Vol-5, Issue-4 PP. 123-126 ISSN: 2394-5788

GLOBAL JOURNAL OF ADVANCED RESEARCH
(Scholarly Peer Review Publishing System)

SPINA BIFIDA WITH TALIPES EQUINOVARUS

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ABSTRACT

Talipes equinovarus is a most common congenital defect. Today it is not only clinicians and physicians who desire to understand the details of diagnosis and treatment, but also the parents of affected children, as because of the inherent economic and emotional burden of a clubfoot diagnosis but also for proper compliance with treatment protocols to prevent future reoccurrences. The cause of this condition is less known. Different mechanisms have been proposed i.e., neurological, bony, muscular, connective tissue and vascular. However the only evidence is that the milder cases are associated with posture within uterus. There is a proof available for the aetiology of congenital Talipes equinovarus genetically. Complex segregation analysis suggests that the most likely inheritance pattern is a single gene of major effect operating against a polygenic background.

Spina bifida is type of birth defect where there is an incomplete closing of the spinal column and meninges around the baby's spinal cord. There are four types of spina bifida: Occulta, closed neural tube defects, meningocele and myelomeningocele. The most usual location is the lumbosacral region. Meningomyelocele is the most severe form. The diagnosis by a simple ultrasound can be correct especially if abortion is considered, and a prenatal MRI scan should be done prior to counselling and before considering the surgical treatment. A Pregnant women underwent a routine ultrasound examination at 19 weeks of gestation. Ultrasound of fetus showed lower midline lesion suspect of meningomyelocele. More ultrasounds were done which showed spina bifida with congenital Talipes equinovarus of right lower limb. Pregnancy was terminated at 19 plus weeks I/V/O. On physical examination of aborted fetus a 2 X 2 X 1 cm midline mass was seen in lumbosacral region covered with meninges.

Keywords: Spina bifida, Talipes Equinovarus

1. INTRODUCTION

Spina bifida (SF) is a neural tube defect and it occurs when the vertebrae don't form properly around part of the baby's spinal cord. SF is found in 2-4/1000 live births- Laurence K (1989). In this disorder a sac composed of meninges- dura and arachnoid mater filled with CSF herniates through a defect in the posterior spine. Its types include spina bifida occulta, meningocele and meningomyelocele. The most common location is the lumbosacral region as cited by Doran PA et al., (1961); Ersahin Y et al., (2001); Kumar R et al., (2003). Occulta is the mildest and most common form and does not usually causes disability or symptoms. Meningocele typically causes mild problems with a herniated dural sac filled with fluid present at the gap in the spine. Meningomyelocele is the most severe form in which the dural sac also contains spinal cord with nerve roots (Doran PA et al., 1961). Foot disorders are quite common in patients with spina bifida. About 30% of children with SF are born with a club foot deformity- Congenital Talpesequinovarus (Joae de CN et

al., 1996). Congenital Talipes equinovarus (CETV) is syndromic when it is associated with the features of genetic syndrome or it can occur in isolation termed as idiopathic. Syndromic CTEV arises with spina bifida, but idiopathic is far most common where is upper limb is normal (Zosia M 2003). In CETV the infant one or both feet are quite rigid, excessively plantar flexed and swing inwards. In the majority of cases, infants appear with only the deformation of the foot or feet, and present clinically without any other grossly

Visible deformations which is termed clinically as isolated clubfoot, idiopathic clubfoot. Most clubfoot cases are of a male predominance and present bilaterally, but when clubfoot presents unilaterally, most often the right leg and foot are affected as opposed to the left leg and foot. Females, although presenting with a statistically lower birth prevalence for clubfoot, are more likely to pass the deformity on to their children than males, and females are also more likely to have siblings with the disorder than are affected males. (Zosia M 2003).Diagnosis by a simple ultrasound can be misleading and in attaining the proper diagnosis especially if abortion is considered, a prenatal MRI scan should be therefore be carried out before counselling and should be repeated before to operative treatment. Surgical treatment of is not only for cosmetic reasons but also to untether spinal cord prophylactically to prevent further neurological damage. In the present report we identify an aborted fetus of SF with club foot.

2. CLINICAL CASE

Pregnant women who underwent a routine ultrasound examination at 19 weeks of gestation, fetus showed lower midline lesion suspect of meningomyelocele. More ultrasounds were done which showed spina bifida with congenital Talipes equinovarus of right lower limb. Because of expected problems to the child pregnancy was terminated at 19 plus weeks I/V/O. On physical examination of aborted fetus a 2 X 2 X 1 cm midline mass was seen in lumbosacral region covered with meninges. It did not contain any neural elements remarkably at the site of the lesion. The spinal cord was slightly tethered dorsally (Fig.1).CETV of right foot (Fig.2) confirmed by means of routine X- ray of the aborted fetus (Fig.3).

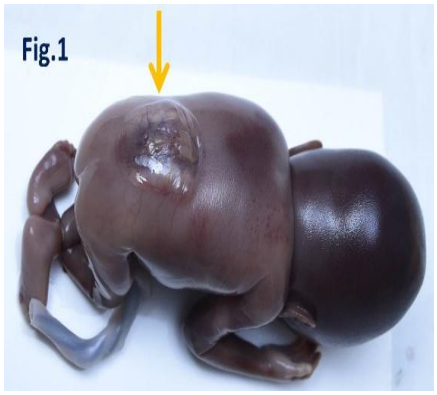

Fig.1.Photograph of the Physical examination showing 2x2x1cm mass covered with Meninges shown by arrow

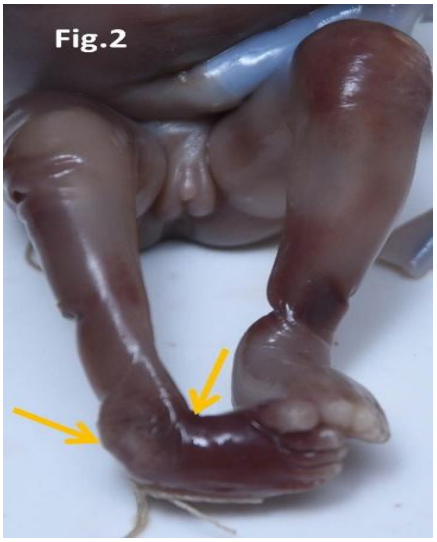

Fig.2.Photograph of aborted fetus demonstrating the Right sided talipes equinovarus, characterized via equines, varus, adduction and supination of foot.

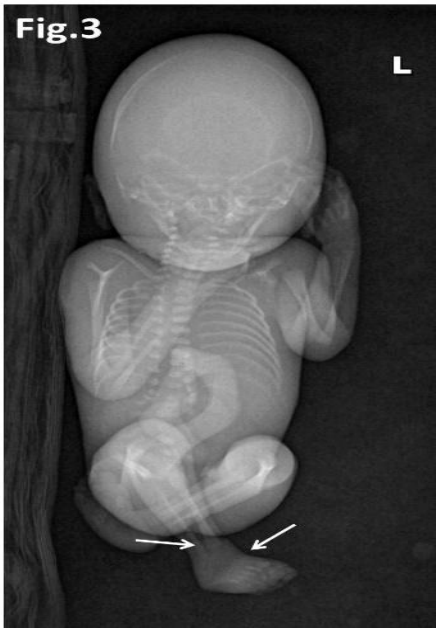

Fig.3.X-ray of Talipes equinovarus of right foot in which bony defect is shown. Note the arrows indicating the defect.

3. DISCUSSION

Spina bifida (SF) - a neural tube defect that appears in the fetus at 28th day of gestation. It is a congenital anomaly involving closure of neural tube with an incidence of 2-4/1000 live births (Laurence K 1989). SF are of different types-Spina bifida cystica where there is a protrusion of spinal cord and meninges with a defect in the vertebral arch. In Meningocele there is a normal development of vertebrae but the meninges (protective coverings around the spine) push out from the spinal opening, and may or may not be covered by a layer of skin. This can occur anywhere on the back but are commonly observed in the lumbar or lumbosacral region. In meningomyelocele sac also contains neural tissue. Meningocele is the most severe type of spina bifida where spinal cord is open and this is due to failure of neural folds to meet and unite resulting in flattening of spinal cord. Ersahin et al., reported 2 cases out of 22 spinal meningoceles located in the upper and midthoracic region and Doran and Guthketch (1961) reported 65 children with a spinal meningocele of which 7 were thoracic type. Steinbok P and Cochrane DD (1991) described two types of spina bifida cystica in the cervical region based on

gross anatomic observations as myelocystocele and meningocele. Pang D and Dias (1993) reported 9 cases of cervical myelomeningoceles and also stated the presence of band of tissue tethering the spinal cord to the adjacent dural sac. Since the sac of meningocele and myelocystocele is devoid of nervous tissue, children born with such a anomaly usually have no neurological deficit. However, neurological damage can occur due to associated anomalies like tethered cord, filum terminale thickening, Chiari malformation, hydrocephalus and hyderomyelia as cited by Ershin Y et al., (2001) and Delashaw JB et al., 1987). As spinal cord can be tethered posteriorly due a band of tissue running into the sac and hence therefore during surgical correction it is essential to perform a thorough intradural inspection to transect all the adhesions in order to prevent future damage due to spinal cord tethering (Delashaw JB et al., 1987).

Diagnosis of spina bifida prenatally by ultrasound is well established and can be done between 14-16 weeks of pregnancy (Unsinn KM et al., 2000). However the detection of all anomalies is limited and differentiating meningocele from meningomyelocele can be tough as above two types have no neuronal deficits and it is important to establish a correct diagnosis when a possible abortion is being di

The exact cause of clubfoot is unknown, but current theories suggest the deformity may be caused by **genetic** or environmental factors. Genetic factors tend to produce symmetrical bilateral clubfoot and often are associated with additional clinical findings and can arise from many different origins, including genomic additions such as Trisomy 18, genomic deletions such as del (17q23), neurologic malformations such as spina bifida, muscular conditions such as dystrophia myotonica, and connective tissue diseases and disorders including congenital joint contractures in two or many areas in the body (arthrogryposis). Environmental factors tend to be most commonly asymmetric in severity although both feet can still be affected to varying degrees. They impede the normal growth and position of the fetus in the maternal womb are implicated in congenital deformities such as clubfoot, and these include specifically uterine constriction due to scarring and fibrosis from multiple prior gestations, a breech position of the fetus in utero, amniotic bands, and oligohydramnios (Nemec U et al.,. 2012; Pagnotta G et al.,.2011).

Anatomy of Clubfoot

Clubfoot is anatomically defined as equinus, varus, adduction, and supination of the fetal foot. In this there is a subluxation of the talo-calcaneo-navicular joint with underdevelopment of the fascia and soft tissue elements on the medial side of the foot. Frequently, underdevelopment of the calf and peroneal muscles are also seen, which commonly results in shortening and fibrosis in the muscles of calf and back muscles of leg and their respected tendons. The ligamentous structures of the fetal foot are however unaffected in clubfoot deformities, although the bones of the fetal foot are often malpositioned and held in relatively fixed, stable positions by the strong and properly formed ligaments (El-Adwar KL, Kotb HT. s. 2010).

Prenatal Diagnosis of Clubfoot

For many years conventional radiography was used to assess clubfoot, however it has limited value because the bones in the feet of the child have not yet ossified and are therefore not radiopaque on plain radiographic films. The techniques used in ultrasound are still evolving in the prenatal determination of clubfoot, but ultrasound provides the observation of the cartilaginous components of the tarsal bones which are missed with conventional radiography. Currently, both transvaginal and transabdominal ultrasound are being used for prenatal clubfoot diagnosis in clinical practice. Transvaginal ultrasound can detect the abnormality for a prenatal diagnosis of clubfoot as early as 12-13 weeks gestation, and transabdominal ultrasound can detect the abnormality as early as 16 weeks gestation. Recently, fetal MRI has been investigated for enhanced detection of clubfoot, but thus farhas only seen real utility in patients presenting with complex clubfoot associated with a myelomeningocele (Nemec U et al. 2012) Fetal MRI has been found to be more sensitive than transvaginal and transabdominal ultrasound in the prenatal detection of clubfoot by 18 weeks gestation, but when the diagnosis can be made with ultrasound alone prior to that time point it has been shown that fetal MRI adds no additional information.3 Improvements in ultrasound therefore have increased the detection of prenatal anomalies such as clubfoot, but no modality has yet been shown to accurately predict the postnatal severity of the clubfoot deformity (Liao H et al., 2012). The ability to accurately diagnose a clubfoot deformity in utero is important however for the emotional and economic planning of the mother in relation to the necessary postnatal treatment modalities currently employed for treating CLU clubfoot, thus prenatal ultrasound has remained the modality of choice for prenatal diagnosis (Nemec U et al. 2012).

4. Conclusion

Genetic and environmental factors are important in the cause of ICTEV. There is evidence that development of bone, joint, connective tissue, innervation, vasculature and muscle may each be implicated in the pathophysiology. Disturbance of the overall process of medial rotation of the fetal foot may be the common pathway linked to all these aspects of development. It is likely there is more than one different cause, and at least in some cases the phenotype may occur as a result of a threshold effect of different factors acting together. The hand is never affected in ICTEV, and thus explanation of its pathology is likely to lead to identification of genes whose effects are exclusive to the foot and lower limb. Advances in genetic mapping techniques, development of mouse models, improved understanding of the control of developmental processes and genetic epidemiology studies are all likely to help to elucidate the causes of idiopathic congenital Talipes equinovarus.

Pregnant women underwent a routine ultrasound examination at 19 weeks of gestation. Ultrasound of the fetus showed lower midline lesion suspect of meningomyelocele. More ultrasounds were done which showed spina bifida with congenital Talipes equinovarus of right lower limb. Because of expected problems to the child pregnancy was terminated at 19 plus weeks I/V/O. On physical examination of aborted fetus a 2 X 2 X 1 cm midline mass was seen in lumbosacral region covered with meninges and containing no neural elements remarkably at the level of the lesion, spinal cord was slightly tethered dorsally. CETV of right foot confirmed by means of routine X ray of the aborted fetus. This case study also illustrates a great point regarding the prenatal diagnosis of clubfoot with ultrasound. Transvaginal ultrasound has been shown to detect clubfoot as early as 12-13 weeks gestation, and transabdominal ultrasound has been shown to detect clubfoot as early as 16 weeks gestation. This woman received an ultrasound in her first trimester. However, it is reported that she received subsequent ultrasound imaging and that no deformities were found, illustrating well the need to develop more sensitive prenatal diagnostic tools for clubfoot. The Apgar scores for the children were within normal ranges indicating healthy infants at birth. This score is a standardized rating system, similar in function to the Pirani score as a standardized rating system, for fetal health immediately following birth. A healthy infant should score between a seven and a ten, and the score should not regress between the one-minute measurement and the five-minute measurement.[13]

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