

Parental Consanguinity and Increased Risk of Idiopathic Congenital Talipes Equinovarus (CTEV)

Idiopathic
Congenital
Talipes
Equinovarus

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ABSTRACT

Objective: The objective of this study was to establish relationship of parental consanguinity as a risk factor for congenital Talipes Equinovarus.

Study Design: Prospective study

Place and Duration of Study: This study was conducted at the Department of Orthopedics Surgery, Lady Reading Hospital Peshawar from 1st July 2014 to 30th June 2015.

Materials and Methods: This study comprised 140 patients. Patients with CTEV presenting to the outdoor department were included and data was taken through a simple questionnaire including consanguinity and nonconsanguinity.

Results: There were 86 (61.42%) males and 54 (38.57%) females. Eighty eight (62.85%) patients have bilateral deformities while the rest 52 (38.57%) patients have unilateral deformities. out of the 52 patients with unilateral deformity. The right side was involved in 37 (71.15%) patient and the left side in 15 (28.84%) patients. Out of these 140 patients with CTEV, 71 (50.71%) patients were born to parents having cousin marriage while the rest of 69 (49.28%) had marriages outside their family.

Conclusion: Consanguineous marriage was significantly associated with an increased risk of idiopathic CTEV. Even after adjusting for potential confounding variables the association remained significant.

Key Words: CTEV; Consanguinity; Idiopathic

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INTRODUCTION

Congenital talipes equinovarus, is one of the most common congenital orthopedic anomaly. Isolated talipes equinovarus, has a birth prevalence of 1.29 per 1,000 livebirths.¹ This congenital anomaly has a male-to-female ratio of 2:1.²⁻⁴ Both the limbs are involved in upto 30-50% of cases. It has been reported that the involvement of right leg is more than the left leg. Chances of a subsequent child to be affected with CTEV increases by 10 % if the parents already have a child with a clubfoot.^{5,6}

CTEV is usually diagnosed on clinical examination (equinus of the hind foot, varus of the hind foot, adduction of the forefoot, cavus of the mid foot).⁷⁻⁹ In utero ultrasound can also be used to make a diagnosis.¹⁰ Congenital clubfoot has no established cause. Although it is suggested that clubfoot has many risk factors, genetic factors clearly are important.⁷

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Most infants who have clubfoot have no genetic or extrinsic cause. Cytogenetic abnormalities (such as congenital talipes equinovarus) involving chromosomal deletion is seen to be a part of different syndromes. It has been suggested that idiopathic CTEV is usually the result of a multifactorial system of inheritance.¹¹ Pregnant women having low folate level is identified as a cause of several congenital malformations, and prevalence of idiopathic CTEV was observed to be reduced after fortification of grains with folic acid, or supplementation in the United States and Denmark.¹² Correlation has been identified between the incidence of CTEV and maternal smoking during pregnancy.^{13,14,15} Maternal smoking is suggested to cause reduction in fetal and placental circulation due the vasoactivity of nicotine, abnormal arterial pattern in the affected foot and fetal tissue hypoxia.¹⁶

Consanguinity, the marriage of individuals with a common ancestor, often has genetic implications for offspring. Several articles have suggested consanguinity as one of the important causal factor in birth defects, including CTEV. In our society the tradition of cousin marriages is more common as compared to the western countries. We found that the occurrence of CTEV is much more common in children who are born of parents who had cousin marriages as compared to others. So we conducted a prospective study to establish the association of CTEV with cousins marriages.

MATERIALS AND METHODS

This prospective study was conducted in the Orthopaedics Department of Lady Reading Hospital Peshawar, over period of one year from 1st July 2014 to 30th June 2015. The study included all the patients who presented to the outdoor patient department. The diagnosis of the CTEV was purely on clinical grounds. After the diagnosis of CTEV was established, data was collected on proforma already designed for this purpose. The proforma used for data collection was simple and included the biodata of the patient along with a simple inquiry from the parents about the cousin marriage. Each patient's family was interviewed in the OPD by a structured questionnaire including demographic and consanguinity information. Informed consent was taken from the parents of the children to be included in the study.

RESULTS

Table No. 1: Descriptive statistics of the patients

Variable	No.	%
Gender		
Male	86	61.42
Female	54	38.57
Deformity		
Bilateral	88	62.85
Unilateral	52	37.14

Table No.2: Side of unilateral deformity (n – 52)

Unilateral deformity	No.	%
Right side	37	71.15
Left side	15	28.84

Table No.3: Frequency of consanguinity of the patients

Consanguinity	No.	%
Non-consanguineous marriage	69	49.28
Consanguineous marriage	71	50.71

Table No.4: Comparison of consanguinity of patients according to genders

Consanguinity	Male	Female	Total
Non-consanguineous marriage	47 (68.11%)	22 (31.88%)	69 (49.28%)
Consanguineous marriage	39 (54.92%)	32 (55.07%)	71 (50.82%)
Total	86 (61.42%)	54 (38.57%)	140 (100%)

A total of 140 patients with CTEV presented to the outdoor patient department during one year mentioned study period. Out of 140 patients 86 (61.42%) were males and 54 (38.57%) were females. Eighty eight (62.85%) patients have bilateral deformities while the rest 52 (38.57%) patients have unilateral deformities. Out of the 52 patients with unilateral deformity. The

right side was involved in 37 (71.15%) patient and the left side in 15 (28.84%) patients. Out of these 140 patients with CTEV, 71 (50.71%) patients were born to parents having cousin marriage while the rest of 69 (49.28%) had marriages outside their family.

DISCUSSION

Talipes equinovarus is a congenital disorder of the foot and ankle that may be unilateral or bilateral. The fore foot is adducted, hind foot in varus and equinus and mid foot in cavus.¹⁷ TEV is often an isolated finding (ITEV) or it may occur as a part of a different genetic syndrome. The management of CTEV become more difficult when it occurs as part of a syndrome or with other abnormalities (such as neural tube defects, i.e. spina bifida).¹⁸

This study further evidence provide support of a causal role of Parental consanguinity in clubfoot. It has been identified that Parental consanguinity is associated with isolated clubfoot. The causative factor of isolated clubfoot, or idiopathic talipes equinovarus, is unknown, but different etiologic theories had been previously suggested in the literature.¹⁹ Male gender had been suggested to have a high risk of involvement, with a 2:1 (male:female) ratio found across different demographic groups.⁷ In our study, the ratio of male to female is 1.6:1. Mechanical forces due to insufficient expansion of the uterus and low amniotic fluid volume is also associated with CTEV. Increased incidence is seen in first-born children and in pregnancies in which there is low amniotic volume and twin pregnancies.^{20,21} Advanced maternal age is also an important risk factor for ITEV.²² CTEV is common in First-born children than children from subsequent pregnancies.²³ The effect of maternal smoking on the risk of clubfoot are consistent in the literature. Previously studies have identified associations between maternal smoking and foot deformities that was statistically significant.^{14,24} An association between CTEV and maternal anemia has also been mentioned in a study.²¹

A relationship between consanguineous marriages and idiopathic congenital talipes equinovarus has previously been established. A study conducted in the rural area of Turkey reported that babies born of 1st cousins parents have 4 times more risk of developing CTEV as compared to the babies who are born of parents who are not relative to each other.²⁵ Another study conducted by Sreenivas et al reported that Out of 174 children included in their study with CTEV, 54 (31%) children were born out of consanguineous marriage.²⁶ In our study we found that out of these 140 patients with CTEV, 71 (50.71%) patients were born to parents having cousin marriage while the rest of 69 (49.28%) had marriages outside their family. 86 (61.42%) patients were male and 54 (38.57%) were female with a male to female ratio of 1.6:1. Eighty Eight 88 (62.85%) patients have bilateral deformities while the

rest 52 (37.14 %) patients have unilateral deformities. Out of the 52 patients with unilateral deformity, the right side was involved in 37 (71.15%) patient and the left side in 17 (28.84%) patients.

CONCLUSION

Consanguineous marriage was significantly associated with an increased risk of idiopathic CTEV. Even after adjusting for potential confounding variables the association remained significant.

Author's Contribution:

Concept & Design of Study: Sajid Akhtar
 Drafting: Anwar Imran
 Data Analysis: Faiz Ali Shah
 Revisiting Critically: Shahab ud Din
 Final Approval of version: Sajid Akhtar

Conflict of Interest: The study has no conflict of interest to declare by any author.

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