B D R News

The official newsletter of The Birth Defect Registry of India,
(A unit of Fetal Care Research Foundation)

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Proceedings of the birth defects registry meeting held on 4.11.2006

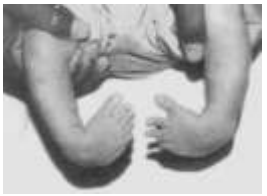
The last meeting of the current year was held on the 4th of November 06 at the premises of Mediscan Systems Annexe, Chennai. Dr. S. Suresh welcomed the gathering. He once again thanked all the members of BDRI family for their sustained support which would soon be completing 5 successful years. He said that as we are progressing in data collection, we also need to move on to the second phase of the study, Project BDR. The Department of Biotechnology, Government of India has accorded permission & grants to conduct research on Folic Acid & Vitamin B_{12} deficiency in Indian Women and its implications in Neural Tube Defects.

Dr Koumudi Godbole of Pune BDR, the principal investigator has taken the initiative to implement this project as a multicentric study in 4 cities Pune, Ahemedabad, Hyderabad & Chennai. FCRF being the Co- investigator would involve the Chennai BDR members in the project. Since Vitamin B_{12} along with folic acid seems to play a vital role in causing birth defects, both have been considered. The study involves collection of blood samples from fetuses and parents, value estimation & comparative study with the Western statistics. While talking about the recent developments in Chennai, he said that FCRF has collected over 600 blood samples from the Newborns of the Government Maternity Hospital (MH), Chennai and screened them free of cost for Congenital Hypothyroidism since August 06. Its sustenance depends on the philanthropic contributions he added. FCRF has also set the process of birth defects data collection in MH which would be a valuable contribution to Chennai BDR. Dr. Suresh repeatedly appealed to member hospitals to enhance the participation of their paediatricians to register more data on birth defects detected during infancy. This was followed by a CME on Club Foot. Dr. K.V. Sridevi spoke on the "Antenatal Diagnosis of Club Foot" & Dr. Vijay Sriram, talked on the "Postnatal Management of Club Foot". The excerpts of the presentations are given below.

Antenatal Diagnosis of Clubfoot -Dr.K.V.Sridevi,
Fellow, Fetal medicine, Mediscan Systems, Chennai.

Dr. Sridevi started her lecture by giving the definition of the term Club Foot. She said that it is an abnormal relationship of Tarsal bones and Calcaneum. Though the definition sounds simple, there is a lot of controversy regarding the definition in the field of orthopaedics she added. This may be due to the fact that the defect not only involves the bones but also the muscles, ligaments, connective tissue and blood circulation. There are several entities of this birth defect such as Congenital Talipes Equino Varus (CTEV), Talipes Calcaneo Valgus & Metatarsal Varus. As the term clubfoot is commonly referred to CTEV, only it was taken up for discussion. This is the commonest congenital correctable orthopaedic deformity of the foot.

It is not a lethal anomaly and the baby can lead a normal life once the defect is corrected. Its presence during antenatal period is quite concerning as it is a potential genetic marker and part of various syndromes. It is associated with 83% of other anomalies in pregnancy & 10-14% of the anomalies during neonatal period. The wide variation is due to its known association with chromosomal defects in the fetus. Most of the chromosomally abnormal fetusus die in utero or during perinatal period. Therefore it is a challenging problem for the Sonologist to identify the potentially correctable club foot and to rule out chromosomal associations thus predicting the prognosis of the fetus to the best extent possible.



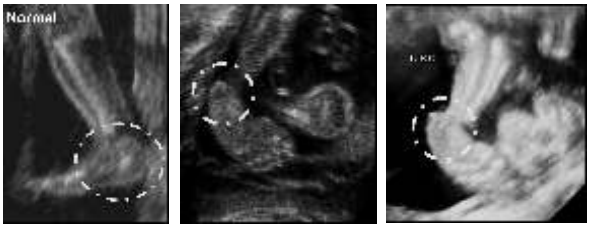
Club foot presents with the following features

Plantar flexion of the ankle
Inversion of the foot
Adduction of the forefoot

The incidence in general population is 1/1000 and the sex ratio is 2.5/1 (Male / female) & 55% of the defects are bilateral. **Etiology** may genetic or nongenetic. It may be caused secondary to Neuromuscular disorders such as Meningomyelocele, Arthrogryphosis, Cerebral palsy & Poliomyelitis. Other extrinsic causes are Oligohydramnios, Uterine anomalies & Early amniocentesis.

CTEV may also be **classified** a into three types such as
1)Positional - mild, 2)Idiopathic - classical &
3)Teratologic - severe.

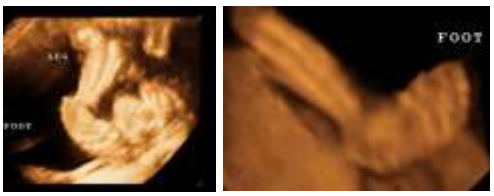
Mild and the classical varieties are the ones which are seen after birth. But during prenatal period all three varieties can be diagnosed. Mild CTEV is seen later in gestation. Usually it does not need any surgical correction. Physiotherapy and casts will solve the problem. Idiopathic defect usually requires surgical correction. Severe form in general is associated with other abnormalities which are lethal. The **pathogenesis** behind is the failure of spontaneous eversion and dorsiflexion of the feet by 10-13wks. Ultrasonogram (USG) is the best modality to diagnose the problem in pregnancy. It can be detected as early as 13 weeks of



Dr. Sridevi while explaining the ultrasound imaging of club foot said that first all three segments- proximal, mid & distal of the fetal extremities should be viewed. She stressed the

importance of assessing the length, echogenicity & shape of these segments to rule out the problem. Normally the lower end of the long bones is perpendicular to the feet. When there is Talipes deformity, the lower end of the long bones and foot are seen in the same plane.

3D USG



Rapid acquisition techniques now allow most extremities to be assessed with 3D ultrasonography. Once the volume is obtained, the structure can be studied carefully without motion. Extremities can be rotated to a standard orientation (sagittal, axial & coronal) and evaluated millimeter by millimeter to clearly see the relationship between the long bones & the ankle. It is often difficult to determine whether abnormal positioning is fixed or transient. Several false positive cases of club foot have occurred. 3D Ultrasound has facilitated evaluation of club foot in some but not all cases. Rotation of the leg into a standard orientation permits meticulous review of relationship between the lower leg, the ankle & the foot. In a normal foot, when the lower leg is optimally rotated, scrolling down a vertical long bone ends with an image of sole of the foot, the so called hang ten position; where as with a club foot, the sole of the foot can not be obtained with the lower leg vertical.

Once the defect is diagnosed, length of the limb, muscle mass & movements of the limb have to be assessed. The length of the foot is decreased & the muscle mass becomes hypoplastic in this condition & they have to be evaluated in terms of gestational age. Along with this, a thorough search for other systemic abnormalities should also be done. When the defect is diagnosed in early gestation, it suggests that the defect is severe. If the defect is diagnosed late in gestation, with the previous scan finding normal, probably it is a milder deformity. Since USG has 5% false positive rate, one has to ensure the presence of the deformity, by observing the fetus while it moves. When the fetus is crowding the uterus in late gestation, close positioning of the legs and feet can be misleading. Hence it is advisable to tap the fetus and view the limbs to find out if there is a defect. The speaker cautioned the audience saying that third trimester is not the right time for diagnosing limb abnormalities & second trimester is only the ideal time to find out this defect.

Dr. Sridevi later presented various case scenarios associated with clubfoot such as Neural Tube Defect, Arthrogryposis, Trisomy 18 & Meckel Gruber syndrome.

Genetic counseling facilitates appropriate decision making during prenatal & postnatal period. It provides a better understanding of the etiology, recurrence risk & prognosis of the fetus.

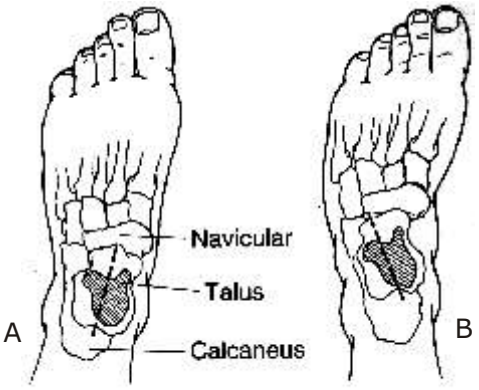
Dr. Sridevi concluded the session by presenting the statistics on club foot derived from Mediscan Systems. 397 cases of club foot were diagnosed by ultrasound between the years 2001- 2005 at Mediscan systems. Of the total number, 243 had associated anomalies & the rest of 153 had isolated club foot. Majority of them (160 nos) were diagnosed between 20 - 25 weeks of gestation when target scanning was done. Karyotyping could be done only for 17 cases out of which only 2 were found to be abnormal.

The take home messages from the speaker were as follow

- The time of diagnosis of club foot denotes the severity of the problem.
- If diagnosed earlier in gestation, it denotes a strong genetic basis and a more severe form. If diagnosed later in gestation (i.e.35 weeks) it could be a late evolving, mild form of postural deformity caused by environmental factors. Eg.oligohydramnios. The Sonologist in this case should not be blamed for not diagnosing it earlier.
- Karyotyping is mandatory for both isolated & associated clubfoot to rule out chromosomal etiology.
- It has 2-3% recurrence risk if the previous child is affected and 2% in case of the father being affected & 5% if the mother is affected, in each pregnancy.

Postnatal Management of Club Foot - Dr. Vijay Sriram, Consultant Paediatric Orthopaedic Surgeon, Chennai

Dr. Vijay Sriram at the commencement of his lecture, said that he would discuss about the Idiopathic Club Foot & not go into details of Syndromic Club Foot. Though many theories have been formulated on the etiology of this birth defect, it is still not precise & definitive. One such theory talks about the role played by genetics in causing the defect. Statistically 25% of the affected have a family history of CTEV. Siblings of the affected have thirty fold increased risk of having the defect. Both siblings in 32.5% in monozygotic twins and only 2.9% in dizygotic twins are likely to be affected. Histologically it is caused by a deformity in the bones or muscles of the foot. Germplasm defect as early as 6 weeks of gestation can cause a deformity in Talus & Calcaneum. Sometimes involvement of the leg muscles due to fibrosis, ischemia due to the absence of tibial artery or tightening can cause club foot. Hippocrates was the first to cite reduced liquor volume as the reason which is not true as feet are formed as early as 6 - 8 weeks of gestation and there is enough space in the uterus for the foot to mould into shape. Early amniocentesis increases the risk to 1.1%. In normal births; the rate is 0.1%.The rate increases to 15% when there is a leak.



A - Superior view of the normal relationship between navicular, talus and calcaneus
B - CTEV - Talus and calcaneum rotate laterally and navicular is subluxated on talar head. The dashed line represents the axis of the talus

In CTEV, the head & neck of talus is deviated medially and downwards; declination angle is reduced; Trochlear surface becomes broad & Talar head descends forwards, inwards & downwards.

The Calcis gets inverted under the talus, rotates in all three planes Anterior half rotates medially & downwards. Posterior half rotates laterally & upwards. One of the **associated changes** in CTEV is atrophy of the calf muscle. When the defect is severe, the atrophy of calf muscle is worse. It may appear thin as in polio affected leg but its strength is fairly good. Other changes include short Tendo Achilles, Flexor Hallucis Longus, & Flexor Digitorum Longus. There is external rotation of the hip and Genu valgum & Genu recurvatum are also present as compensatory features in the affected. When the child walks with foot turned inwards, the knees turn outwards and backwards to maintain balance.

While examining the child with CTEV, both the components of the foot are examined. Hind foot is seen for Equinus & Varus deformity. The heel pad will be empty as the calcaneum is displaced and the foot is nicely turned inwards. In the fore foot there is adduction, supination & cavus. One should not forget hip examination as quite a few of them have dislocation of hips. The spine should also be examined as club foot is closely associated with spinal problems. The neurological status of the child has to be evaluated to rule out syndromic associations like arthrogryposis by observing the movements of the foot & toes.

Dr. Vijay Sriram while talking about the grading of the club foot said that the most commonly used system is Dimeglio Grading as shown below:

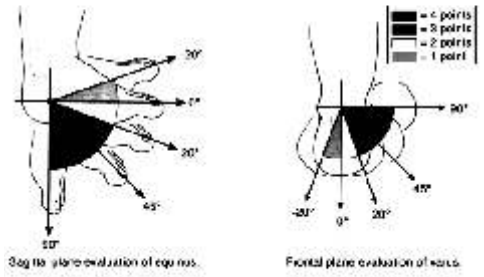
Classification of clubfoot according to Dimeglio

Classification

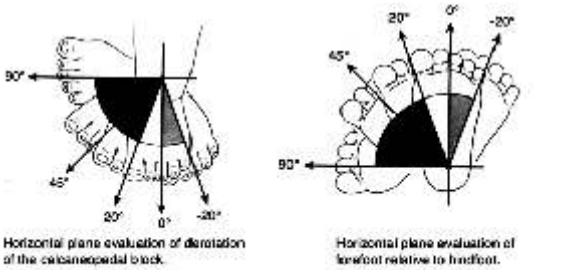
Classification Grade	Type	Frequency (%)	Score
I	Benign	20	(<5)
II	Moderate	33	(=5<10)
III	Severe	35	(=10<15)
IV	Very severe	12	(=15<20)

Assessment of Clubfoot by Severity Scale

Characteristics: Reproducibility	Points	Characteristics: Other parameters	Points
90°-45°	4	Posterior crease	1
45°-20°	3	Medial crease	1
20°-0°	2	Cavus	1
<0° to -20°	1	Poor muscle condition	1



Sagittal plane evaluation of equinus. Frontal plane evaluation of varus.



Horizontal plane evaluation of derotation of the calcaneopedal block. Horizontal plane evaluation of forefoot relative to hindfoot.

(Classificiation of clubfoot according to Dimeglio. Each major component of the clubfoot (eg. Equinus, heel varus, medial rotation of the calcaneopedal block, forefoot adduction is graded clinically from I (benign) to IV (very severe). Additional points are added for deep posterior and medial creases, cavus and poor muscle condition. The total score is categorized into one of four groups; grade I (benign), grade II (moderate), grade III (severe), grade IV (very severe). (Adapted with permission from Dimeglio.A, Bensahel.H, Souchet.P, Mazeacu.P, Bonnet.F; Classification of clubfoot, J.Ped.Ortho B;1995;4;129)

Treatment is given after the defect is graded. The aim of the treatment is to obtain a plantigrade mobile foot and not a cosmetically appearing good but stiff foot the surgeon said. He informed that previously surgeons were concentrating more on the cosmetic aspect leaving the foot with poor muscle conditions. Presently their approach is to give the child the best functional foot possible to lead a quality life. There are many treatment methods such as 1) Kite Method 2) Ponseti Method 3) Posteromedial Release & 4) Exclusive Reconstructive Surgery.

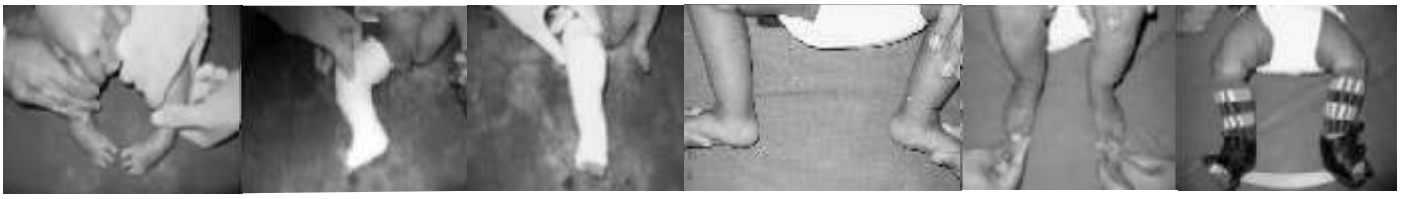
Kite method is one of the oldest method & common form of manipulative treatment. Serial casting is started very early and the foot is gradually abducted. The calcaneum is everted by manual pressure. This procedure involves many months of casting and is tedious on the part of the parents. After the deformity is corrected, Equinus correction is done with or without a minimal surgery.

The next method which is widely practised now is called the **Ponseti** method. This was developed by Dr. Ponseti of Iowa,U.S.A as early as 1940. Though it was not popular earlier, now it is used by the majority. It involves manipulative correction with serial casting from the II week of life. Cast is changed on weekly basis. Cavus is first corrected by supinating the foot which is a different approach from Kite's method. The entire foot is then abducted to correct the rest of the deformities. There is no manual pressure applied over the calcaneum. Casting is required for 5 - 6 weeks.

Once the foot abducts to 30^0, Percutaneous TA tenotomy is done to achieve 15^0 of ankle dorsiflexion. Then again casting is done and retained for a month. Later foot abduction brace is given for 2 - 5.5 years depending on the child's cooperation. Physiotherapy is also taught to the mothers and the child is made to do the exercises on daily basis.

Percutaneous, TA tenotomy

Dr. Vijay Sriram explained **Ponseti method** with beautiful illustrations of bone model.



Sometimes one has to resort to Posteromedial Release procedure as all club foot are not similar. It involves releasing only the tight structures of the foot. It is usually done around 6 months of age when conservative therapy fails. By this time the foot should have grown to a decent size of at least 8 cm. The speaker informed that Dr. Ponseti does this as an outpatient procedure, here it is done as theatre procedure.He listed out the structures released in posterior, medial & lateral parts of the foot. (Refer table below)

Posteromedial Release Tight Structures		
Medially	**Posterior**	**Lateral**
Abductor Hallucis Plantar fascia Tibialis posterior tendon Talo-navicular capsule	Tendo Achilles Posterior capsule of the ankle joint Sometimes the subtalar joint	Calcaneocuboid Joint Talofibular ligament Calcaneofibular ligament

During **discussion,** Dr. Suresh expressed his views on club foot. He said that imaging the muscle mass with USG is quite difficult and even 3D imaging has not helped much in this aspect. Fetal MRI may provide more useful information on the muscle mass he added.

1. Will the muscle mass measurement going to change the course of treatment if the parents opt to continue the pregnancy?

Not really; as there are no standard controls for muscle mass assessment for different gestational periods. There is also no role at present for fetal MRI in isolated club foot.

2. When CTEV is detected in pregnancy, should the parents be asked to consult the Orthopaedic surgeon? How does this help the parents?

Yes, the parents should consult the Orthopaedician in pregnancy. The surgeon concerned would facilitate finer details of the corrective procedure involved after birth, the period of hospitalization, cost involved and the outcome all of which would be reassuring the parents to continue the pregnancy. The specialist also has the knowledge of recent techniques engaged in corrective procedures.

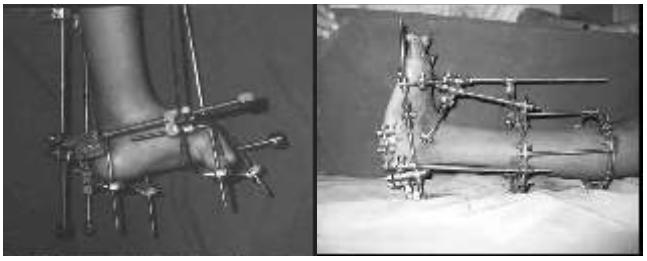
3. Can the handicap be corrected for optimal function?

It is possible to correct any abnormality of the foot to function close to normal.

4. Can the affected after correction become a Dancer or an Athlete?

Dr. Vijay Sriram while replying to this query said that one of Dr. Ponseti's patients who was corrected for CTEV, went on to become a Olympic Champion.

Recontructive procedures are needed for those who have failed to undergo corrective procedure earlier and for those in whom the problem has relapsed. Osteostomies & External fixators are used to get the correction done mostly with good outcome rates. Though the results are good, they have high morbidity rates. The speaker while concluding the lecture said that parents & pediatricians should never delay corrective procedures and let the foot assume postures that are difficult to correct. He also assured that CTEV is a totally correctable anomaly and the affected can lead a normal life after correction.



BDRI extends a hearty welcome to the nodal and participating members of AKOLA BDR, Maharashtra

BDRI

&

Rotary Madras Metro

Dt 3230

Partnership Program for Birth Defects Prevention

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