B D R News

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

Volume 7 Issue 2: July 2007

Proceedings of the BDR meeting held on 25th August 2007

The third BDR meeting of the current year was held on 25/08/07 at Mediscan Systems Premises, Chennai. Dr. Indrani Suresh (Associate Director - BDRI) welcomed the gathering. She was happy to announce that, the FOGSI (Federation of Obstetric & Gynaecological Society of India) in their quest to understanding the problems of Indian women, has planned to initiate a birth defects registry by next year. The FOGSI BDR would function under the auspices of BDRI, Chennai. FOGSI, under the presidentship of Dr.Narendra Malhotra (Ahmedabad) has nominated Dr. S. Suresh, Dr.P.K.Shah and Dr.Ashok Khurana as the national coordinators of the registry. To start with, this will enroll 100 members comprising all FOGSI regions in the country. The Central Registry is working on the functioning modalities of this project. She also said that the II phase of the BDRI mission of finding out the etiology of common birth defects in our country have already been commissioned as planned. She hoped that the members would have browsed through the Government sponsored NTD study project in the last newsletter.

Moving onto the scientific session of the day, Dr. Indrani said that time and again medical practitioners come across neural tube defects. The common defects like anencephaly, encephalocele etc are picked up early in pregnancy. Quite frequently long segment spina bifida is seen late in pregnancy. When there are no cerebellar signs & the defect is isolated, it mostly ends up as closed spinal defect having better prognosis. She hoped that the CME on Antenatal & Post natal Management of Spinal Defects would throw light on the appropriate methods of dealing with the affected fetuses & the families. The excerpts of the presentations are given below:

Antenatal diagnosis of closed spinal defects

Mrs.Vijayalakshmi Raja, Senior Sonographer, MediScan, Chennai

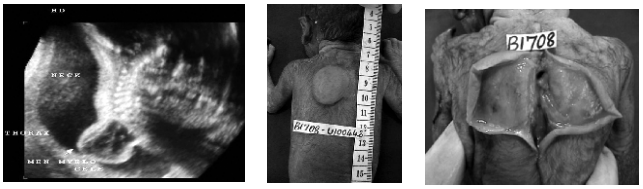
The speakers, Mrs Vijayalakshmi & Dr. Indrani Suresh presented the role of high resolution, high frequency ultrasound in fetuses & newborns with spinal defects.

Ms. Vijayalakshmi remarked that antenatally missed out external lesions like cleft lip and palate, hand and foot anomalies and closed spinal defects produce great anxiety in the family. Their immediate reaction would be "how this was not seen on ultrasound"? The speaker discussed about a few challenging antenatal case scenarios with closed spinal defects.

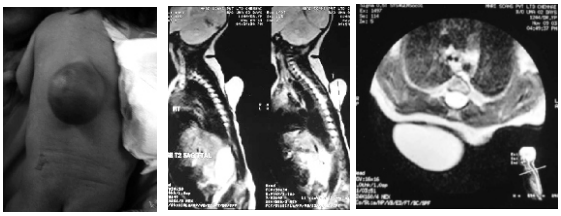
According to Pilu et al (Prenatal Diagnosis of Open and Closed Spina bifida, ISUOG) spinal defects can be **classified** as follows:

- **Open Spina bifida -** Meninges exposed to the exterior with "Head signs" (Lemon, Banana signs) & elevated Alpha feto protein (AFP)
- **Closed Spina bifida -** Skin covered dysraphism with normal posterior fossa &AFP within normal range.

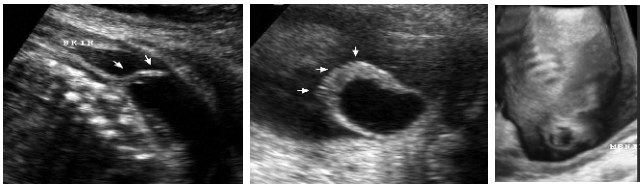
3D ultrasound provides more insight about the thickness of the covering, contents of the sac, the extent of the spinal defects, spinal cord abnormalities and associated vertebral anomalies. In all the cases discussed, the posterior cranial fossa was normal. In the case of a **thoracic meningomyelocele,** the patient opted for termination of pregnancy. It was a skin covered defect, however, on autopsy, tethering of cord was diagnosed. Retrospectively, 3D imaging revealed the presence of tethered cord.



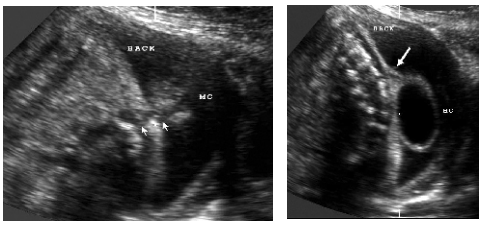
Larger the lesion, higher is the incidence of tethering & split cord abnormalities. Another case of **cystic meningocele** in the thoracic region undiagnosed during targeted scan was delivered and operated. MRI in the newborn revealed no cord structures.



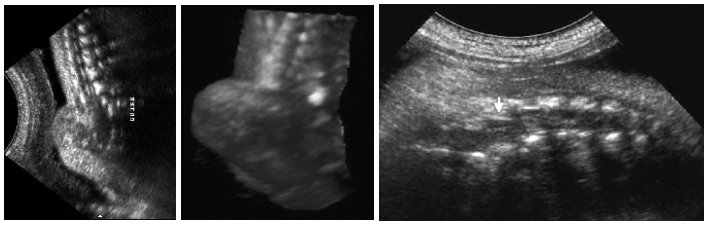
High resolution imaging can help in delineating the membrane and the skin covering separately. In the following case, imaging at 24 weeks of gestation, revealed streaks of hair over the swelling.



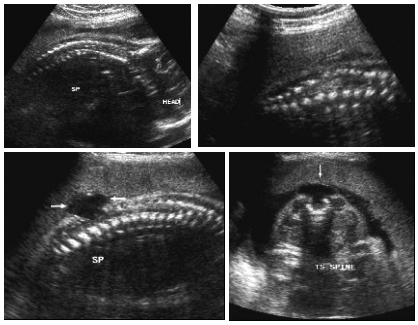
This suggested the presence of a skin-covered lesion. Near the upper margin of the swelling, wedge shaped elevation was seen secondary to elevation of the skin over the lesion.



Postnatally meningocele was confirmed without the tuft of hair & this child remains unoperated at 4 months.Compared to the cystic lesions, echogenic lesions are relatively diagnosed with ease. Associated tethered cord and split cord malformations are common with **lipomeningocele.**



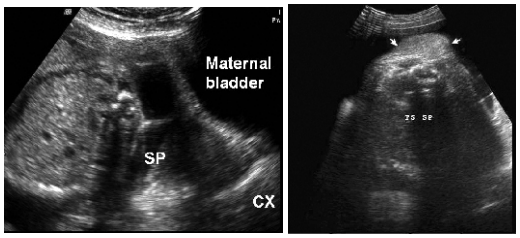
Rarely **mild variety of Arnold chiari malformation** (herniation of cerebellar tonsil) can be mistaken for closed spinal defects. In chiari malformation generally the membrane is exposed. Significant widening of the posterior ossification centre helps in differentiating from closed defects.



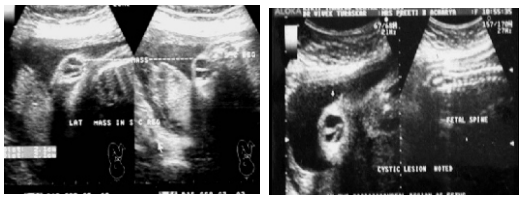
In the above case herniation was not detected by USG. If the patient had opted to continue the pregnancy, as it was a cystic meningocele, will surgical resection of the meningocele result in obstructive hydrocephalus? Such situation necessitates the importance of postnatal MRI prior to surgical resection.

False negative diagnosis of Spina bifida can occur in spinal defects without head signs. In such cases the lesions are:

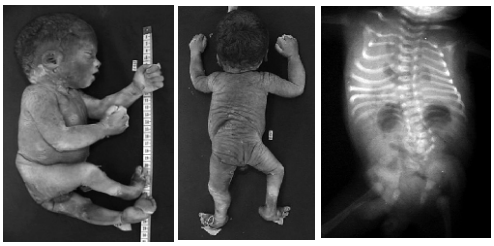
small in size / in the sacral region / predominantly cystic / close to the uterine wall / the fetus may be in breech position / the widening of the posterior ossification centre is not clearly made out in transverse and coronal sections.



The speaker mentioned that assessing the posterior cranial fossa beyond 30 weeks due to shadowing from the occipital bones; differentiating between epithelial membrane vs. skin, are some of the **limitations of ultrasound imaging.** In another case, there was a cystic mass with septations which was misinterpreted as **spina bifida with meningomyelocele.**



Postnatal autopsy picture showed no swelling in the back & .X ray revealed sacral dysgenesis.



Data collected over a period of 10 years (1997 - 2007) from Mediscan Systems, revealed a total number of 39 cases of closed spinal dysraphism. Out of these, follow up could be obtained in 13 cases. In 8 cases autopsies were done after termination. Of the 3 cases operated, in one child surgery was performed within 24 hours and at one year is doing well, one succumbed after 15th postoperative day due to meningitis & the other in whom surgery was done at 11mnths of age after the onset of neurological deficit, survives with severe disability. Early surgery proves to have favourable outcome. In 2 other cases no correction has yet been performed & the children are at 3 years & 4 months of age at present.

The speaker ended the lecture saying that once a closed spinal defect is diagnosed, Pediatric Neurosurgeon has to be consulted to predict the outcome after correction. In patients diagnosed late, mode of delivery, postnatal protection of the lesion are some of the important factors which have to be taken care of. When the diagnosis is made during targeted scan, MSAFP should be estimated to differentiate between closed / open defects.

SPINAL SONOGRAPHY

Dr. Indrani Suresh, Director, Mediscan Systems, Chennai

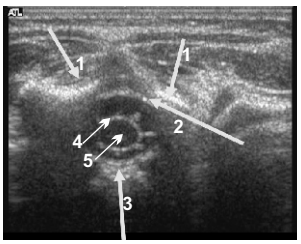
Dr. Indrani Suresh commenced her lecture saying that high resolution transducers provide useful information in the newborns in pediatric units. However, MRI is the gold standard in closed spinal defects. She discussed, whether it is essential to subject all the newborns with occult spinal defects to an MRI before surgery or information provided by a Sonologist to the Surgeon would suffice. In a series of 4 cases referred from the Institute of Child health, Chennai, 2D, extended field of imaging and 3D ultrasound of the spine were performed. As the patients could not afford MRI, surgeries were performed based on the ultrasound information. The surgical correlation was encouraging, she said. In newborn, optimal imaging of the spine is obtained, as the spinous processes are not completely ossified.

Several indications for a spinal ultrasound are: pigmentation, nevus, tuft of hair on the mid line, deformities like tethered cord, spinal dysraphism & hydrocephalus .

By about 22 weeks of gestation, the spinal cord ends at L2 level as seen in the adults. The deep anterior fissure is seen as a dot in the transverse section of the spine, which should not be mistaken for a spinal canal.

Spinal imaging may be done in various positions: Prone, Lateral decubitus, Sitting and Erect

Linear 10 Mhz transducer helps in better anatomy delineation. In the transverse section the following anatomical landmarks are visualized.

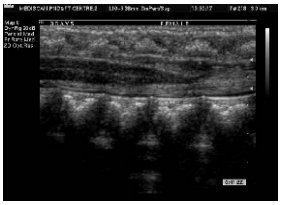


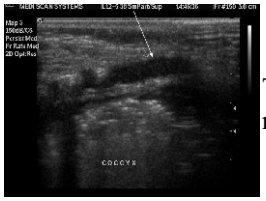
1. Dorsal elements
2. Dura seen close to bone
3. Vertebral body ventrally Subarachnoid fluid seen
4. Echogenic border of hypoechoic cord
5. Echogenic central complex

In the para sagital section, three echogenic lines are seen, the outer lines are the borders of the spinal cord and the central echogenic line is the deep anterior fissure.

The spinal cord is narrowest at the thoracic level and is wider at the cervical and lumbosacral levels. In the sagital section, the tapering conus medullaris and the cauda equina fibres are seen.

The cauda equina fibres are cluster of nerve roots have "spider" like configuration & the nerve roots oscillate well on real time. The oscillation of the nerve roots is reassuring which rules out tethered cord



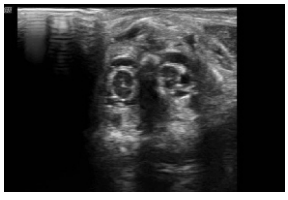


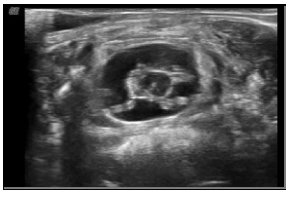
The coccyx is cartilaginous which should not be mistaken for a mass.

Case studies

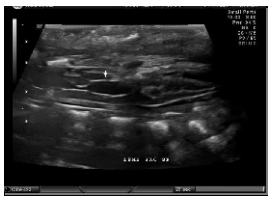
In all the following 4 cases which were discussed, antenatal ultrasound was reported as normal. Imaging of the neonatal spine was preceded by a cranial ultrasound. Lateral ventricles and the posterior fossa were normal.

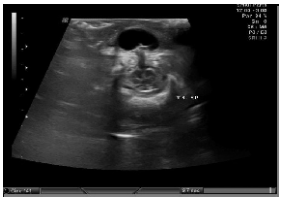
Case I - A full term normally delivered, 14 days male child weighing 2.8 kilograms was born with a swelling in the sacral spine. Ultrasound imaging was performed in long and transverse section of the spine. Cervical & thoracic spines were normal. There was splitting of the cord by a bony spur with reunion at the conus medullaris level. The diagnosis assigned was Diastematomyelia.



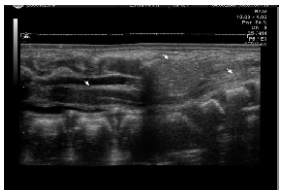


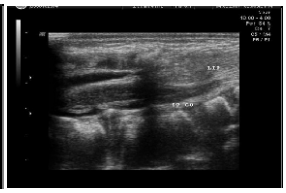
Case II - A 13 days old male child was referred for imaging. There was meningomyelocele with hydromelia



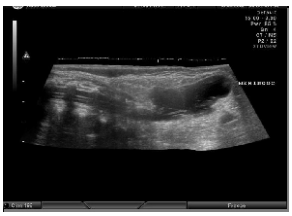


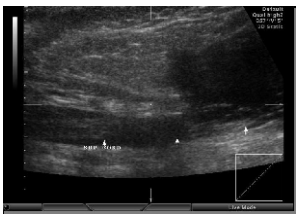
Case III-A mother with bad obstetric history & previous child with NTD was scanned during her IV pregnancy.The fetus, postnatally showed lipomeningocele in sacral region with tethered cord.





Case IV-In the last case, a cystic meningocele with no nerve roots was seen.





While concluding her presentation, Dr. Indrani quoted an abstract from Clinical Paediatrics (December 1995) on certain facts about occult spinal dysraphism. They were as follows

- The progressive neurologic dysfunction caused by occult spinal dysraphism can be prevented with early clinical recognition, radiographic diagnosis, and neurosurgical treatment.
  However, detection of occult spinal dysraphism in the infant is difficult because neurologic symptoms often are not apparent until the child becomes ambulatory.
- Occult spinal dysraphism, however, can be suspected in the asymptomatic neonate when cutaneous stigmata, such as hemangiomas, hairy patches, deep and/or eccentric dimples, or subcutaneous masses are seen over the lumbosacral spine
- Because of the serious, often irreversible, sequelae of a delayed diagnosis, spinal sonography of high-risk infants with midline, lumbosacral, cutaneous stigmata should be considered as an effective, noninvasive screening method.

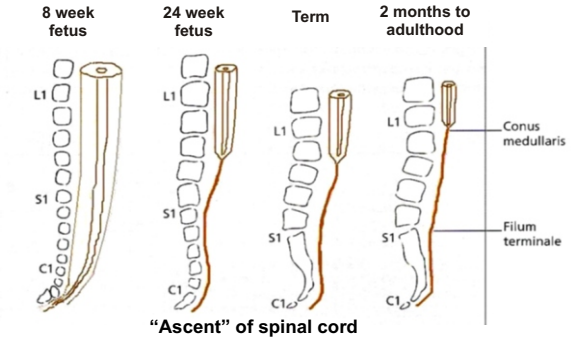
She also disclosed the facts published in **European Journal of Radiology (April 2006)** about the use of Ultrasound imaging in cases with simple cutaneous markers & in newborns with potentially dangerous dimples associated with dermal sinus. When there is a suspicious result on sonography, MRI is indispensable. Great advantages of sonography are the real time examination and the potential to show oscillations of the conus, filum and cauda equina.

LUMPS, BUMPS & HOLES - a primer on Spinal Dysraphism

Dr. C.Mohan, Paediatric Surgeon, Institute of Child Health, Chennai

Dr. Mohan started the lecture in a lighter vein by giving explanation to the title he chose for his presentation. He mentioned that since most of the occult spinal defects present as lumps, bumps / holes, he had named it so. Spinal dysraphism is a spectrum of birth defects of the developing spinal cord which may occur either as closed or open defect. He discussed briefly the embryology of the neural axis.

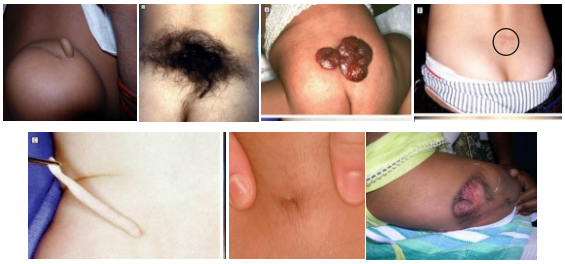
Recapitulating the formation of the neural tube from the neural plate & the closure of the anterior & posterior pores, he said that during development, the surface ectoderm attached to the neural tube starts separating from the neural tube (process of disjunction). When there is failure of fusion of the neural folds, the process of disjunction is affected resulting in spinal dysraphism or occult spinal defect.



"Ascent" of spinal cord

Conus medullaris is the most distal bulbous part of the spinal cord which tapers down as the Filum. The surgeon explained that the conus extends up to the coccyx during 8 weeks of gestation. It stays at L3, L4 levels at 24 weeks, L2 level at term & at L1 level from 2 months of age through adulthood.

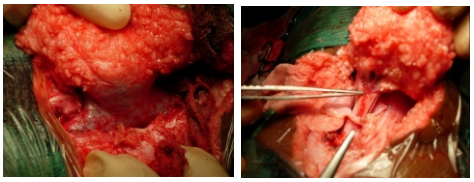
Tethered Cord Syndrome (TCS): is progressive neurological deterioration resulting from traction on the conus and the spinal nerve roots because of local adhesions which may be due to various causes. It is important to find out the position of the conus medullaris in relation to the spine while releasing the tethered cord. TCS is diagnosed when Conus medullaris if found below the L2 level. As already discussed, there are many cutaneous markers that are suggestive of occult spinal defects such as subcutaneous mass or lipoma, hairy patch on the back, dermal sinus, vascular lesion like haemangioma, skin appendages like skin tag or tail & scar like lesions.



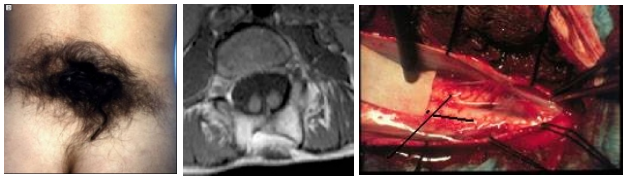
If more than two markers are present there are higher chances of the defect being present & vice versa. Hypertrichosis has a high predictive value for underlying split cord malformation, he added any midline sinus found on the back above the gluteal cleft has communication to the duramater until proved otherwise and is also strongly suggestive of occult spinal defect (OSD). Coccygeal sinus found deep inside the gluteal cleft is innocuous.

Dr. Mohan explained various presentations of OSD with many illustrations. While talking about the postnatal imaging of these defects he said that Ultrasound is the best modality for infants below 6 months. It involves low cost, can be repeated easily & requires no sedation. He said that good imaging for surgical intervention depends on the technical skill of the operator. MRI is ideal for imaging OSD in older children.It facilitates better anatomical details required for surgery. Compared to USG, MRI involves high cost & sedation before imaging, he said.

The surgeon went on to show pre & post operative pictures of children with lipo meningocele, lipo meningomyelocele at different levels (thoracic & sacral) & split cord malformation. He said that tethered cord is released after excising the lipoma



In case of split cord, the bony spur is excised & if there is duplicate dural covers it is made single while correcting the defect.



He concluded saying that surgery should be done before the onset of neurological deficit as the sequential effects of neurological deficits once established are not reversible.

Error regretted !

The Karyotype given for KLINE FELTER syndrome may be read as XXY and not XXX as published in BDR News Jan 2007 Issue

BDRI extends a hearty welcome to the nodal (J.N Medical college and KLE Society's Prabhakar Kore Hospital & MRC) and participating members of Belgaum BDR!

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