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 04/02/06

The first birth defects registry meeting of the current year was held on the 4th of February at the premises of Mediscan Systems. Dr. Indrani Suresh, Director BDRI, welcomed the gathering for the CME on Birth Defects. She narrated the activities and new developments that happened over the past six months. She said that the registry has about 20 branches spread over the nation, many being in the state of Tamilnadu. She congratulated and extended a warm welcome to the newly joined nodal centres at Ramanathapuram, Madurai, Trivandrum, Pune & Mumbai. She also mentioned about the Newborn Screening program launched by FCRF as a measure towards birth defects prevention and requested the members to support the project to identify specific metabolic disorders in our population apart from providing data on birth defects. She informed the audience about the publication of an article by Dr. G. Thangavel (Epidemiologist-BDRI) titled "Methodological issues in setting up a Surveillance System for Birth Defects in India" based on our data collection methods, in the "National Medical Journal of India" (Abstract of the article given in the last page). She called for the support of NGOs to sustain data collection in registries which are finding it difficult to continue the mission founded for a cause. She thanked all the members for their untiring cooperation and she hoped to march ahead with their support to reach the goals.

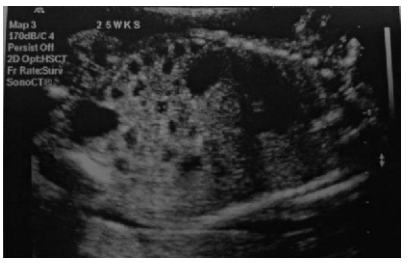
We thank all the participants of Chennai BDR for their interesting case presentations. The excerpts of the case presentations are given below.

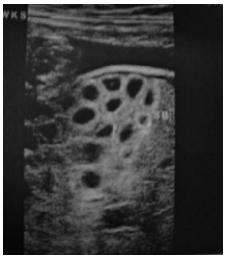
CONGENITAL CHLORIDE DIARRHOEA

Dr. R.Sowbarnika, Sri Ramachandra Medical College Hospital & Research Institute.

Dr. Sowbarnika presented an interesting and rare case of Congenital Chloride Losing Diarrhorea in a newborn.

Case Study: Mrs. V, 25 years with an obstetric history of G2P1L0A0 was referred at 31 weeks of gestation with an antenatal ultrasound (USG) report of polyhydramnios and fetal dysmotility syndrome/ ? Hirschsprung's disease. She was consanguineously (III degree) married for 3years. During her previous pregnancy, the USG of the fetus at 27 weeks was suggestive of Hirschsprung's disease. She delivered a male child normally in a Government Hospital. The child was not evaluated for Hirschsprung's disease probably because the child passed normal stools. This child died on the 26th day due to ? Neonatal sepsis/Jaundice. In the present pregnancy, she had a dating scan done in first trimester and an anomaly scan at 24-25 weeks which revealed dilated bowel loops and polyhydramnios.





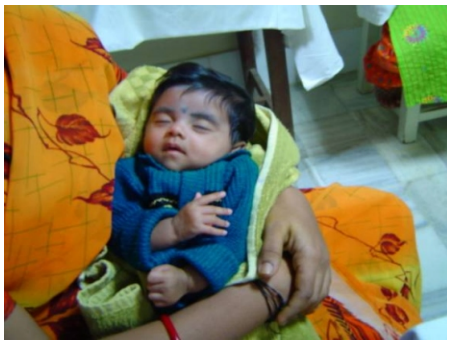
Cordocentesis was done and karyotype was found to be normal. She was again scanned at 25-26 weeks and had the same findings with an AFI of 28 & ? Hirschsprung's disease. She was given two doses of steroids at 32 weeks in view of polyhydraminos and anticipated preterm labour. She also had routine blood investigations which showed normal values. Although she was advised early admission, she got admitted only at 36.6 weeks with preterm labour. She had abdominal pain, leaking per vagina (p/v) for 4 hours (no bleeding) and her fetal movements were good. Her general and systemic examinations were within normal limits. Her uterus was over distended, acting and cervix was dilated to 1.5 cm. Fetal heart rate was 140/min.
She delivered a female baby the next day of her admission with good APGAR and she weighed 2.42 kgs. The child did not have vomiting or abdominal distension. In view of Hirschsprung's disease, rectal wash was given after 10 hours. *As expected there was no explosive passage of meconium instead the baby passed light coloured stools.* The USG report of abdomen was normal. The child was put on breast feeds from D2. On D3 she had Hyperbilirubinemia and was given phototherapy for 7 days. The baby was discharged later.

The child got readmitted after a week with weakness in the left upper limb. The Neonatologist suspected an Erb's Palsy due to forceful lifting of the child by the caretaker. But the mother also said that the baby had frequent passage of loose stools. There was visible gastric peristalsis. Investigations revealed hyponatremia- Na (118mg) & Hypochloremic (Cl-84mg) Metabolic Alkalosis without evidence of sepsis. The child was treated for Hyponatremia but did not improve on treatment. At this juncture, Congenital Adrenal Hyperplasia (CAH) was thought to be the cause of diarrhoea and investigated. Her Serum Cortisol was normal and 17 OH-P was in the upper limit of normal range. The baby was started on 3N saline and Inj.Cortisone which was changed to oral and tapered later. She was discharged on oral NaCl solution and the mother was taught to feed this after feeds every 2 hours. Subsequently the baby did not seem to thrive and exhibited the same problems loose stools & refractory electrolyte abnormality. On discussion, Dr. Sujatha Jagadeesh of FCRF suggested the possibility of Congenital Chloride Losing Diarrhoea in the child. Her stool analysis revealed a very high excretion of Chloride (80 mmol/L normal 2-3 mmol/L) clinching the above mentioned diagnosis.

Review of literature

This rare metabolic error first described by Gamble & Darrow of Finland in 1945, is an autosomal recessive condition manifested due toa defect in chloride reabsorption by the intestinal mucosa (distal ileum& colon).The defect occurs in the anion pump which transfers Cl & HCO3.The mean age of diagnosis is 3.2 months of age. The incidence rates reported are 1/3200 in Kuwait & 1/5500 in Saudi Arabia and also it is said to be high in Finland. As already discussed, it has classic symptoms of watery diarrhoea even in utero causing polyhydramnios, preterm birth,and no passage of meconium, abdominal distension and severe dehydration endangering life. The metabolic problems include, hyperbilirubinemia, hyponatremia with increased Renin & Aldosterone, hypo kalemia, hypochloremia and metabolic alkalosis.
Diagnosis is confirmed by fecal chloride levels > 90 mmol/L. The gene responsible for this condition is called CCD and the locus is mapped to chromosome 7q 31 adjacent to the Cystic Fibrosis gene locus (Kere et al).30 mutations have been reported so far in this condition. If not diagnosed early, this could lead to certain renal changes similar to hypertensive angiopathy.

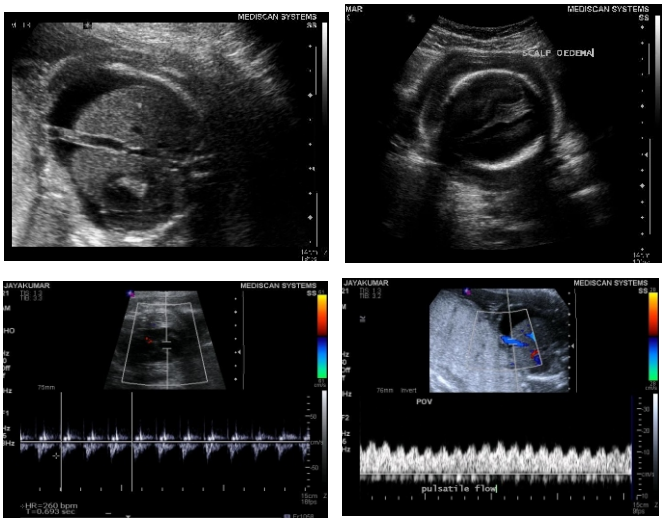
Dr. Sowbarnika concluded her lecture saying that it is wiser to diagnose this problem early and treat with simple oral NaCl / Kcl solution which prevents growth retardation, psychomotor delay and renal changes. The child has been followed up on two weekly intervals and her electrolytes are checked. The baby at present is healthy and growing well. During discussion, the need for "team work" among the medical professionals concerned in diagnosis & management of difficult cases was stressed for the benefit of the patient.



FETAL ARRHYTHMIA WITH NON IMMUNE HYDROPS (NIH) Dr. Indra Nedumaran, CSI Rainy Multi Specialty Hospital

Dr. Indra presented a case of NIH with fetal arrhythmia and the efforts of the medical team to save the baby until the end .

Case Study: Mrs K, 21 years old primi gravida was referred with USG findings suggesting Hydrops at 23-24 weeks of gestation. She had no dating scan done. She had no significant personal or family history of genetic problems. Clinically, her general health status including blood pressure, pulse rate & uterine size were normal. The fetal heart rate was found to be irregular and fast and hence she was referred for another scan. The scan findings were single fetus of 24 weeks of gestation with generalized edema, pleural effusion, pericardial effusion, gross ascites, no structural anomalies and FHR of 200 - 220 beats/mt. The fetal ECHO was normal. Mother's blood group was "O"positive & her IgG of Toxoplasmosis, Rubella and CMV were higher than normal indicating immunity to previous exposure.



Oral maternal transplacental therapy was started with T. Flecanide 100mg/bd and followed up with serial scans. 5 days after the administration, fetal heart rate was lowered to 130 beats/mt and after 2 weeks scalp edema and pleural effusion had disappeared. Ascites persisted. The mother at 27.3 weeks had preterm labour and hence she was treated with steroids and IV tocolysis. She continued taking flecanide.
She was again admitted at 28 weeks with leaking PV and active labour. She spontaneously delivered a male infant weighing 1.6 kg. The baby was depressed and the APGAR was 5/1mt. The baby was kept in NICU. The child was grossly edematous and about 130ml of ascitic fluid was tapped. His heart rate continued to be 180-200/mt. Since the baby did not respond to Digoxin, he was given Ionotropic support with dopamine. The baby was on ventilator due to severe respiratory distress. Chest X ray showed hypoplastic lungs, pleural effusion and no pulmonary edema. Surfactant was also given. On D3 the baby improved and was weaned from the ventilator. But on D4 he developed thrombocytopenia and pulmonary hemorrhage and reventilated. It persisted for another 4-5 days. By D7, dysmorphic features like low set ears, bilateral micropthalmia, corneal haziness and microcephaly were noticed. The baby also had PDA and did not pass meconium. The baby continued to have distress with features of CCF and thrombocytopenia. Chest X ray revealed bronchopulmonary dysplasia & cardiomegaly. Paediatric cardiologist felt that it could be PPHN / LV dysfunction secondary to pulmonary problems and the prognosis was poor. The baby tolerated small tube feeds. He was given Fursemide along with Digoxin. The baby's condition deteriorated suddenly and died on the 17th day. Dr. Indra briefed about the literature of Supra Ventricular Tachycardia (SVT) and use of the drug Flecanide.

Literature Review

SVT is an easily reversible and single most common cause of NIH. It is diagnosed accurately with cardiac imaging in early pregnancy.Early intervention may result in normal outcome with normal neuro development. The survival rate is around 85-95%. Associated anomalies or syndromes of SVT are rare.
The management of this condition warrants multi disciplinary approach. Transplacental drug therapy by administering oral anti arrhythmic drugs like Digoxin, Amiodarone and Flecanide in combination improves the outcome of the baby. Digoxin is effective when there is no hydrops. Sotalol is a promising agent. Flecanide is very effective for intrauterine treatment with or without hydrops.

Conversion into sinus rhythm can be expected 72 hours after initiation of therapy, but it may take up to 14 days. It has no terotogenic effects and 80% of maternal concentration of the drug crosses the placenta. Direct fetal therapy is warranted when the fetus does not respond to maternal therapy. Fetal intramuscular / intraperitonial injections which provide a sustained release is sought here. Sotalol is the drug usually used and Digoxin is added on when sinus rhythm is not achieved. Common antenatal complications include polyhydramnios and preterm labour. Most of them deliver by LSCS. Babies have to be re-evaluated postnatally. Most will require treatment of SVT if present. Digoxin / Amiodarone are used for long term therapy. Fetal tachyarrhythmias have better prognosis than bradyarrhythmias.

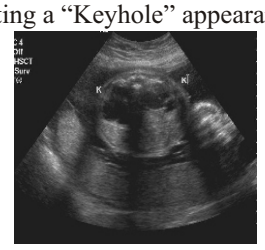
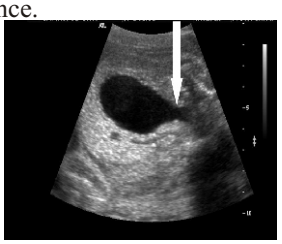
While discussing this case, Dr. Indrani Suresh enumerated a few points about SVT as given below :

1. SVT is mostly an incidental finding picked by the obstetrician on Pocket Doppler/Fundal height not matching the period of amennorrhoea due to polyhydramnios.
2. Since there may not be any changes in the movement pattern of the fetus with SVT, mother will not be able to sense the problem.
3. Hydropic babies with normal cardiac structure do well when treated.
4. SVT arises due to an aberration in the conduction system.
5. Autopsy though in this case was not done, would have thrown more light into the etiology of the anomaly.
6. It is ideal to have photo documentation of cases with dysmorphic features to get an accurate opinion. The dysmorphic facies described in the case presented could have been due to the edematous condition of the baby.
7. Though Flecanide has 80% penetration level across the placenta, it did not produce desired effect in this case probably due to placental hydrops.
8. The next pregnancy warrants close monitoring (USG at 11-13 weeks, 20 weeks and later at every 4 weeks) as the mother is at a higher risk compared to the background population.

She congratulated the team for their best supportive medical management throughout, even though the baby did not survive as he had other complications as well.

FETAL URINARY TRACT OBSTRUCTION - VESICO AMNIOTIC SHUNTING
Dr. Radhika Ramesh, Mediscan Systems.

Dr. Radhika Ramesh discussed in her presentation about how to select cases for vesico amniotic shunting procedure for a better outcome. She said that the commonest cause of fetal lower urinary obstruction is the posterior urethral valve (PUV). Other causes include, 1. urethal atresia 2.persistent cloaca and 3.hypospadias, epispadias/stenosis. She reviewed 167 cases of bilateral hydronephrosis reported at Mediscan Systems over a period of 3 years. Most of the cases (N-54) presented obstruction after 35 weeks of gestation.
When the first scan was done for bilateral hydronephrosis cases, 40% had normal liquor and 60% of them had oligohydramnios. *While those in the III trimester would deliver shortly and postnatally the baby could be managed, it is those in the late II trimester who pose a dilemma about intervention. By prenatal intervention one could prevent the consequences of lower urinary obstruction thereby saving the baby from progressive renal damage.*
The incidence of PUV is 1/5000 males. As already mentioned it is the commonest cause of severe obstructive uropathies. It occurs due to the failure of complete disintegration of urogenital membrane leaving membranous tissue within posterior urethra. It is diagnosed with ultrasound showing distended thick walled bladder with a dilated posterior urethra depicting a “Keyhole” appearance.

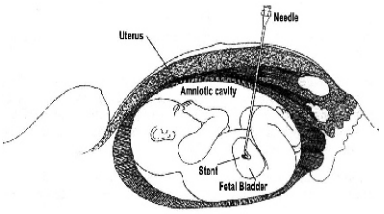
Other features include dilated ureters with bilateral hydronephrosis, variable liquor volume & increased cortical echogenicity with or without cysts. As cortical cysts are associated with irreversible, advanced renal damage fetuses presenting with this finding are not eligible for intervention. While working up a case for intervention, firstly the above mentioned USG features have to be diagnosed after 24 weeks of gestation. Other associated anomalies and perinephric urinomas have to be ruled out as they have poor prognosis. Fetal renal function has to be evaluated by vesicocentesis along with fetal karyotyping.

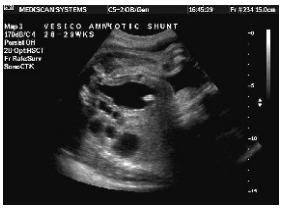
The following table explains the prognosis for fetal urine biochemistry

	Good Prognosis	Bad Prognosis
Sodium	< 100mEq/L	> 100mEq/L
Chloride	< 90mEq/L	> 90mEq/L
Osmolality	< 210mOsm/L	> 210mOsm/L
Calcium	< 2 mmol/L	> 2 mmol/L
Phosphate	< 2mmol/L	> 2mmol/L
β2 microglobulin	< 2mg/L	> 2mg/L

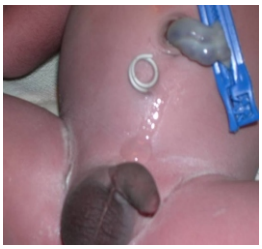
Eligibility for vesico amniotic shunting procedure
Cases with isolated lower urinary obstruction with an onset after 24 weeks of gestation and with a keyhole sign of the bladder on USG and good liquor volume are favourable for intrauterine vesico amniotic shunting procedure. Urine chemistry should show Na level of < 100 mEq/L (salt conservation), osmolality < 210 and β2 microglobulin < 2 mg/L indicating the absence of tubular damage.

Dr. Radhika went on to explain a case where successful shunting was done with a good outcome. Mrs. T.S with an obstetric history of G2P1L1 had a scan done at 23-24 weeks showing bilateral hydronephrosis and distended bladder suggestive of bladder outlet obstruction. The mother had a previous normal and healthy female child. The couple were counselled for the need for further testing to assess the prognosis and they agreed for the tests. Fetal urinary/blood sampling were done and the values were normal. She was called after 4 weeks for a scan and once again fetal urine sampling was done.
The values at 27-28 weeks revealed marginal deterioration of renal function and there was an increasing pressure on the upper urinary tract. Opinion was sought from a paediatric urologist and it was decided to do a vesico amniotic shunting to alleviate renal damage. The mother was started on tocolytics and antibiotics prior to procedure. A double pig tail shunt using 16 gauge trocar and needle was put under local anaesthesia. One end of the shunt coils inside the bladder and the other end in the abdominal wall into the amniotic cavity.





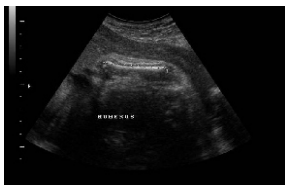
The shunting procedure was well illustrated with video clippings. The bladder size regressed the next day after shunting. A male baby was delivered by LSCS at 38 weeks with good APGAR.

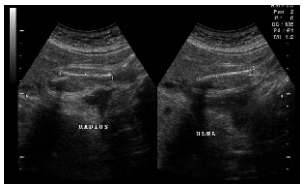


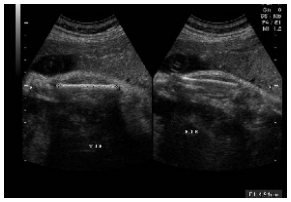
The baby underwent PUV fulgration and circumcision on D2 after birth. Post operative urine output was good and the renal parameters were also normal. After 2 weeks there was no urethral dilatation and bilateral hydronephrosis persisted.
At 2 months of age the baby had a good stream of urine with grade IV vesico ureteric reflux on the left side. By one year of age the urinary function was normal although there was some evidence of hydronephrosis which may take some time to resolve. Long term outcome of these children shows that majority have acceptable renal and bladder function and quality life. 1/3rd of surviving babies required dialysis and transplantation. Their neuro development was found to be normal. To conclude in utero diversion procedures are helpful only in selected cases. The fact that long term prognosis can not be predicted at the time of shunting has to be well explained to the parents.

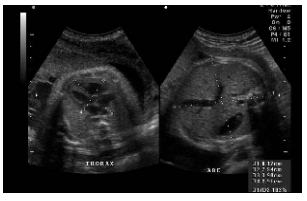
A CASE OF PRENATAL SHORT LIMBS - VALUE OF FOLLOW UP
Dr. Sujatha Jagadeesh, Fetal Care Research Foundation

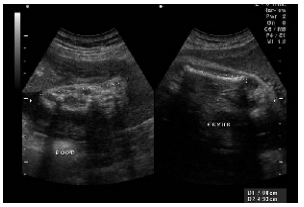
Dr. Sujatha Jagadeesh talked about the value of having long term follow up of a case antenatally diagnosed to have short limb skeletal dysplasia. A couple with III degree consanguinity was referred for a pregnancy with the USG finding of short limb skeletal dysplasia (SLSD). All long bones were less than 5th centile for gestational age.

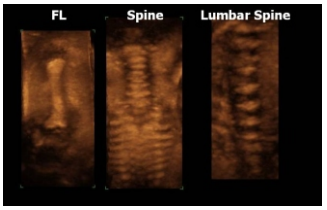




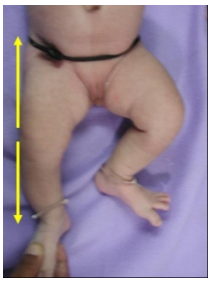


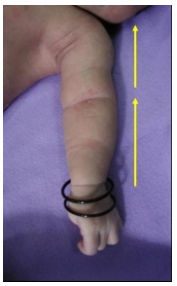


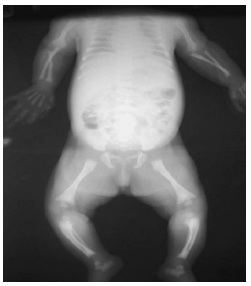




The prognosis of the fetus was to be assessed. Their family history did not reveal anything significant except that the grand mother of the fetus had a short stature of 135cm. Since the pregnancy was already 33 weeks and the contour, echogenicity of all long bones, thoracic diameter, skull and vertebrae were normal, the pregnancy was continued.
A girl baby was delivered at term and she had good cry and fed well. Clinically she had frontal bossing, depressed nasal bridge and rhizomelic shortening of the upper limbs. Rest of the examination of the baby was within normal limits







Her postnatal X ray showed short humerus,abnormal lower condyle, dislocation of superior radio-ulnar joints with normal pelvis and spine. Orthopaedic opinion was sought at this point and reduction of superior radio ulnar joints was attempted. But it failed. Since the baby had good elbow movements, surgical correction was also not opted during this period. When the baby was followed up at one and a half years of age, her developmental milestones were normal. Her upper limbs reached up to the iliac crest as against upper mid thigh.

The child's anthropometry recorded from birth to 1year are given below:

Age	At birth	7 mths	1 year	1.5 yrs
Ht/Lt cms	48 25%le	54 <5%le	67 <5%ile	74 <5%ile
Upper seg.	30.5	36	43	46.5
Lower seg	17.5	18	24	27.5
H.C	33.5 25%ile	41 5%ile	43.5 5%ile	44.5 <5%ile
Wt kgs	3.25	5	7	9

As seen, though the anthropometric measurements were good at birth, the child's catch up growth was not satisfactory.

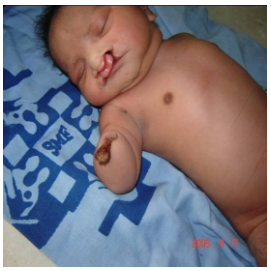
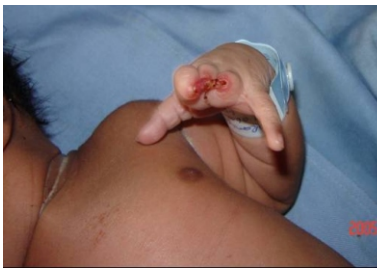
Dr. Sujatha described this genetic disorder as **Omodysplasia.** She also said, that all rhizomelic shortening with normal mental development are not termed as Achodroplasia. The term "Omo" stands for " Shoulder" in Greek. Omodysplasia is characterized by severe short stature, shoulder dysplasia due to defective growth of distal end of humerus, hypoplastic everted condyle, proximal radio- ulnar diastasis and anterolateral dislocation of radial head. This has both autosomal dominant / recessive mode of inheritance. Short humerii and characteristic facies are common for both modes of inheritance and in

dominant type the final height (4-4.5 feet) attained is better compared to recessive type.

The differential diagnosis for this condition include Skeletal dysplasia with Rhizomelia Patterson Lowry type, Humero Spinal Dysostosis and Familial Rhizomelic Dysplasia where short humerus is present as one of the features. Dr Sujatha on ending her presentation listed out the protocol for decision making for both lethal /non lethal types of skeletal dysplasias She also reminded the audience that SLSDs' do not warrant karyotyping as they are single gene disorders.

LIMB REDUCTION DEFECTS
Dr. S. Bhuvana, Sundaram Medical Foundation

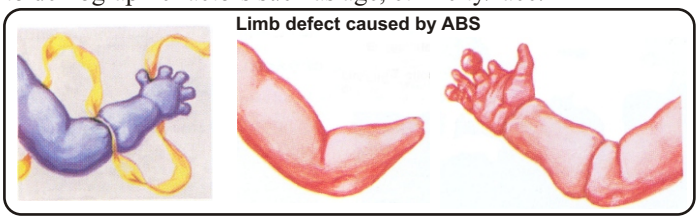
Dr. Bhuvana presented a case of limb reduction defect with a differential diagnosis of amniotic band syndrome.
Case Study: Mrs. R, 28 years with an obstetric history of G3 P1 L1 A1 with no consanguinity was referred for an USG. Her scan findings showed the gestational age as 18-19 weeks and no obvious anomalies. At 36 weeks she was admitted with bleeding p/v. She was well but for uterine contractions and irregular fetal heart rate on examination. Her blood investigation for coagulation profile and CBC showed all parameters fairly within normal limits except leucocytosis. In view of persistent irregularity of fetal heart, ECHO was done to rule out fetal cardiac anomalies. There was no cardiac abnormality other than occasional premature atrial contractions. Liquor was on the lower limit of normal and face & limbs surveillance was not possible due to reduced liquor. In view of bleeding p/v and elevated total count, sepsis was suspected and hence labour was induced. She delivered a female child normally, weighing 2.65 kg with good APGAR. Hind waters showed grade 1 meconium stained liquor. The baby presented with bilateral cleft lip/ complete cleft palate, shortening of right upper limb and transverse defect below the elbow level with gangrenous palm attached. The middle 3 digits were absent in the left hand which looked amputated rather than developmental error.

Cardiac rhythm was occasionally irregular. The couple was explained the need for surgical opinion and correction.
The parents were not willing due to financial constraints and took the baby home against medical advice.
Dr. Bhuvana went on to explain the diagnosis in this case with possible queries such as

1) Whether these problems could have been picked up in the mid trimester scan? (OR) at
2) What gestational age this mishap could have happened?

Limb reduction defects occur in about 1/2400 births and they are more commonly present in the upper limbs than in the lower limbs. They are diversified conditions depending on maternal age & ethnicity with different prenatal etiological causes. Reduction Polydactyly of thumb / big toe are seen in 2.5% of cases. defects such as shortened radius/tibia, missing fingers/toes make up for about 1/3 of the total. Multiple preaxial defects like 30-50% of the babies with limb defects have other associated anomalies. About 15--20% of the affected die before 1 year of age mostly due to associated defects. Though limb defects are picked up by prenatal USG, there is no screening program to diagnose them early. While discussing the etiology of limb defects she said that periconceptional folic acid containing multivitamins reduce the risk of delivering babies with limb defects by 36%. Folic acid seems to have beneficial effect on longitudinal defects of the arms & legs rather than transverse (amputation type) defects. The effect of other terotogens like corticosteroids, pesticides and tobacco (smoking) in causing limb defects have not been proved. The risk does not alter due to demographic factors such as age, ethnicity/race.



The speaker later gave a detailed account on the differential diagnosis that could be assigned for this case -
Amniotic Band syndrome (ABS). It is difficult to antenatally diagnose this condition. The incidence varies from 1/1200 1/15000 live births. 50% of cases have associated cleft lip/palate & club foot and 80% of them have associated hand & finger anomalies. The common view is that ABS occurs when the inner membrane (amnion) ruptures without injury to the outer membrane (chorion) exposing the baby to fibrous sticky tissue bands from the ruptured amnion which may float in the amniotic fluid. These fibrous tissues can entangle the baby reducing the blood supply, causing birth defects. In some cases a complete “natural” amputation of a digit or limb may occur before birth or the digit(s) or limb(s) may be necrotic (dead) and require surgical amputation postnatally. The timing of rupture is believed to occur between 28 days after conception 18 weeks of gestation. However late bands can occur and present at birth even after a normal USG earlier. Mostly they are detected by the effect they cause on fetal anatomy (missing limbs). Since ABS is not a genetic disorder it has no recurrence risk. Dr. Bhuvana concluded her talk with the management of limb defects with prostheses and physical & occupational therapy. Surgery is often needed in lower limb defects to straighten and stabilize the legs for prostheses fitting

VALUE OF AUTOPSY IN AN IUFD
Dr. Poornima Ramkumar, Fetal Care Research Foundation.

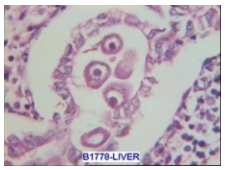
Dr. Poornima Ramkumar in her presentation claimed that a detailed autopsy helps in appropriate diagnosis of fetuses that undergo unexplained fetal demise in the womb. She explained her views with a case study. A woman with primi gravida and an H/O II degree consanguinity was referred for an ultrasound. The scan showed a single moribund fetus of 23-24 weeks of gestation with oligohydramnios, symmetrical IUGR, placentomegaly, ventriculomegaly and echogenic small bowel. The fetus died in utero within a couple of days after the scan

and the fetus was subjected to post mortem examination. The autopsy findings were

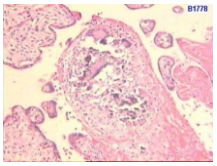
1) IUGR corresponding to 20-21 weeks of gestation,
2) Dysmorphic facies
3) Hypoplastic thymus & lungs
4) Cardiomegaly & hepatosplenomegaly.

Histopathology of organs showed CMV inclusions in the liver, kidneys, pancreas with placenta showing destructive villitis with granulomatous inflammation.

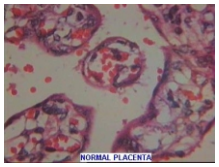
Hence the cause for IUGR & IUD was " Congenital CMV Infection" proving the value of autopsy.



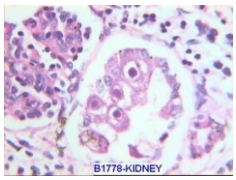
'Owl eye ' inclusion bodies of CMV virus in Hepatocytes which are ballooned out .



Destructive granulomatous villitisin placenta characteristic of CMV infection.



Normal Placenta



HPE of cells of various organs

Tubular cells in glomerulus are enlarged and show classical "owl eye nucleus" .

Abstract of the article published in National Medical Journal of India, Natl Med J India. 2005 Sep-Oct;18(5):259-62.

Methodological issues in setting up a surveillance system for birth defects in India.

Fetal Care Research Foundation,
203, Avvai Shanmugam Salai, Royapettah,
Chennai 600014, Tamil Nadu

Suresh S, Thangavel G, Sujatha J, Indrani S.

India is undergoing an epidemiological transition- communicable diseases are on the decline due to better living conditions and healthcare delivery. On the other hand, the relative increase in the prevalence of non-communicable, chronic and genetic diseases threatens to be a public health problem in India. One such group of disorders is congenital malformations. Though several studies have been done on congenital malformations in India since the early 1960s, coinciding with the thalidomide tragedy in the West, no uniform methods are available for the surveillance of birth defects. Each study has come out with varying results, not only because of the geographic variation in birth defects but also due to the varying standards adopted by each study in data collection, case definition and other methodological issues. Setting up a mechanism to understand the extent and nature of birth defects would involve the creation of a birth defects registry. The goals and objectives of such a registry should be formulated before it is set up. There are three types of registries-descriptive, analytical and preventive. These can also be classified as population - or hospital-based. Whether a registry is population - or hospital-based depends largely on the movement of mothers for delivery, registration of vital events in an area defined by the programme, as well as the resources available to the registry. Data can be collected in a passive or active manner, which also depends on the resources available to the registry. Every registry should have its own working definition of eligible cases to be reported, depending on the diagnostic services available in that area, and multiple sources of information should be used to improve the ascertainment rate. All the diagnostic terms should be coded and the information collected should be stored in a well-constructed database, preferably a relational type. Registries must evaluate their methods of data collection periodically to estimate the number of false-positive and false-negative reports. Ethical issues, cost and funding for the employment of various specialized professionals should be considered before setting up a registry.

Citation

Suresh S, Thangavel G, Sujatha J, Indrani S. Methodological issues in setting up a surveillance system for birth defects in India. *Natl Med J India 2005; 18:259-62.*

New BDRs Commissioned...

Welcome to our fold, together we shall realize our goal !

REGISTRIES	NODAL HOSPITALS
Madurai	Sundaram Scans
Ramanathapuram	Kanagamani Hospital
Trivandrum	Trivandrum Medical College Hospital
Pune	Deenanath Mangeshkar Hospital & Research centre
Mumbai	Dr. Bedekar Hospital

We are proud to announce that, Dr.Sujatha Jagadeesh, Geneticist & Dysmorphologist (FCRF) was invited to participate in the Taiwan Human Genetics Society Workshop & also in the HUGO Conference. Geneticists from all parts of the globe meet & share their rich academic experiences in this forum.

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