys, with a male-to-female ratio of 2:1, and has a strong association with family inheritance and consanguinity. **Objectives**: To determine the prevalence of clubfoot and identify associated risk factors in the Al-Baha Region. **Methods**: The research team analyzed the medical records of patients from the orthopedic clinic at King Fahad Hospital between January 2020 and January 2024. A data sheet, designed by experts and consultants, captured demographic data, defect location, possible risk factors, associated anomalies, and treatment plans. Data analysis was conducted using Chi-square tests through SPSS v28. **Results**: The study found that 94.1% of patients were male, and 76.5% had bilateral deformities. Nearly half (47.1%) of the patients had parents with consanguineous marriages. Identified risk factors included preeclampsia, oligohydramnios, gestational diabetes, and family history of deformities. No significant correlation was found between maternal drug exposure or infections and clubfoot. Treatment, which primarily involved serial casting and Achilles tenotomy, resulted in 41.2% of patients walking normally post-treatment. **Conclusions**: This study highlights the prevalence of clubfoot in the Al-Baha region and its association with risk factors, emphasizing the importance of genetic screening, early diagnosis, and public awareness to improve outcomes.

Keywords: Clubfoot, TEV, Congenital deformity, Prevalence, Risk Factor, Consanguinity, Genetic Predisposition, Congenital Talipes Equinovarus.

Introduction

Clubfoot, or Congenital talipes equinovarus (CTEV), is a structural defect that occurs early in pregnancy (Smythe et al., 2017). It affects 1 in 1000 live births. If untreated, it can cause permanent disability and severe social and economic consequences (Ansar et al., 2018;

Grimes et al., 2016; Parker et al., 2009). It occurs two times more frequently in boys with a male-to-female ratio of 2:1 and with family inheritance. And bilaterality in about 50% of cases (Salvatori et al., 2020; Vukasinović et al., 2010). The risk of clubfoot can increase due to various factors, such as consanguineous marriage, smoking, family history, being male, and exposure to certain antidepressant drugs like selective serotonin reuptake inhibitors (SSRIs). These are the most significant factors that are clinically associated with higher odds of clubfoot development, where family history poses the greatest risk (Chen et al., 2018; Sahin et al., 2013)

A case-control study was conducted on screening for Congenital Talipes Equinovarus (CTEV) in a rural city in eastern Turkey between 2009 and 2011. The study found that babies born to first-cousin parents had more than four times the risk of idiopathic CTEV, and the risk for those born to distant relatives was 2.9 times higher than for children of unrelated parents (Sahin et al., 2013). According to a 2015 study conducted in Pakistan, The Club Foot is strongly associated with consanguineous marriages and has a heterogeneous geographical distribution and family history (Bhatti et al., n.d.). Looking at previous literature, A retrospective study from 2015 to 2019 of 18,515 births at King Saud Medical City found that clubfoot affected 2.3/1000 (0.23%) of babies born to Saudis at KSMC. Forty-two patients with clubfoot were born at KSMC out of the 100 cases that underwent evaluation at the clinic. Thirty-one percent of these patients were from consanguineous marriages (Fakeeha et al., 2021). Similarly, research on 89 people in Abha City between August 2022 and March 2023 discovered that almost two-thirds of the cases were male and that only three of the patients had a family history of CTEV (Kardm et al., 2023) .

We noticed there are not many studies or statistics available on Clubfoot in Saudi Arabia and Al-Baha. The purpose of this study focused on looking at congenital defects associated with clubfoot as well as the prevalence and risk factors of the disorder in Al-Baha City. The goal of the study was to provide the basis for upcoming studies on clubfoot in the region.

Methods

Study Design and Area

The current patient records pertaining to the diagnosis of clubfoot are analyzed in this study using a retrospective database design. The period covered by the research is January 2020–January 2024. The research was conducted in King Fahad Hospital in Al-Baha region.

Study Population

All individuals in the Al-Baha region were diagnosed with clubfoot within the specified time.

Sampling Frame

Patient records from King Fahad Hospital in Al-Baha, those with clubfoot diagnoses, are included in the sampling time frame.

Data Collection

We collected data using a customized data sheet that included all necessary elements. A clinical case sheet provided a comprehensive summary of each patient's medical background by summarizing their clinical history, examination results, and treatment specifics. Checklists were included to improve the consistency of the data collection procedure and ensure that all pertinent data points were gathered in an organized manner.

Data Collection Tool

The data collection tool included parental characteristics, specifically consanguinity between parents, important risk factors, and the existence of similar conditions or other anomalies in the family, as well as demographic data like gender and place of residence. Clinical data, such as the type and degree of the deformity and any associated conditions, was obtained. Along with recording the term of the pregnancy, the method

of delivery, and the fetal presentation, the sheet also evaluated any maternal exposures that may have occurred during the trimesters. Neonatal data is collected, including the baby's weight at birth, the age when the problem was first noticed, and when treatment commenced. Also included in the details are the treatment plan and the child's present state, including whether they need assistance or are ambulating regularly. Finally, a log of any issues that arose during the course of treatment.

Inclusion Criteria

All patients were diagnosed with clubfoot and lived in the Al-Baha region between January 2020 and January 2024 in King Fahad Hospital.

Exclusion Criteria

Patients who were not diagnosed with clubfoot or who do not live in the Al-Baha region.

Data Analysis

Descriptive statistics, such as counts, proportions (%), and mean values with standard deviations, were utilized in the research study to summarize the data. The Statistical Package for Social Sciences (SPSS), version 28, was used for all statistical analyses.

Ethical Considerations

The research study complied with ethical standards for retrospective studies and was authorized by the research committees at Faculty of medicine Al-Baha University under reference number [REC/SEC/BU-FM/2024/43]. Because of the nature of the study, participants' privacy was protected by anonymizing data to prevent identification and keep privacy, and informed consent was acquired using pre-existing medical information. Since the study only involved the analysis of pre-existing data and no direct participant engagement, there is no known risk associated with it.

Result

Demographic Information

Based on the demographic data of 17 patients with clubfoot identified in the Al-Baha region, 94.1% of the patients (n = 16) are male. Albaha accounted for the majority of patients (76.5%, n = 13), with lesser patients from Al Aqiq (5.9%, n = 1), Al Makhwah (5.9%, n = 1), Baljurashi (5.9%, n = 1), and Qilwah (5.9%, n = 1). Regarding parental consanguinity, 47.1% (n = 8) had consanguineous parents (Table1).

Table 1: Demographic Information

Items		n	%
Gender	*Female*	1	5.90%
	Male	16	94.10%
Residency	*Al Aqiq*	1	5.90%
	Al Makhwah	1	5.90%
	Albaha	13	76.50%
	Baljurashi	1	5.90%
	Qilwah	1	5.90%
Consanguinity between parents	*No*	9	52.90%
	yes	8	47.10%

Medical History and Risk Factors

A majority of the patients, 76.5% (n = 13), exhibited bilateral deformities, while 5.9% (n = 1) had a left foot deformity and 17.6% (n = 3) had a right foot deformity. In terms of risk factors, 11.0% (n = 2) were twins, and 5.5% (n = 1) reported preeclampsia, oligohydramnios, or gestational diabetes mellitus.

Table 2: Medical History and Risk Factors

Items		n	%
Deformity	*Bilateral*	13	76.50%
	Lt foot	1	5.90%
	Rt foot	3	17.60%
Risk Factors	*Twins*	2	11.00%
	Preeclampsia	1	5.50%
	Oligohydramnios	1	5.50%
	Gestational DM	1	5.50%
	None	13	72.50%

	congenital short Achilles tendon in the brother	1	5.90%
Similar condition? any deformities in the family	*None*	11	64.70%
	Sister	1	5.90%
	The father has same condition, and this condition runs in father's family	3	17.60%
	Yes (Not specified)	1	5.90%
Associated Medical Condition	*Asthma*	3	16.70%
	Down syndrome	1	5.50%
	Spina bifida	1	5.50%
	Undescended testis	3	16.70%
	Nothing reported	10	55.60%

Regarding family history, 5.9% (n = 1) reported a congenital short Achilles tendon in a brother, while 64.7% (n = 11) had no similar conditions in the family. Additionally, one patient reported a sister with a similar condition, and 17.6% (n = 3) indicated that their father had the same condition. The associated medical conditions are as follows: 16.7% (n = 3) of patients had asthma, while 5.5% (n = 1) had Down syndrome and another 5.5% (n = 1) had spina bifida. Additionally, 16.7% (n = 3) of patients were reported to have undescended testis (Table 2).

Obstetric Information

According to the obstetric data, 17.6% (n = 3) of the babies were preterm (less than 37 weeks), and 82.4% (n = 14) were considered full term (40±2 weeks). Regarding mode of delivery, 35.3% (n = 6) had a caesarean section, whereas 64.7% (n = 11) experienced a normal spontaneous vaginal delivery (NSVD).

Table 3: Obstetric Information

Items		n	%
Term	*Full term (40+/-2 weeks)*	14	82.40%
	Preterm (less than 37 weeks)	3	17.60%
Mode of Delivery	*Caesarean section*	6	35.30%
	normal spontaneous vaginal delivery (NSVD)	11	64.70%
Why Caesarean section	*CS due to previous CS*	2	11.80%
	oligohydramnios	1	5.90%
	Preeclampsia	1	5.90%
	Severe vomiting which leads to hypotension	1	5.90%
	Twins	1	5.90%
Fetal presentation	*breech presentation*	2	11.80%
	cephalic presentation	14	82.40%
	transvers presentation	1	5.90%
Exposure during the Trimesters	*None*	14	82.40%
	Drug	1	5.90%
	Infection	2	11.80%
If yes, in which trimester	*None*	14	82.40%
	1st	1	5.90%
	2nd	2	11.80%
baby weight	*Fetal macrosomia*	1	5.90%
	Low Birth weight	6	35.30%
	Normal range (2.5 - 3.5 kg)	10	58.80%

Preeclampsia (5.9%, n = 1), oligohydramnios (5.9%, n = 1), severe vomiting resulting in hypotension (5.9%, n = 1), and twins (5.9%, n = 1) were the reasons for cesarean births. 82.4% (n = 14) of the fetuses presented in the cephalic position, with 11.8% (n = 2) presenting breech and 5.9% (n = 1) presenting transversely. 5.9% (n = 1) of participants reported drug exposure during the first trimester, and 11.8% (n = 2) reported infections during the second trimester. In terms of baby weight, the distribution was as follows: 5.9% (n = 1) had fetal macrosomia, 35.3% (n = 6) had low

birth weight, and 58.8% (n = 10) had weights between 2.5 and 3.5 kg that were considered normal (Table 3).

Clinical Timeline and Current Status

Table 4 shows when the problem was first noticed: 94.1% (n = 16) reported recognition immediately after birth, while 5.9% (n = 1) identified it during pregnancy. There were variations in when the therapy was started: among the five, 29.4% (n = 5) began in the first week, while another 29.4% (n = 5) began by the eighth week. The other initiation times were the following: the second week (5.9%, n = 1), the fourth week (17.6%, n = 3), the twentieth week (11.8%, n = 2), and the twenty-fourth week (5.9%, n = 1). The treatment plans were 100.0% (n = 17) undergoing serial casting and Achilles tenotomy.

Current status indicates that 41.2% (n = 7) are walking normally, while 23.5% (n = 4) are unable to walk. Additionally, 11.8% (n = 4) cannot walk normally due to suspected neurological problems, and another 11.8% (n = 2) can walk well, but the left foot is still a little crooked. The complications were 5.9% (n = 1) experienced hyperflexibility, while another 5.9% (n = 1) had leg length inconsistency. A significant 41.2% (n = 7) reported an inability to walk, and 17.6% (n = 3) experienced tightness of the tendon. Additionally, 29.4% (n = 5) reported no complications.

Discussion

Congenital talipes equinovarus (CTEV) or clubfoot is one of the prevalent musculoskeletal deformities characterized by equinovarus (turned in foot) or cavus (longitudinal arch). It is a congenital condition, meaning it is evident at birth where a newborn's foot is twisted, losing the normal shape of the foot. CTEV is a significant contributor to disability, impeding the child's quality of life and potentially extending into adulthood (Alomran et al., 2024). This calls for a thorough assessment of the condition to devise and implement a multidisciplinary intervention that holistically assists the affected. Our study targeted the Al-Baha region of Saudi Arabia and included 17 patients.

According to this study, 76.5% (n = 13) of the patients had bilateral deformities, 17.6% (n = 3) had a deformity on the right foot, and 5.9% (n = 1) had a deformity on the left foot. These results were considered high compared to other studies. For instance, according to Kardm (Kardm et al., 2023), 40.5% of the cases were bilateral. We collected obstetric information, which revealed variations in the participants. Among the 17 participants, 14 or 82.4% were born full term, i.e., over 40 weeks old, and 17.6% or 3 were born preterm or before 37 weeks. 11 participants, or 64.7%, were born through normal virginal delivery, whereas 6 participants, or 35.3%, were born through caesarean delivery. Other factors considered were the child's position, birth weight, drug use, and complications during various trimesters.

Research in different parts of the world reveals that clubfoot is a prevalent condition among the populations. However, the prevalence differs based on country and specific populations. Studies from different countries and populations agree that the condition is higher in males than females. In particular, the research indicates that the condition is more common among males than females. For example, about 56% of the participants with the condition were male, and 42% were female (Ugorji, 2020). Bilateral CTEV was the most common at 75% among the patients. Research in Denmark revealed that the prevalence was 1.52 per 1000 live births (Hedley et al., 2023). Another study in the Czech Republic placed the prevalence at 1.9 per 1000 live births (Janatová et al., 2023). The research conducted for 14 years differed based on regions. At the global level, the prevalence of the condition was estimated to be 0.6% to 1.5/1000 live births (Moteb et al., 2022). These studies align with our research, where 16 out of 17 patients were male, showing the prevalence.

The research explored different risk factors contributing to clubfoot. Two, or 11%, of the participants had twins, preeclampsia,

oligohydramnios, and gestational diabetes mellitus, at 5% or 1 in each case. One participant reported having a sister with the condition, as well as three whose fathers were suffering. However, 11 participants did not exhibit any of the risk factors. Three of the participants had asthma, three had undescended testicles, and one each had spina bifida and Downs syndrome.

Different studies have shown that some genes are more prone to CTEV than others. For instance, COL1A2 and AKT3 genes are attributed to the condition (Wang et al., 2022). This is because these genes are involved in a variety of bone formation-related activities. For instance, the genes are responsible for bone metabolism, toughness, and osteogenesis. The research investigated the upregulation and downregulation of these two genes, revealing their impact on the development of children. It therefore implies that interference with the genes plays a great role in developing CTEV by impacting the role of bone formation (Mustari et al., 2022). A comprehensive meta-analysis, which included 8 studies, 833 CTEV patients, and 1280 healthy individuals, investigated the role of genetics in CTEV. The research found that there is a close relationship between COL9A1 polymorphisms and CTEV. Children exposed to this gene, specifically rs1135056 and rs35470562, were more susceptible to the condition (Golshan-Tafti et al., 2024). The results explain why the twins in the case study may have suffered from the condition. They likely inherited the genes from the parents.

Although clubfoot may be an isolated condition caused by different factors, some of them stood out among the population. For instance, some diseases like asthma, Down syndrome, twining, maternal diabetes, and parental consanguinity. These were similar to findings from retrospective research, which also attributed the condition to factors related to the parent and diseases. According to this study, monozygotic twins are more likely to suffer from the condition compared to dizygotic twins in (Kardm et al., 2023). This further reinforced our findings showing that twining is likely to cause the condition. The most common reason for the condition in twins apart from genetics is the position of the children while in the womb.

The parents have been identified as some of the carriers of the most risk factors. Parents are genetically exposed to the condition, increasing the prevalence of the condition. Other parents, on the other hand, expose their children to social and environmental factors leading to clubfoot. Some of the parental factors leading to the condition include maternal chronic illness and familial predisposition (Van Schelven et al., 2021). Although these were not significant in the study, they were impactful to the condition. However, it was found that dysplasia of the hip (DDH) was closely related to the condition. Individuals with DDH were two and a half times more likely to suffer from the condition compared to those without the condition.

Research in Norway involving 60,844 children born between 1996 and 2012 also found a close relationship between DDH and clubfoot. This research found that 4.3% of the children with a leg deformity also experienced DDH (Håberg et al., 2020). These conditions are, however, not limited to the children but also transmitted through the parents to their offspring. Parents also have the responsibility of examining the children to identify if they are suffering from the condition. Our research found that 8 out of 17 children had a **consanguinity** with the parent. This aligns with findings by **Saini** (Saini et al., 2023), where 1 (5%) of the patients was from a consanguinity marriage. Prenatal examination plays a crucial role in promptly identifying the condition. Congenital anomalies have been identified as some of the most common features of the disease. Some children experience different levels of complications. Some are highly flexible; others suffer inconsistency when walking, and some have short legs (Alosaimi et al., 2022). Therefore, it is necessary to address the risk factors in order to save them.

Recommendations

Most of the risk factors are closely related to maternal and paternal health. We suggest that educating parents can be a proactive measure in evaluating their health and tackling the social, genetic, and environmental factors that can exuberate the problem (Alasbali et al., 2023). Early screening is necessary to address it early (Mohsenh et al., 2023). The parent should ensure the child is treated after diagnosis (Dreise et al., 2023).

Limitations

There are several limitations to this study. First, it was conducted in a single hospital within one region of Saudi Arabia, which limits the generalizability of the findings to the broader population. Second, the sample was predominantly male, with 16 out of 17 participants being male and only one female, making it challenging to draw conclusions applicable to both genders. Third, the sample size was small (17 participants), which may reduce the statistical power and reliability of the results. A larger sample size would strengthen the robustness and generalizability of the conclusions. Finally, the study's exploration of genetic factors was limited to consanguinity, missing an opportunity to gain deeper insights into hereditary influences on clubfoot.

Conclusion

The research provides valuable insights into the prevalence and risk factors for clubfoot in Saudi Arabia's Al Baha region. The population under study had a high rate of the condition. There is a high number of patients with consanguinity, indicating they may have acquired the condition from the ancestors in the parental lineage. Despite not directly revealing the prevalence of the condition in the population, the research did indicate its prevalence, given that the participants came from a single region. Some people are at a higher risk of suffering than others based on genetic, social, and environmental predispositions. Interventions must be implemented to manage the condition. The condition affects children at a very young age. It is imperative to implement educational interventions aimed at assisting parents in identifying and managing this condition early. Education during the prenatal stage can inform parents about the habits they should avoid protecting their children. Genetic screening can also be useful because it helps manage the genes that contribute to bone deformities. Public awareness through the internet and social sites can be a powerful tool. This should inform society about the risky populations and mitigation mechanisms. However, some children have already been affected and are enduring suffering. Stabilizing the health of the children requires comprehensive interventions. CTEV can be destabilizing since it interferes with normal life. Healthcare professionals need to identify interventions that serve diverse populations. However, further studies are necessary to determine the prevalence both regionally and nationwide. The studies should accommodate all the populations, including males and females. They will create a clear picture of the condition and compare it with other nations where the disease prevalence is well known.

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