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RESEARCH ARTICLE

ROLE OF PARENTAL CONSANGUINITY IN DEVELOPMENT OF CONGENITAL TALIPES EQUINOVARUS

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ABSTRACT

Introduction: Congenital clubfoot is one of the most common congenital anomalies encountered in orthopaedics. Its etiology has been proposed to be multifactorial. We studied the association of various factors, with special reference to consanguinity between parents as the possible etiological factor in the development of this condition. **Materials and methods:** 78 children with congenital talipes equinovarus were included in our study, and a detailed history was taken, with emphasis on parental consanguinity, and an examination was done to assess the idiopathic, or non-idiopathic nature of clubfoot. **Results:** Idiopathic CTEV was the most common type diagnosed. Of the 78 children with CTEV, 21 (27%) were born out of consanguineous marriages. Only 1 out of those 21 children had a non-idiopathic CTEV. **Conclusions:** There is a high incidence of parental consanguinity among parents in the children diagnosed with congenital clubfoot, in our region. However, parental consanguinity was not associated with increasing severity of clubfoot. More studies, and possibly genetic testing, however, are needed to clarify the association of disease severity with parental consanguinity.

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INTRODUCTION

Clubfoot, or congenital talipes equinovarus (CTEV) is one of the most common orthopaedic anomalies, the description of which goes back to 400 B.C., when Hippocrates first described the condition. The incidence of clubfoot is 1-2 per thousand live births (Gurnett, 2008). It is bilateral in about 50% cases.It is approximately twice more common in males than in females (Gurnett et al., 2008; Lochmiller et al., 1998). The deformity has four components; forefoot adductus, midfoot cavus, hind foot varus, and ankle equinus (Ponseti et al., 1963). The deformity, though easily recognizable at birth, can now be diagnosed by antenatal ultrasonography at 18-20 weeks of gestation (Bakalis et al., 2002). Several theories have been proposed so as to explain the etiology of clubfoot. One of the theories (Gurnett et al., 2008; Lochmiller, 1998), which has also been seconded by Ponseti (Ponseti, 1963) suggests that clubfoot is the result of an arrest during the development of foot during intra-uterine life. Another view (Palmer) is that there is a multifactorial system of inheritance, with the possibility of intra-uterine factors having some effect (Wang, 1988).

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Yet another proposed explanation is the Polygenic theory, which was supported by Davis, and he showed there was a decrease in the incidence of clubfoot from first to second to third degree relatives (Wynne-Davies, 1972). Insley (1967), in his paper reported the association of clubfoot with a deficiency of part of long arm of chromosome 18. Wang et al. (2013), in his paper proposed a potential role of SOX9 over expression in idiopathic congenital talipes equinovarus. Consanguineous marriage is the marriage of individuals having common ancestor(s). It is known to predispose offspring to congenital disorders (Rittler et al., 2001), supposedly by inheriting identical copies of mutant alleles of autosomal recessive disorders. The purpose of the present study was to make observations on the incidence of parental consanguinity in patients of CTEV, and make note of associated factors in addition to it; especially as consanguineous marriages are not uncommon in this region.

MATERIALS AND METHODS

This study was conducted at the Government Medical College - Hospital for Bone and Joint Surgery, Srinagar, a tertiary orthopaedic referral centre in the Kashmir division of the Indian state of Jammu & Kashmir. The study period was from June 2017 to June 2019.

During the study period, a total of 108 patients who were less than 1 month old, presenting to our out-patient department first with complaints of a deformed foot were evaluated by a team of three Orthopaedic residents. The patients with any of the known or suspected causes of clubfoot were also included in our study (central nervous system defects, meningomyelocele, meningocele, arthrogryposis multiplex congenita, metabolic disorders, and neuromuscular abnormalities). Postural clubfoot was excluded from our analysis. After the exclusion process, 78 patients of congenital talipes equinovarus remained in the study. Before including the babies in our study, a detailed, informed consent regarding the inclusion of their data in our study was taken from their parents. A detailed parental history was taken and a full physical examination was done subsequently, and the findings were recorded. History included details of the baby, e.g. birth order, enquiring about consanguinity between the couples, and family history of similar deformities. Examination included scoring the feet by the Pirani scoring system (World Health Organization, 2019) for clubfeet. This scoring system took into consideration mid-foot and hind-foot deformities only. Three signs for Mid foot include a deep medial crease, curved lateral border, and coverage of the lateral part of the talar head. Three signs for Hind foot include posterior creases, empty heel, and rigid equinus. Each is scored according to the following principle:

0, no abnormality.
0.5, moderate abnormality.
1, severe abnormality.

Thus, each foot can have a Mid foot score between 0-3 and a hind foot score between 0-3 and a total score between 0-6
All of these patients were managed by the Ponseti casting technique.

Type of CTEV	Bilateral	Right	Left
Idiopathic	42	22	8
Secondary	6	0	0

RESULTS

A total of 78 children presented to our department with clubfeet. Out of these 78 patients, 21 children (27%) were born out of consanguineous marriage. The most common type of consanguinity was marriage between first cousins (14 couples). This implies uncle's son marrying aunty's daughter, or vice versa. Remaining couples were marriage between second cousins. Out of the 78 children evaluated, 48 patients (61.5%) had bilateral CTEV, whereas the remaining 30 patients (38.5%) had unilateral CTEV. 8 were left sided and 22 were right sided. There was a male predominance in our patients, 54 patients (69.2%) were males, and 24 patients (30.8%) were females. Idiopathic CTEV was by far the most common type diagnosed. 72 patients were diagnosed with Idiopathic CTEV, and 6 patients had secondary CTEV.

Of the 6 patients with non idiopathic CTEV,

- One child was born out of a consanguineous marriage
- 3 of the patients with non-idiopathic CTEV were males, 3 were females
- 2 patients had AMC, 2 patients had neural tube defects, 1 patient had polydactyly, and 1 was associated with congenital deformity of upper limb

49 patients out of 78 in our study were first born. 29 patients in our study had a Pirani score of 6/6, 29 patients had a score of 4.5/6, 20 patients had a score of 3/6 at presentation. All of the cases with non idiopathic CTEV had Pirani scores of 6/6. Only 1 case in our study was diagnosed by antenatal ultrasound before delivery, at 22 weeks of gestation. Subsequent amniocentesis was not done. Deformities of all other patients were noted at birth, and orthopaedic advice sought thereafter. No mother in our study had a history of addiction to tobacco, alcohol or any other substance.

DISCUSSION

In a study conducted by Sreenivas et al. (2012), in 2012, 31% of CTEV children in their study had been born out of consanguineous marriages. In our study also, 27% of children have a history of parental consanguinity. Whether this has a direct role in the causation of CTEV is yet to be determined, as idiopathic CTEV is a polygenetic condition. In the same study by Srinavasan et al, on consanguineous couples, the distribution of CTEV in children born out of consanguineous marriages was: 53.7% idiopathic, and 46.3% non-idiopathic. This incidence of non-idiopathic CTEV was higher than that noted in general population. However, our study showed that only 1 child had non-idiopathic CTEV, out of 21 children with CTEV born out of consanguineous marriages (4.76%). The number of patients however, in our study, was too low to draw a statistical conclusion. The incidence of non-idiopathic CTEV in consanguineous couples in our study was only 4.76%.

In his study published in 1972, Wynn Davies et al. (1972) concluded that congenital talipes equinovarus had a multifactorial inheritance pattern, wih a male: female ratio of 2:1. Gurnett et al. (2008), reported a male: female ratio of 2:1, and Lochmiller et al. (1998) reported male:female ratio of 2.5:1. In our study, the male: female ratio was 2.7:1, and it is comparable to previously published reports of a male preponderance of the condition. However, non-idiopathic CTEV in our study had a 1:1 male: female ratio. In the study by Gurnett et al, bilateral CTEV was more common than unilateral, in both idiopathic, and non-idiopathic group. Majority of unilateral CTEV was idiopathic in their group. In our study also, it was found that bilateral CTEV was more common in the idiopathic group. And all of the non-idiopathic CTEV patients had bilateral deformities. CTEV is the most common deformity noted in patients of spina bifida, and frequency varies from 7 to 40% depending on the method of diagnosis (Hunter, 1984). Preconceptional folic acid supplementation has been proposed to reduce the risk of neural tube defects, as shown in the study by Czeizel et al. (1998). 70 mothers in our study took folic acid during pregnancy, but none of them had a history of folic acid intake in the preconceptional period. Maximum number of children with CTEV in our study were firstborn (45%), followed by second born children. In the study by Gurnett et al, 76% patients had idiopathic CTEV, and 24% had secondary CTEV. The prevalence of secondary CTEV varies from 11 to 48 %, in different series (1,5,14). In our study, 7.6% patients had non-idiopathic CTEV, whereas 92.3% had idiopathic CTEV.

Conclusion

Our study on the associations of CTEV in our population showed that there is high incidence of parental consanguinity, as is widely practiced in our region.

However, the proportion of non idiopathic CTEV in our study was less than that observed in literature. Also, the proportion of non-idiopathic CTEV in consanguineous couples was also lower than previously described. More studies, and possibly genetic testing, however, are needed to clarify the association of disease severity with parental consanguinity. Also, the practice of consanguineous marriages needs to be dealt with on a societal basis to avoid development of congenital disorders in general.

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