B D R News

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

Volume 9 Issues 1 & 2 combined: January & April 2009

Proceedings of the Birth Defects Registry meeting held on 09.02.2009 & 09.05.2009

The first & second quarterly registry meetings were held on 09th February & May of the current year at MediScan, Chennai. A CME on Neural Tube Defects (NTD) with special reference to Closed NTD & its management were discussed in the first meet. During the welcome address, Dr. Indrani Suresh (Director, BDRI) enumerated the developments of BDRI & announced about the flexible enrollment of membership available now. BDRI offers both individual / team memberships to those who opt for it. Registration is just a click away! Those who would like to become members need only to access the website www.mediscansystems.org , look for the consent form of either FOGSI BDR or BDRI, fill in, submit & get enrolled. With the advent of FOGSI members joining BDRI, such changes have been made to motivate more & more members join the mission for a national cause. The existing members could continue their support as in the past. Reporting data online will be facilitated shortly to members who wanted do so. She spoke on the prospects of closed neural tube defects that have better outcome considering the management of NTD in general. She also mentioned that, although 24 nodal centers had been inaugurated by BDRI, only 17 registries are actively contributing data. She expressed her hopes to revive again those registries discontinued for some reasons & gather data once again as the project has to move well forward.
Dr. Pooja Vazirani, Fellow Fetal Medicine, MediScan &
Dr. B. Chidambaram, Consultant Paediatric Neurosurgeon, spoke on the Antenatal Presentation of Closed Spinal Defects & its Postnatal Management respectively.

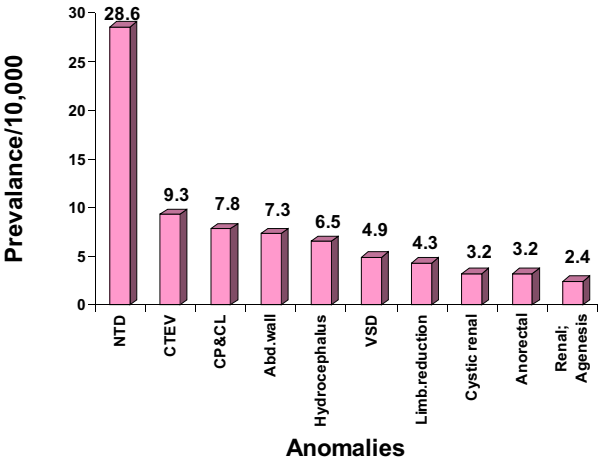
The Annual Birth Defects Statistics of year 2008 was reported by Dr. N. Suganya, Epidemiologist-BDRI,at the second BDR meeting. This was followed by Dr. Ranjit, Consultant Paediatric Cardiologist, Sri Ramachandra Medical College Hospital & RI, speaking on Correctable Congenital Cardiac anomalies.

While welcoming the audience Dr.S.Suresh (Director, BDRI) thanked the existing BDRI members & the newly enrolled FOGSI members for their immense support. The registry would carry on the good work started for a cause with improved coverage of births. He hoped to collect & analyze more volume of data with new enrolment & online data reporting facilities provided to existing/ prospective members.

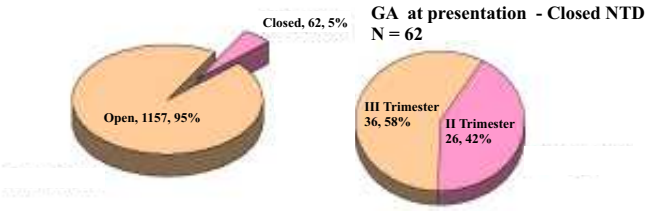
The excerpts of the presentations & the Annual Report of Birth Defects Statistics, Year 2008 are given below:

Mediscan Statistics of Neural Tube Defects
Dr. I.V. Amit Kumar - Scientific Officer, ICMR Project MediScan.

Dr. Amit Kumar presented the statistics of Neural Tube Defects derived from the data collected at Mediscan over a decade (1998-2008). He said that NTD is the commonest congenital anomaly around the globe and it tops the list of the BDRI sample data all through the 7 years of analysis. The incidence of NTD was found to be 28.6 /10000 live births among the crude birth prevalence of major anomalies.

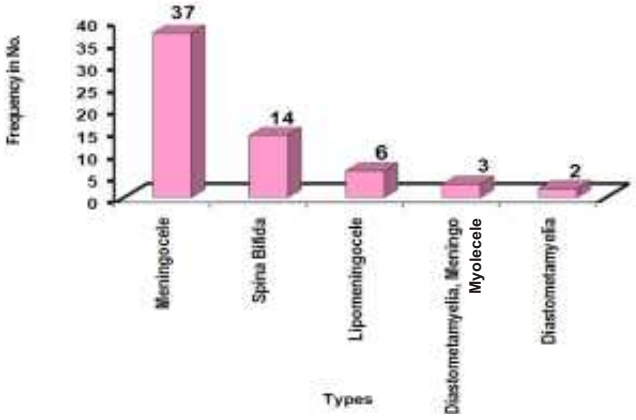


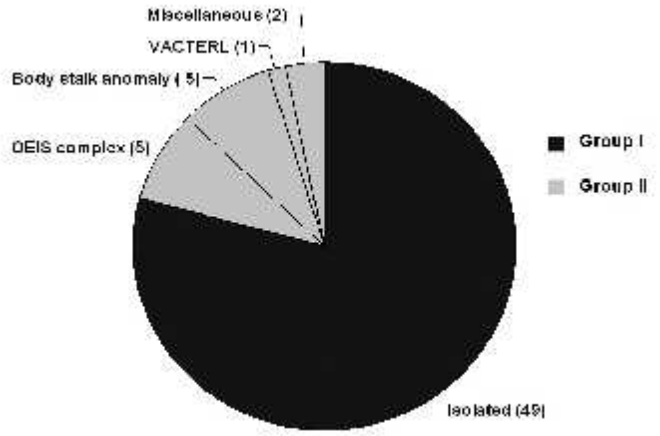
95% (n=1157) of cases referred to Mediscan in the retrospective study, were diagnosed to have open type defects and 5% (n=62) had closed NTD on Ultrasound imaging.



42% of NTD were detected in late second trimester and 58% in the third trimester revealing the fact that a significant group had late diagnosis of NTD even today. The following diagrams gave an insight in to the magnitude and spectrum of the closed types of NTD.

Frequency of Closed NTD in the prenatal series (1998 - 2008), n= 62

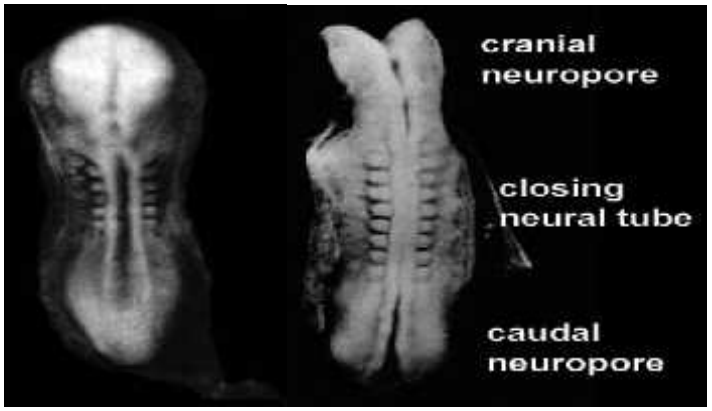




Antenatal Presentation of Neural Tube Defects

Dr. Pooja Vazirani, Fellow, Fetal Medicine, MediScan

Dr. Pooja Vazirani began her talk, briefing the embryology of NTD. The formation of the neural plate and neural tube starts around 22-23 days after fertilization in the region of the fourth to sixth pairs of somites. The caudal one third of the neural plate and tube represents the future spinal cord. The cranial two thirds of the neural plate and tube, as far caudal as the fourth pair of somites, represents the future brain..



Fusion of the neural folds and formation of the neural tube proceeds in cranial and caudal directions. Small areas of the tube remain open at both ends.The cranial opening - rostral neuropore, closes at approximately 24 days &¨the caudal neuropore closes at around 28 days. Primary neurulation closes the proximal neural tube and secondary neurulation results in closure of lower spinal cord. Presentation of NTD at various sites along the neural tube is due to multi site closure involved as explained. The sites of closure happens at

Sites	Region
1	the midcervical region, proceed cranially & caudally
2	the prosencephalon / mesencephalon boundary
3	the stomadeum proceeds rostrally & convenes cranially with closure site 2
4	the rhombencephalon and proceeds rostrally & ends with
5	unidirectional closure of the caudal end beyond S 2 level.

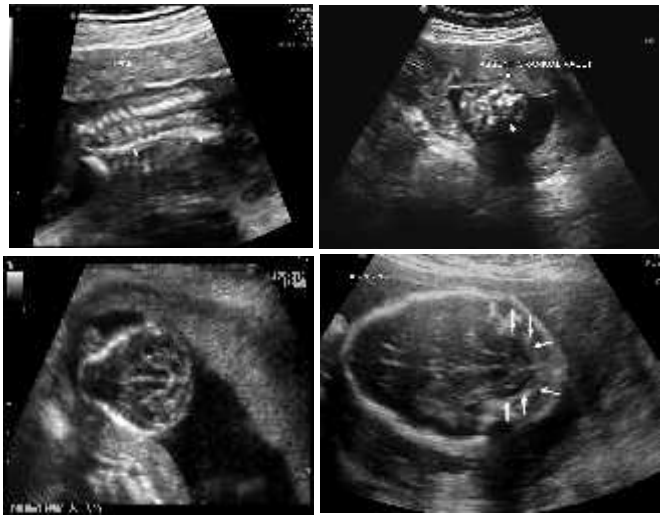
The **type & presentation of NTD** depends on the site of non closure as given below:

1) Spina bifida cystica - site 1
2) Meroencephaly (anencephaly) - site 2
3) Craniorachischisis - sites 2, 4, and 1
4) Nonfusion is rare at site 3
5) NTD that occurs from the second lumbar vertebra to the second sacral vertebra - site 5

Based on clinical significance and embryology, NTD may be classified as:

a. Cranial neural tube defects
b. Open spinal dysraphism
c. Closed spinal dysraphism

Cranial NTD comprises of Anencephaly, Iniencephaly, Rachischisis & Meningomyelocele. **Open NTD** may be picked up antenatally by certain direct presentations like Anencephaly, Rachischisis, Meningocele and Meningomyelocele. Indirect USG markers are lemon and banana signs of the cerebellum. Biochemical marker for NTD is MSAFP (Maternal Serum Alpha Feto Protein) measured between 16 - 20 weeks of gestation.

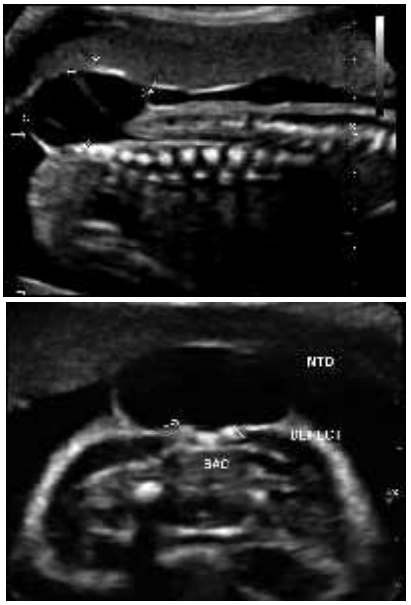


The **pathophysiology** of these lesions is due to the leakage of cerebrospinal fluid (CSF) within the amniotic cavity. The hypotension caused in subarachnoid spaces results in ArnoldChiari II malformation. 1. Lemon sign presents when the soft frontal bone is drawn in due to the negative intracranial pressure. 2. Banana sign appears when negative pressure is created in the small posterior fossa which pulls it down changing the cerebellar shape.

Closed spinal dysraphism is an elusive entity. Its real incidence has not yet been clearly established. It accounts for about 10% of cases.Generally speaking, most cases go undetected in the prenatal period . As for closed spinal dysraphism is concerned, it arises in the conus medullaris , lower lumbar & sacrococcygeal region .This region is continuous with the caudal cell mass. By day 30, vacuoles coalesce and become continuous with the neural tube. Disorders of the caudal neural tube are caused due to defective secondary neurulation, non fusion of neural arches involving the caudal spinal cord & or its coverings. **Occult dysraphism** is commonly **associated** with vertebral anomalies and have a skin covering.

They include 1.Meningocele, 2.Lipomatous malformation, 3.Dermal sinus & 4. Split cord malformation.

Meningoceles are 1) commonly located in the lumbosacral region, 2)covered with skin 3) rarely involves more than 2-3 vertebrae 4) the sac consists of arachnoid and dural meninges with CSF. Most of them do contain some neural element - myelomeningocele.



The neurologic outcome is normal in most of the newborns. Immediate post natal surgical correction and resection of the herniated meninges is necessary to save babies with such defects.

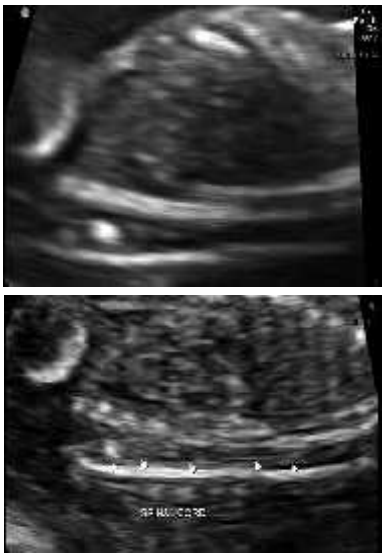
Lipomatous malformation includes all closed NTD's with excessive lipomatous tissue. This tissue can be situated within or attached to the spinal cord or the filum terminale. Its origin is controversial. It may occur as dorsal, transitional or terminal lesion - lipoma of the filum terminale. The common lumbosacral lipomeningocele is a transitional lipoma. Extended imaging helps diagnose filum terminale lesions. Most newborns have good prognosis & normal lower limb and urologic function.

Dermal sinus presents in deep squamous epithelium lined tract on or near the midline of the back. It is common in the lumbosacral region & contains hair or communicates with the duramater. They result from abnormal adhesions between the ectoderm which forms the neural tube and that which forms the skin. Surgical excision of these lesions is warranted during postnatal period, as there is risk of infection & compression of the underlying neural elements by progressive enlargement of the dermal sinus.

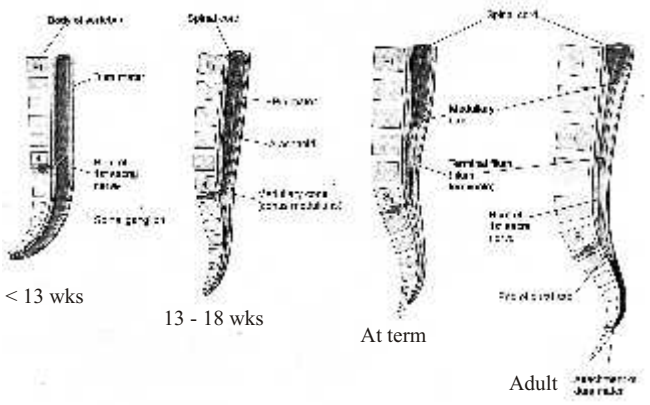
Split cord malformation is another entity which is a rare congenital anomaly caused due to "splitting" of the spinal cord in a longitudinal (sagittal) direction. It may have an osseous (bone), cartilaginous or fibrous septum in the central portion of the spinal canal.

It produces a complete or incomplete sagittal division of the spinal cord into two hemicords. Prenatal diagnosis of closed defects includes imaging of the spine from 9th thoracic and 1st sacral levels as upper lumbar vertebra is the commonest site of these defects. An extra posterior echogenic focus between the fetal spinal laminae, splaying of the posterior element with skin intact & lack of head signs are suggestive of closed defects.

Prenatal diagnosis of closed NTD

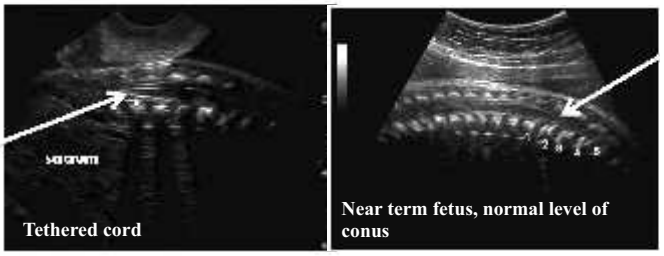


Knowledge of the level of spinal cord and its normal apparent ascent is essential in pathological conditions of the spine. There is a difference in the growth of the embryo and its spinal canal & that of the spinal cord. Due to this, there is as apparent ascent of the spinal cord during growth & development.



However the filum terminale maintains its original position & it is the conus medullaris that shows the apparent ascent. Abnormal positioning of conus medullaris (below S1 level) of the cord results in undue pulling & tethering causing damage to the nerve tissue. This has its own impact on the prognosis of the defect.

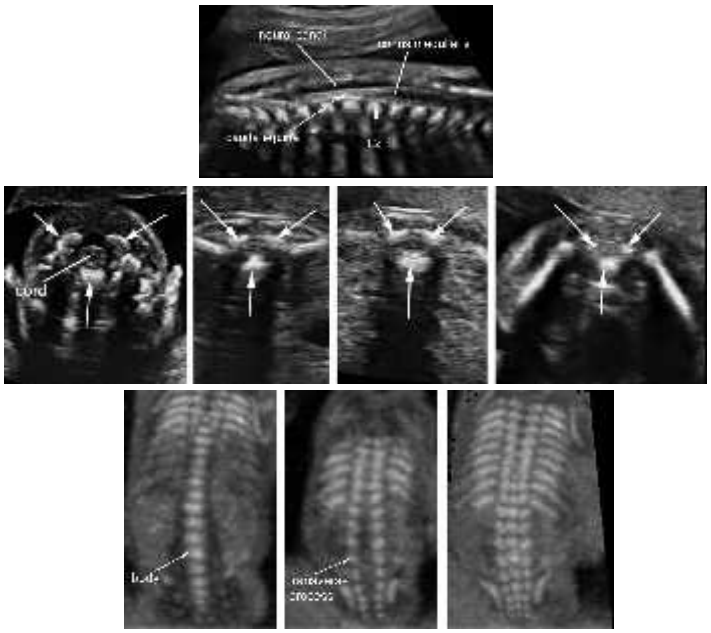
< 13 wks



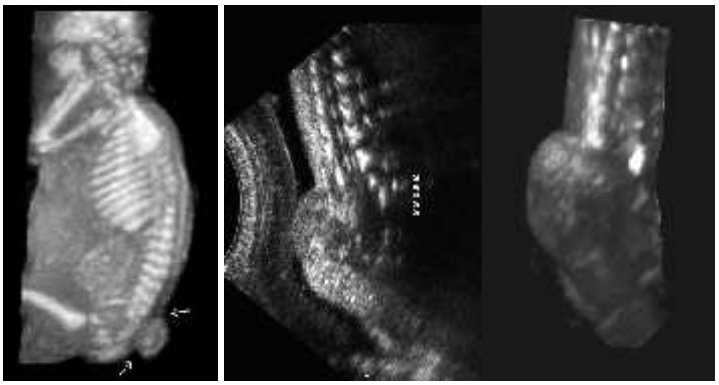
Absence of indirect signs such as raised AFP levels, head signs poses a challenge to the Sonologist for antenatal diagnosis of closed defects. In the absence of head signs, one has to look for spinal lesion at the lumbar & sacral levels. Careful examination is required to look for neural tissue. Recent references report that cystic mass is not a hallmark of closed NTD. As in the past, cyst wall thickness also does not indicate the diagnosis as open or occult defect. Cystic lesion with non homogenous echogenic area is usually seen in lipomas of the lower lumbar region. In such case, one should check for tethering of the spinal cord.

Head signs in NTD are definitely a blessing in disguise as its presence helps in detection of the lethal and highly morbid, open spinal defects. Though absence poses a challenge for diagnosis, it is reassuring of a closed defect. There can be exceptions to the general rule as in cases of open spina bifida with minimal degrees of Chiari II malformation. This is however rare.

Ultrasound imaging and diagnosis of NTD may be done using 2D / 3D & high resolution scanning techniques. With 2D / 3D machines, spine is examined ideally in prone position in patients with ideal maternal built & good liquor window. ***Spine should be imaged in all three planes - transverse, longitudinal & coronal.*** Extended imaging involves imaging of the spine cord & the conus medullaris.

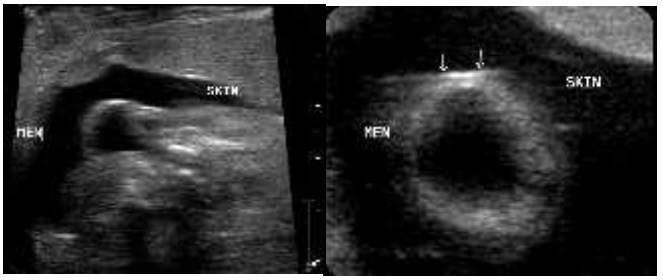


3D imaging is complementary to 2D imaging. Multiplanar views are more informative than rendered view. The level of defect correlates well with the 2D and postnatal imaging studies.



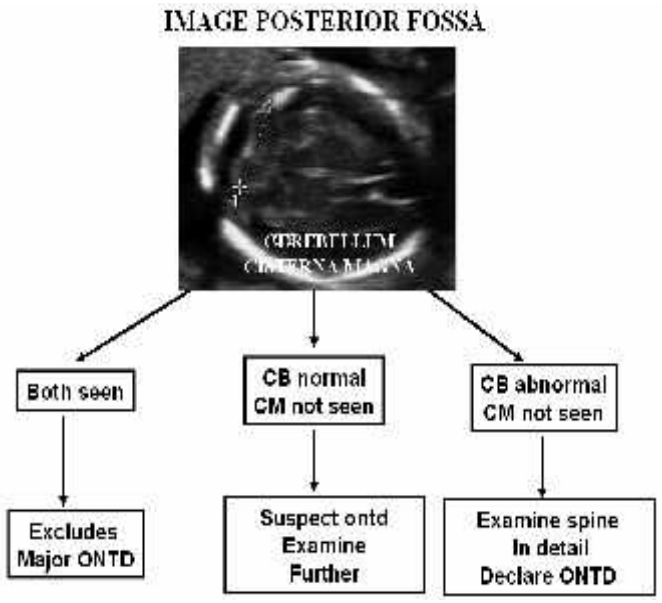
High resolution ultrasound has improvised the examination of the spine and the spinal cord. Ultrasound beam in the longitudinal position can be directed across the unossified spinous process which identifies the spinal cord within. The speaker while concluding mentioned that probes like L7.5- 8, L9 & L17 facilitate excellent imaging of the spinal cord delineating the skin from the underlying spinal defect. Distinction between open and closed spina bifida has prognostic implications. As mentioned, the outcome for infants with closed spina bifida is good, although neurologic symptoms of variable entity are frequently present. Above all correct diagnosis aids in genetic counseling of the family & provide them appropriate information.

Probe L17 MHz



DISCUSSION

During discussion Dr. Suresh gave an explanation for revisiting the topic on NTD in the BDR meeting. He emphasized that time has come now to declare a war against NTD in India. As seen from the statistics presented, NTD should not be detected in late second trimester with high end USG technology available today. It is easier to diagnose severe open defects between 11-13 weeks of pregnancy. He insisted that every Sonologist should image Posterior Cranial Fossa by following the algorithm as given below:



It is not enough to measure only the BPD, but cerebellar diameter should also be noted down. He suggested that it is also worthwhile estimating MSAFP levels on mandatory basis between 15-21 weeks of gestation for better detection of the defects at the population level.

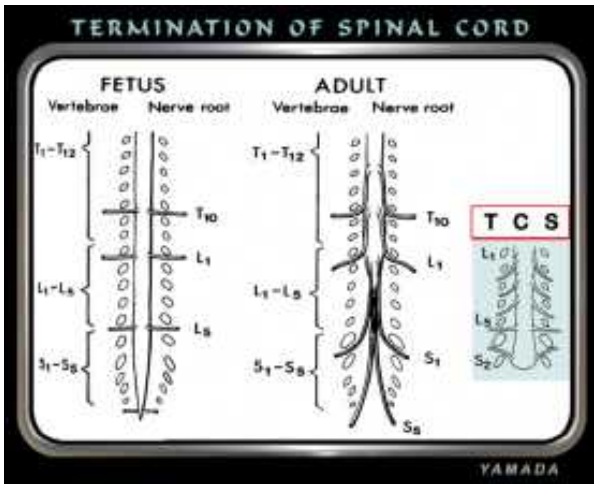
Postnatal Management of Closed Neural Tube Defects

Dr. Chidambaram Balasubramaniam,
Consultant Paediatric Neurosurgeon

Dr. B. Chidambaram provided the audience a candid picture about the current aspects of surgical management of closed neural tube defects presenting not only in the newborn period but also later in life. He said that the implications of surgical management of Tethered Cord Syndrome (TCS) & meningomyelocele are similar from the surgeon's & patient view point & further elaborated on TCS.

Professor S.Yamada, who has extensively researched on this defines TCS as a functional disorder that is manifested by neurologic symptoms in the lower extremities & bladder dysfunction. It is not only functional but also progressive, the surgeon added. This condition was first described in later 1800 & subsequently in 1900. Much has been written about it since then. The advances in prenatal & postnatal imaging techniques have paved way for rediscovery & rethinking of many aspects related to TCS.

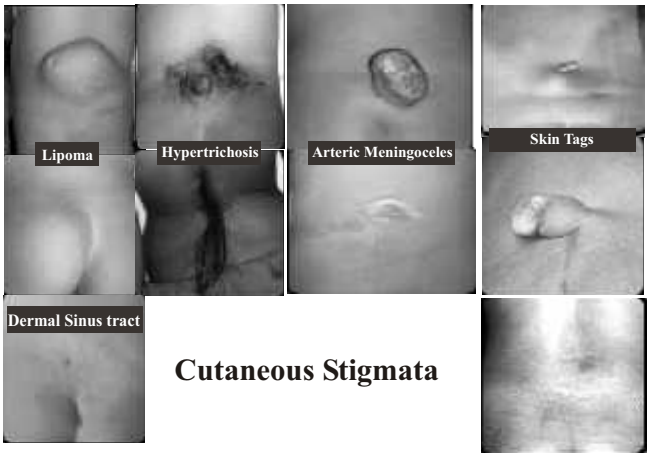
The pathology is often confined to the lower lumbar cord and conus which are low lying. *Tethering of the cord in the normal position is also seen*. It is known that the bones grow faster than the cord & the cord is held back. The basic pathology is anchoring of the cords that result in stretching during growth, which causes ischemia of the neurons. This is evidenced by the shift of the redox ratio of cytochrome aa3 to a more reduced state. Wrinkling & tears are seen in the cell membrane causing structural damage and this is maximal at the site of tethering. Normally the conus ends approximately between L1, L2 If the conus is seen below L2, it is abnormal. Antenatally it may be low placed even at L5 level. One has to also observe the roots while diagnosing the problem. Since conus in the fetus is low placed, the roots exit horizontally. As the baby grows, the cord ascends. The following diagram shows the exit of nerve roots in a normal fetus, adult and a child with TCS.



As seen the cord is held taut when it is not tethered. In a child with TCS there is micro trauma to the spine every time the child bends down or gets up

Various presentations of tethered cord include
1) Lipoma - Lipomeningomyelocele, Lipomas, Fatty Filum,
2) Split cord malformation - Dermal sinus tract, myelocystocele,
3) Atretic meningocele - tight filum,
4) Postoperative tethering following release of tethering,
Post meningomyelocele repair tethering &
5) Cervical meningomyelocele.

They manifest as cutaneous stigmata in more than 50% of the children postnatally. **Lipomas may present** as a small skin tag, a mass on or off the midline of the spinal cord, barely perceptible bulge in the back and if the gluteal cleft is pushed to one side, a lipoma on the other side of the spinal cord should always be suspected. **Hypertrichosis** seen as a tuft of hair or as a pony tail almost & always indicate split cord malformation. **Dermal sinus tracts** may be seen as a dimple, pinhole or a sizeable hole with inflammation & infection. When the site is pressed a cheesy material may ooze out from the dermoid inside. **Atretic meningoceles** are thought to be naturally healed defects. They may be seen as a blister or a small aberration with thin epithelium & this type of NTD is now called **Limited Dorsal Myeloschisis**. A small **skin tag** is not to be mistaken for a tail. True tail will have bony coccyx in it & it is very rare. **Haemangioma** on the midline of the back is not to be ignored & it denotes the underlying defect.



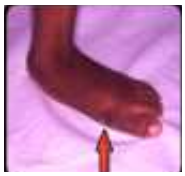
In cases of **Diastomatomyelia - Split Cord Malformation** (SCM) type I & II, depending on the type, the timing of repair varies. In type I, the cord is split into two hemi cords by a bony spur. The cords may unite above or below the split, but each hemi cord has a neural tube. In type II, no bony spur is found, there is arachnoid / fibrovascular septum splitting the cord in to two but both the hemicords have a single neural tube. SCM is used to be wrongly synonymous with the term **Dipolomyelia** where there is true duplication of the spinal cord with two hemi cords with its dorsal & ventral roots. This is a rare condition. Antenatal reporting of the type of SCM as I / II will help the surgeon decide on the timing of surgery he added.

There are neurologioc probems such as sensory motor deficit or orthopaedic problems of spine, lower lumbar anomalies & urologic problems such as incontinence & neurogenic bladder. The neurologic symptoms that present in almost three fourths of the affected include pain, weakness, areflexia, hyporeflexia, spasticity & sensory deficits. Children complain of pain that are poorly localized indicating a problem in the cord & not in the roots as one may experience in conditions of spinal disc prolapse. Unusually it can present as a non healing ulcer in the foot & this warrants examination of both the inguinal nodes & the back. Parapaeresis of the lower limbs is seen but weakness of the foot & toe is more common.

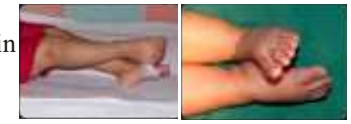
Neurologic Problems

Sensory and Motor Deficits

Non healing ulcer



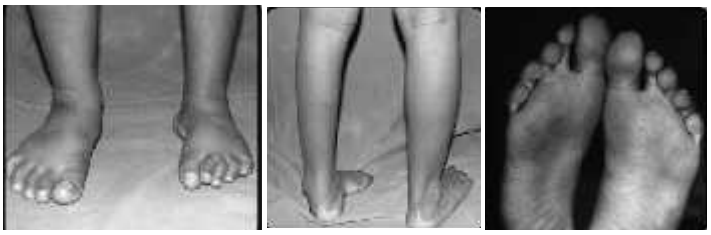
Parapaeresis in Lipoma



Weakness of LL Feet & Toe in Lipoma

Orthopaedic symptoms may present as asymmetry of limbs as shown:

Orthopaedic Problems
Limb deformity and size difference



Limb size difference is also seen in parietal lobe problems & hence this should not over looked. Other indicators of intraspinal anomaly are left sided scoliosis, wide interpedicular disctance & absent abdominal reflexes & rapid progression of symptoms. It is important to know that left sided scoliosis is not an idiopathic problem but a neurogenic problem.

The speaker mentioned that in a study along with Dr. Vijay Sriram, Consultant Orthopaedician, Chennai, it was observed that 26 children out of 31 children with spinal deformity had TCS.This revealed the close association of NTD with spinal deformities.

Urologic manifestation of symptoms presents with urinary incontinence & neurogenic hyperflexic bladder. Fecal incontinence is very rare. A child with congenital spinal problem will have severe constipation. Fecal incontinence indicates a severe damage of the conus or cauda equina. There is high incidence of associated anomalies found with closed NTD such as Anorectal, Urogenital, Renal & Tracheo Esophageal Fistula.

Management-Surgical

Neither the decision to operate nor is the surgical management of straight forward cases of tethering is difficult. The easy ones are tight filum terminale lipomas of filum. Next in the order are splitcord malformations, myelocele, dorsal lipomas & cervical myelomeningoceles. The most difficult ones are cases of re-tethering & transitional lipomas. While operating on a lipoma, it is safer to leave a bit of lipoma behind than to remove a bit of conus medullaris. No technology can prevent re-tethering. Tethering can follow any spinal surgery more so it is common after a surgery for TCS & meningomyelocele. Re-tethering after meningomyelocele repair may be due to scarring at the site of surgery or a new pathology.

Re-tethering is diagnosed only on clinical & urological parameters and not on imaging parameters. Children who have undergone repair of meningomyelocele must have follow up MRI to look for a second pathology. Post operative re-tethering has an incidence of 3-15% & it may increase further. This recurrence is seen in the ages between 2-15 years. The cord anchors in two sites 1. cervicomedullary junction & 2. at the site of surgery resulting in a "bow sling" effect ". This causes injury during movement & growth of the cord.

As mentioned earlier, TCS can present even when the conus is in normal position. The definitions of low "conus " as defined by Baston is :some variations in the level of termination (+ / - one segment) as in any other form although pure urologic problems may be unknown. The diagnosis is based on clinical urologic & imaging findings. If there are urologic symptoms due to neurogenic bladder, then untethering is recommended. The results are variable. Judgement is based on overall picture. Post surgical improvement in sensory motor deficits are seen upto 60%. More than 80% have amelioration of pain. Less than 65% show improved sphintcter / bladder function.

With recent technique of "clean intermittent catheterization" all those incontinent school - going children can be made socially continent in their environment. Their families need to be well motivated to adapt to such modifications throughout. Rate of surgical morbidity is very low with almost nil mortality. The symptomatic child needs untethering. Ideal timing is before the deficits become fixed. The earlier surgery is done in dermal sinus tract better is the outcome. It is wiser to stabilize the deficit on a prophylactic basis by performing the surgery as the natural history of the disease process needs to be studied further. But surgery is warranted in conditions other than lipomas. Despite successful surgery, nearly 10% deteriorate. Many of them who remain asymptomatic would remain so throughout. There are patients presenting with symptoms aas late as 18 - 19years of age & also in adulthood. Based on their clinical symptoms, the benefits of surgery are weighed in such cases. Surgical option is prophylactic in nature & hence it is fruitful when performed before the onset of symptoms in children.

Dr.Chidambaram while ending his lecture, listed a few queries to be answered still as for closed spinal defects are concerned. They were:

1. Is the pathophysiology only due to mechanical or metabolic problems?
2. Is prophylactic surgery the best since a subset deteriorate anyway?
3. How to deal with children who are asymptomatic?
4. How to deal children with neurogenic or non neurogenic bladder with conus in normal position?

He said that the biggest of all challenges is the "awareness" about the whole spectrum and the management of closed spinal defects among the professionals involved!

Discussion

When a fetus is antenatally diagnosed to have TCS and there is no neurological deficit postnatally, what would be the right time of correction?

The surgeon while answering said, the decision about the time of surgery depends on the family who are anxious and know least about the problem. He emphasized that meningomyeloceles are congenital anomalies that are most complex yet practically managable with normal longevity. Hence when it is antenatally diagnosed, pregnancy can be continued, if it is not major and the baby can be operated around when he or she gains 5kgs of weight to with stand the procedure. He also added that if an anomaly seen in the spine while doing a scan it is mandatory to look for anomalies in the brain and rule out any structural defects like Arnold Chiari malformation.

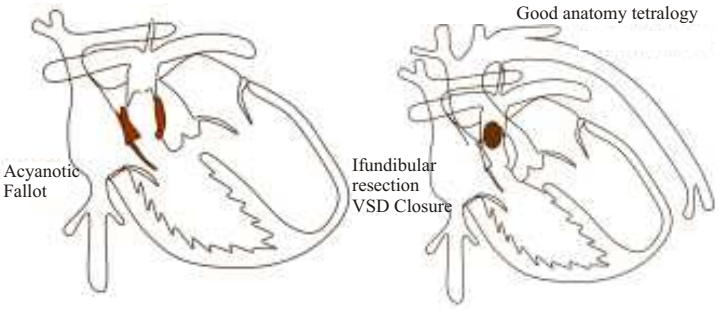
Correctable congenital cardiac anomalies

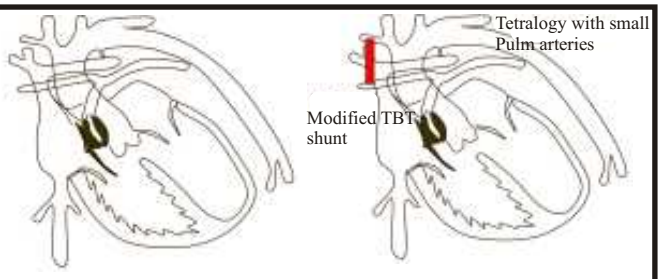
Dr M S Ranjit, Consultant Pediatric Cardiologist and Professor of Pediatrics , Sri Ramachandra University, Chennai

Dr. M.S Ranjit gave his lecture on CORRECTABLE CARDIAC ANOMALIES with good outcome on long term. He started off with a note that he would not speak on simple correctable congenital cardiac anomalies whose results were well known or about complex anomalies where no correction but only palliation was possible. His lecture was restricted to the topic chosen.

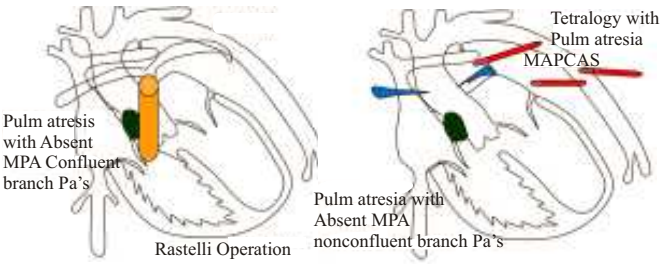
1. TETRALOGY OF FALLOT (TOF): Though Tetralogy of Fallot is a correctable anomaly, predicting long term outcome based on antenatal diagnosis would be difficult. The reason being that the disease has a wide spectrum with variable presentation and it could evolve from a mild to an extreme form. Extreme forms have different prognosis and specific details regarding postnatal management may not be available on Fetal Echo. Further questions which need to be answered are regarding the need for neonatal intervention or later, and whether single or multiple stage repair required. The **mildest form of TOF** is called the **Acyanotic Fallot,** where surgery is similar to a Ventricular Septal Defect (VSD) closure. TOF with good anatomy also has good prognosis which includes only VSD closure with infundibular resection. TOF with small pulmonary arteries require a **Modified Blalock Shunt** followed by intra cardiac repair. TOF with a small pulmonary annulus requires a **Trans-annular patch,** which may result in florid pulmonary regurgitation. It can cause right ventricular dilatation, dysfunction and arrhythmias in the long run.

TOF Correction



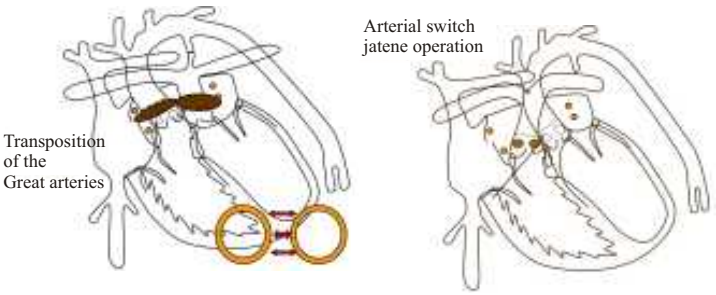


Regarding long term outcomes, he quoted a few literature for evidence. It was found that early repair of TOF resulted in significant acceleration of weight and height, with normalization of long term growth and fulfillment of genetic growth potential. Another study quoted that the rate of long-term survival after the postoperative period is excellent but remains lower than that in the general population and the risk of late sudden death was small. Pulmonary valve replacement is being considered for patients with florid pulmonary regurgitation and specific monitoring protocols are being devised for the same. A recent study published in May 2009 showed that valve sparing repairs are useful in preventing long term Right Ventricular Dysfunction. Regarding pregnancy in women with operated Tetralogy, it is well tolerated and an excellent neonatal outcome is expected. The recurrence risk of congenital cardiac disease, most often TOF, is high. When intellectual, neuropsychological, and behavioral functioning in children with TOF were assessed, a lower estimated full-scale intelligence was identified than in healthy peers and a neuropsychological profile characterized by primarily mild motor deficits and difficulties with language tasks were found. This is mainly contributed by the association of 22q microdeletion in these patients. Parents indicated attention problems and rated the child's school competencies to be lower than in healthy control subjects.Extreme forms of Tetralogy, like TOF with Pulmonary atresia, needs a Blalock Taussig Shunt followed by a conduit repair or a Rastelli operation. For those with Major Aorta Pulmonary Collateral Arteries, it requires a multi- stage repair involving high risks.



TRANSPOSITION OF GREAT ARTERIES (TGA): in TGA physiology, the circulation which normally runs in series is seen to run in parallel and is dependent on a shunt at the atrial, ventricular or ductal level for mixing of both circulations. Arterial Switch Operation (ASO) is performed in this condition, where the great arteries are switched over and anatomic correction of the circulation is obtained.

The major task in this operation is the transfer of coronary arteries which requires great expertise.



The potential problems following a ASO is Supravalvar Aortic and Pulmonary stenosis, Sub aortic stenosis, constriction of coronary orifices & left ventricular dysfunction. Many studies have shown encouraging results for TGA correction but the need for long term follow up to watch for complications cannot be overemphasized. Other surgeries for TGA are 1) Mustard and Senning operation or 2) Atrial Switch surgery, where physiological correction is achieved. Long term results are quite encouraging, yet constant follow up is needed.

ATRIO VENTRICULAR SEPTAL DEFECTS (AVSD):

These are wide spectrum of anomalies ranging from partial to complete or a clefted mitral valve. Primum defects are easy to repair but post operative complications like florid AV valve regurgitation and Left ventricular outflow tract (LVOT) obstruction are common. Complete AVSD s' are present in children with or without Down syndrome. The incidence of severe pulmonary hypertension and early onset of pulmonary vascular obstructive disease is common and hence early surgery is warranted. An article in the Annals of Thoracic Surgery 2008 has shown that definitive repair for complete AVSD can be performed in early infancy with excellent results. The two -patch technique is a safe and reproducible surgical method that can achieve low mortality and good midterm outcomes even in very young infants. He concluded his talk on the so called correctable anomalies with a remark that "you correct nothing, you only repair things"!

Discussion

1. When cardiac anomalies are antenatally diagnosed, which are the conditions that can be reassured to parents as correctable and compatible with normal life?

   TGA, TOF with good size of pulmonary arteries can be given reassurance other than the AV septal defects.

2. What are the cardiac conditons that warrant Karyotyping?

   TOF has significant associations with chromosomal anomalies and hence KT is warranted. Isolated TGA is seldom associated With chromosomal disorders.

3. What is the outcome of TAPVR ? (Total Anomalous Pulmonary Venous Return)

   TAPVR is of three types - supra cardiac, cardiac & infra cardiac. Infra cardiac types are more prone for obstruction which may alter the prognosis on the whole.

Annual Birth Defects Statistics Year 2008

Dr.N.Suganya, Epidemiologist, MediScan

Birth Defects Registry of India Annual Report 2008

Introduction

Worldwide the prevalence of congenital malformation is about 2 - 3%. Although nation wide prevalence estimate of birth defects in India is not known, a few small hospital-based studies indicate that it would be high. However, there is no systematic surveillance exist for birth defects in India. Having understood the lacunae in this area, Fetal Care Research Foundation (FCRF) established Birth Defects Registry of India (BDRI) in 2001. This is the eighth successive annual statistical report of BDRI. It presents the birth prevalence of birth defects estimated from 17 regional registries; viz. Chennai, Erode, Trichy, Lalgudi, Madurai, Nagercoil, Ramanathapuram, Dindigul, and Sivakasi in TamilNadu, Hyderabad in Andhra Pradesh, Bangalore, Mysore and Belgaum in Karnataka, Mumbai, Pune, Akola and Jalgaon in Maharashtra and Vis Nagar of Mehsana district in Gujarat. Data from Trichy and Lalgudi were combined as because they represent the same geographic area (Administrative district).

Program Description

BDRI is a hospital-based descriptive surveillance program, which passively collects (voluntary reporting) data on structural and chromosomal birth defects from hospitals of defined geographic areas. Collected data are checked by the Dysmorphologist before storing into the database. The diagnostic terms are then coded according to ICD10. Finally statistical analysis is done and the yearly annual report is presented to the members.

Results

During 2007 there were 1,21,515 births reported from the member registries. Of which 97.6% were live born. (Table 1). There were 1238 cases with birth defect(s). The over all crude birth prevalence is 101.9/ 10,000. (Fig 1). Tables 2A to 2J show the detailed anomaly specific and system specific crude birth prevalence. CNS anomalies were found to be high across all registries except Hyderabad and Nagercoil, (Fig. 2).

Salient features

1. High prevalence of birth defects were reported from Vis Nagar, Madurai & Dindigul registries followed by Chennai.
2. The prevalence of CNS & Musculoskeletal anomalies is high as compared to other system anomalies in all most all regions.
3. NTD repeatedly tops the list of anomalies this year too among the rest followed by CTEV & CLCP.
4. Low prevalence of birth defects were reported from Sivakasi & Ramanathapuram.

Table 1: Frequency of birth categories.

Categories	N	%
Live birth	118607	97.6
Intrauterine fetal death / still birth	2579	2.1
MTP for anomaly	329	0.3
Total births	121515	100

Fig 1: Crude birth prevalence of all anomalies and folic acid preventable anomalies across the registries

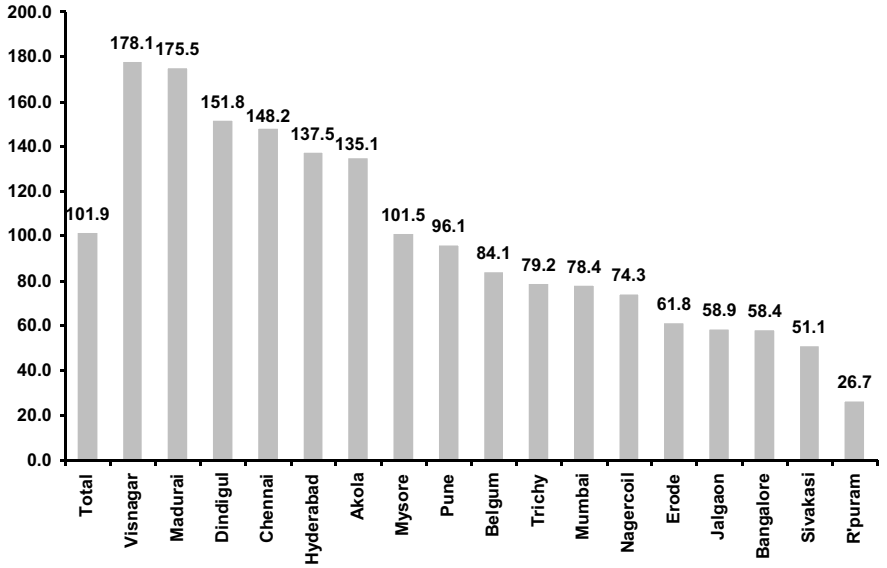
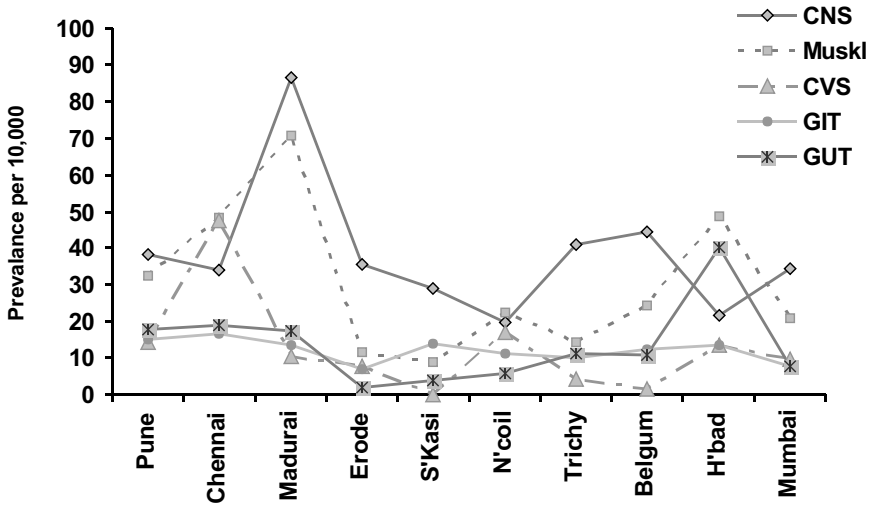


Fig 2: Crude birth prevalence of selected system anomalies across selected registries



Limitations of the data

Though the overall crude birth prevalence is 101.9 per 10,000, it is still underestimated since the expected prevalence is at least 2%. The plausible reasons could be, a. data not population based, (e.g. only 15.2% of total births are covered by the program in Chennai) b. data collection is not active, c. most of the minor anomalies might not have been reported, d. though multiple sources of data ascertainment was used, few cases from neonatologists and from pediatricians were reported. Though the estimated prevalence may not reflect the true population prevalence in those regions, it shows that the pattern and type of congenital malformations most likely to occur in those areas.

Table 2A-J: Anomaly specific and system specific crude birth prevalence of congenital malformations across all registries

Diagnostic Grouping	Number of cases	Prevalence / 10,000
A. Congenital Anomalies of the Central Nervous System (Q00 – Q07)	**482**	**39.7**
A01 Anencephaly (Q00.0) **(Incl. Acrania, Exencephaly, Iniencephaly)**	163	13.4
A02 Encephalocele (Q01.0 – Q01.9) **(Incl. Frontal & Occipital Encephalocele/ Meningocele)**	23	1.9
A03 Microcephaly (Q02)	18	1.5
A04 Congenital Hydrocephalus without Spina bifida (Q03.0 – Q03.9) **(Incl. Dandy – Walker malformation, Ventriculomegaly)**	96	7.9
A05 Spina bifida without anencephaly (Q05.0 – Q05.9) **(Incl. Meningocele, Meningomyelocele, Myelocele, Rachischisis, excluding Spina bifida occulta)**	154	12.7
A06 Holoprosencephaly (Q04.2)	8	0.7
A07 All other congenital malformations of brain, spinal cord & nervous system (Q04, Q06 & 07) (Incl. Agenesis of corpus callosum, absence of nerves, cerebral cysts and cerebellar malformations, etc.)	20	1.6

Diagnostic Grouping	Number of cases	Prevalence / 10,000
B. Congenital Anomalies of Eye, Ear, Face & Neck (Q10 – Q18)	**69**	**5.7**
B01 Anophthalmos / Microphthalmos / Macrophthalmos (Q11.0 – Q11.9)	10	0.8
B02 Absent external auditory meatus (Q16.1)	11	0.9
B03 Low set ears (Q17.4)	42	3.5
B04 All other congenital anomalies of Eye, Ear, Face & Neck (Q10 – Q18)	6	0.5

Diagnostic Grouping	Number of cases	Prevalence / 10,000
C. Congenital Anomalies of the Circulatory System (Q20 – Q28)	**176**	**14.5**
C01 Common Truncus / Persistent Truncus arteriosus (Q20.0)	2	0.2
C02 Double outlet right ventricle (Q20.1)	6	0.5
C03 Transposed Great vessels (Q20.3)	6	0.5
C04 Ventricular Septal Defect (Q21.0)	36	3.0
C05 Atrial Septal Defect / Patent or persistent foramen ovale (Q21.1)	14	1.2
C06 Atrioventricular septal defect / Endocardial Cushion Defect / Ostium primum (Q21.2)	3	0.2
C07 Tetrology of Fallot (Q21.3)	9	0.7
C08 Pulmonary valve Atresia (Q22.0)	0	0
C09 Ebstein's anomaly (Q22.5)	0	0
C10 Hypoplastic right heart syndrome (Q22.6)	8	0.7
C11 Other tricuspid valve abnormalities (Q22.8)	0	0
C12 Bicuspid aortic valve (Q23.1)	0	0
C13 Hypoplastic left heart syndrome (Q23.4)	15	1.2

C14 Dextrocardia (Q24.0)	2	0.2
C15 Patent ductus arteriosus (Q25.0)	33	2.7
C16 Anomalies of arch of Aorta (Q25.1 & 25.4)	9	0.7
C17 Anomalies of pulmonary artery (Q25.5 – 25.7)	15	1.2
C18 Persistent left superior vena cava (Q26.1)	0	0
C19 Single umbilical artery (Q27.0)	11	0.9
C20 Other specified and unspecified congenital heart anomalies (Q20.2, Q20.4, Q20.8, Q22.3, Q22.4, Q23.0, Q23.2, Q24.8, Q24.9 & Q25.8, Q26.2, 26.9)	35	2.9

Diagnostic Grouping	Number of cases	Prevalence / 10,000
D. Congenital anomalies of the Respiratory system (Q30 – Q34)	**12**	**1.0**
D01 Congenital cystic adenomatoid malformation of lung (Q30.0)	5	0.4
D02 Absence / Malformation of nose (Q30.1 – Q30.9)	0	0
D03 Laryngeal atresia (Q31.8)	0	0
D04 Tracheal atresia (Q32.1 – 32.9)	1	0.1
D05 Agenesis of lung (Q33.6, Q33.8)	5	0.4
D06 Other Respiratory anomalies	1	0.1

Diagnostic Grouping	Number of cases	Prevalence / 10,000
E. Congenital anomalies of the Gastrointestinal tract (Q35 – Q45)	**169**	**13.9**
E01 Cleft palate (Q35.0 – Q35.9)	18	1.5
E02 Cleft lip (Q36.0 – Q36.9)	19	1.6
E03 Cleft palate & cleft lip (Q37.0 – Q37.9)	61	5.0
E04 High arched palate (Q38.5)	5	0.4
E05 Other congenital malformations of tongue and mouth (Q38.2, Q38.3)	1	0.1
E06 Atresia of esophagus without fistula (Q39.0)	10	0.8
E07 Tracheoesophageal fistula with atresia (Q39.1)	0	0
E08 Tracheoesophageal fistula without atresia (Q39.2)	11	0.9
E09 Gastric outlet obstruction (Q40.0)	0	0
E10 Tubular Stomach (Q40.2)	2	0.2
E11 Absence, atresia and stenosis of small intestine (Q41.0 – Q41.9)	7	0.6
E12 Imperforate anus (Q42.3)	28	2.3
E13 Other Congenital malformations of large intestines (Q42.1)	0	0
E14 Meckel's diverticulum (Q43.0)	0	0
E15 Anomalies of liver and gall bladder (Q44.0 – Q44.9)	5	0.4
E16 Absent pancreas (Q45.0)	0	0
E17 Anovestibular fistula / Rectovestibular fistula (Q64.7)	0	0

E18 Other specified and unspecified gastrointestinal tract (Q43.1 – 43.9)	12	1

Diagnostic Grouping	Number of cases	Prevalence / 10,000
F. Congenital Anomalies of the Genital and Urinary Systems (Q50 – Q64)	**220**	**18.1**
F01 Congenital malformation female genital organs (Q50.0 – Q52.9)	0	0
F02 Undescended testis (Q53.0 – Q53.9)	13	1.1
F03 Hypospadiasis (Q54.0 – Q54.9)	23	1.9
F04 Other congenital malformations of male genital organs (Q55.0 – Q55.9)	7	0.6
F05 Indeterminate sex (Q56.4)	16	1.3
F06 Renal agenesis (Q60.0 – Q60.6)	21	1.7
F07 Cystic kidney disease (Q61.0 – Q61.9) (Incl. Infantile or Adult polycystic kidney and Multicystic dysplasia)	45	3.7
F08 Congenital hydronephrosis (Q62.0)	32	2.6
F09 Pelviureteric junction obstruction (Q62.1)	10	0.8
F10 Other ureter anomaly (Q62.4 – Q62.8)	1	0.1
F11 Other congenital malformations of kidney (Q63.0 - Q63.9) (Incl. Fused / Horseshoe kidney)	2	0.2
F12 Ectopia vesicae / Bladder exstrophy (Q64.1)	0	0
F13 Congenital posterior urethral valve (Q64.2)	5	0.4
F14 Other congenital malformations of bladder & urethra (Q64.3, Q64.8)	11	0.9

Diagnostic Grouping	Number of cases	Prevalence / 10,000
G. Congenital Anomalies of the Musculoskeletal System (Q65 – Q79)	**383**	**31.5**
G01 Congenital dislocation of hip (Q65.0, Q65.1)	4	0.3
G02 Talipes equinovarus (Q66.0)	132	10.9
G03 Other Congenital malformations of feet (Q66.1- Q66.9) (Incl. Rocker bottom foot)	8	0.7
G04 Congenital Musculoskeletal deformities of head, face, spine & chest (Q67.0 – Q67.9) Incl. Dysmorphic face (Q67.0)	30	2.5
G05 Congenital deformities of knee (Q68.2) Genu recurvatum	13	1.1
G06 Polydactyly (Q69.0 – Q69.9)	47	3.9
G07 Syndactyly and polysyndactyly (Q70.0 – Q70.9)	20	1.6
G08 Upper limbs - reduction defects / shortening (Q71.0 – Q71.9)	8	0.7
G09 Lower limbs - reduction defects / shortening (Q72.0- Q72.9)	8	0.7
G10 Unspecified limbs - reduction defects / shortening (Q73.0 – Q73.8)	10	0.8
G11 Arthrogryposis (Q74.3)	2	0.2
G12 Other congenital malformations of limbs (Q74.8 & Q74.9)	7	0.6

G13 Hypertelorism (Q75.2)	14	1.2
G14 Other congenital malformations of skull & face bones (Q75.0– 75.9)	44	3.6
G15 Spina bifida occulta (Q76.9)	0	0
G16 Other congenital malformations of bony thorax and spine (Q76.0 – Q76.8) (Incl. Scoliosis, Hemivertebrae etc)	23	1.9
G17 Osteochondrodysplasia with defects of growth of tubular bones & spine (Q77.0 – Q77.9)	21	1.7
G18 Osteogenesis imperfecta (Q78.0)	3	0.2
G19 Diaphragmatic Hernia (Q79.0)	36	3.0
G20 Absence / Eventration of diaphragm (Q79.1)	2	0.2
G21 Exomphalos / Omphalocele (Q79.2)	36	3.0
G22 Gastroschisis (Q79.3)	9	0.7
G23 Thanatophoric Dysplasia (Q77.1)	2	0.2
G24 Other congenital malformations of abdominal wall (Q79.5, Q79.6, Q79.8) (Incl. Limb body wall complex, Cloacal anomaly)	12	1.0
G25 Other specified and unspecified congenital malformations of musculoskeletal system	1	0.1

Diagnostic Grouping	Number of cases	Prevalence / 10,000
H. Other Congenital Anomalies (Q80 – Q86 & Q89)	**9**	**0.7**
H01 Icthyosis (Q80.8)	0	0
H02 Simian crease (82.8)	2	0.2
H03 All other congenital malformations not elsewhere classified (Q81.8, Q82.3, Q84.8, Q89.1, Q89.2)	7	0.6

Diagnostic Grouping	Number of cases	Prevalence / 10,000
I. Multisystem Anomalies / Syndromes	**29**	**2.4**
I01 Meckel Gruber Syndrome (Q61.9)	3	0.2
I02 Pierre Robin syndrome (Q87.0)	16	1.3
I03 Sirenomelia sequence (Q87.2)	3	0.2
I04 VACTREL (Q87.2)	1	0.1
I06 Other Syndromes (Q75.1, Q87.1, Q87.3, Q87.5, Q87.8, Q87.9)	6	0.5

Diagnostic Grouping	Number of cases	Prevalence / 10,000
J. Chromosomal Anomalies (Q90)	**21**	**1.7**
J01 Down's Syndrome (Q90.0 – Q90.9)	15	1.2
J02 Edwards' Syndrome (Q91.3)	3	0.2
J03 Turner's Syndrome (Q96.0)	3	0.2

Well done! Keep it up!!!

BDRI team wishes to place on record its appreciation to the regional registries of Pune, Madurai & Belgaum for their detailed Data coverage.

BDRI invites members of FOGSI to join FOGSI BDR & contribute data for a national cause. Obstetric centers of India may also join BDRI by browsing www.mediscansystems.org

Congratulations !

Dr. Anu Vij (Anurag Nursing Home) for commissioning the first FOGSI BDR - Navi Mumbai chapter with a team of participants & successfully steering ahead the project over 6 months.

This news letter is available at http://www.mediscansystems.org. quarterly - January, April, July and October.
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