BDR NEWS

The official news letter of The Birth Defects Registry, Chennai
(Unit of Fetal Care Research Foundation)

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Proceedings of the 4th meeting of the Chennai Birth Defects Registry held on the 24th of January 2002

The fourth meeting of the Chennai Birth Defects Registry was held on the 24th of January 2002. Dr. S.Suresh welcomed the gathering. The continuing medical education (CME) meeting as planned, was turned out to be a useful learning session for the members. The overwhelming response from the members was heartwarming. This newsletter carries an abstract of all the birth defects cases presented at the meeting from various member institutions.

Dr. Sujatha Jagadeesh said that the registry will present the statistics on annual birth defects in the next meeting to be held in March 2002 and proposed vote of thanks at the end. Let us hope to have more informative, interactive sessions on birth defects in future and establish a National registry for birth defects in the years to come.

Case presentations

1. Hypoplastic left heart syndrome

Dr. Arnab Basak, E.V.Kalyani Medical Centre, Chennai

Mrs. D.S, 23 years G2 P1 was referred for ultrasound in the second pregnancy. She has one live male child. No history of consanguinity or birth of anomalous babies in her family . Ultrasound (USG) and fetal echocardiography revealed a single fetus with "hypoplastic left heart syndrome" (HLHS). The parents were counseled about the poor prognosis of the fetus and they opted for termination of pregnancy which was done at 22 weeks. Autopsy revealed a hypoplastic left heart with atretic left ventricle, aortic and mitral valves and hypertrophied right ventricular wall. These were consistent with the USG diagnosis.

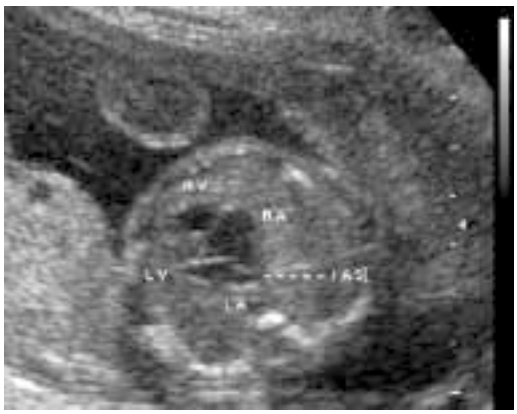


Fig. 1.1: USG picture of HLHS

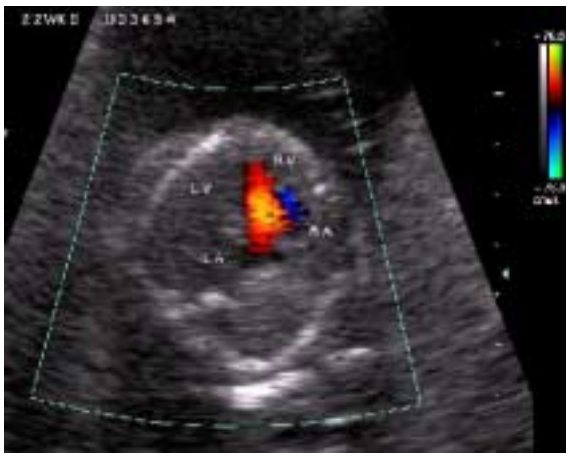


Fig. 1.2: Doppler USG shows absent blood flow in left heart

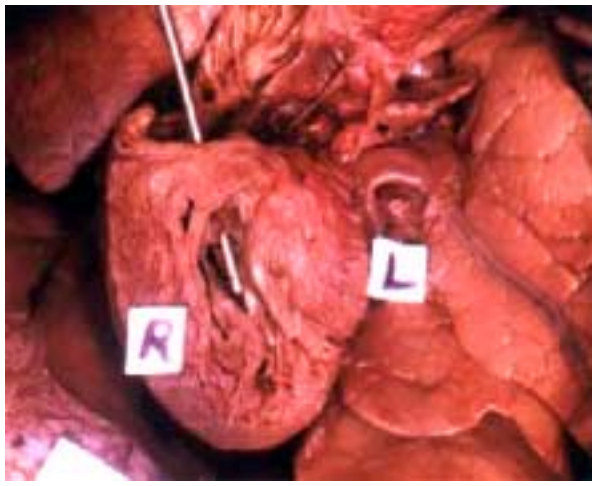


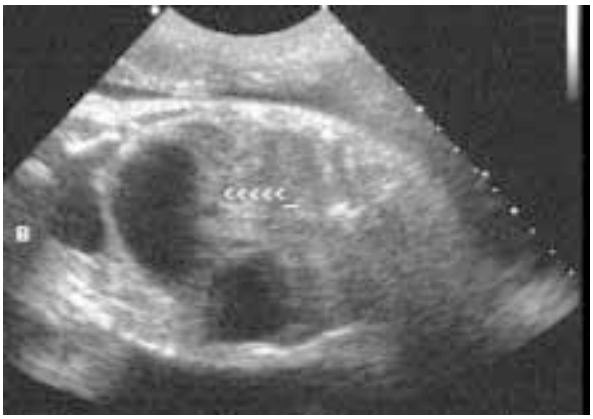
Fig. 1.3: Autopsy picture shows obliterated left ventricular cavity & thickened right ventricular wall

The presentation concluded with a detailed discussion about the characteristic features on USG for the diagnosis of HLHS and its associations, etiology, counseling and management options. The diagnosis is facilitated by demonstrating absence of filling of the left sided chambers of the heart by colour flow mapping. HLHS is usually a sporadic disorder though a few reports of autosomal recessive inheritance have been reported. Chromosomal associations are very rare and hence a fetal karyotype may not be necessary. The prognosis is poor and the babies die within a few days after birth. Norwood three-stage surgery is available as a corrective measure but survival rate is almost negligible. Cardiac transplantation may be the only option. The risk of recurrence is 5-10% and a cardiac echo is mandatory between 22-24 weeks in subsequent pregnancies.

2. Distended proximal bowel loop - Antenatal diagnosis at 34 weeks of pregnancy

Dr. Mona Pandurangi and Dr. Mythili Sivaraman,
Sahishnatha Vijaya Institute of Child Health and Maternity Centre, Chennai

A 23-year old primi gravida with known family history of anomalies was scanned at 20 weeks. No abnormalities were detected at that time. Patient was admitted at 34 weeks with preterm labour. The ultrasound (USG) showed a dilated bowel loop in upper abdomen and polyhydramnios. Kidneys were of normal size and no other anomaly was found.



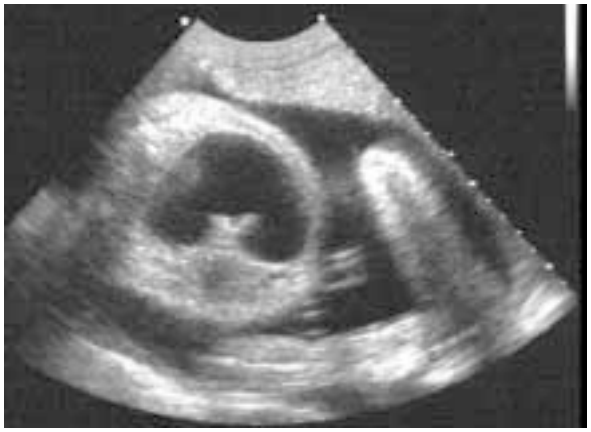


Fig. 2.1 & 2.2: D*ilated bowel loop seen in USG*

Parents were informed of the possibility of proximal bowel obstruction and the need for surgery in the immediate neonatal period. The mother was given steroids for short-term tocolysis. She went into active labour at 34 weeks and was delivered by forceps. The baby weighed 2.3kgs and the APGAR scores were 8 and 9 at 1 and 5 minutes respectively. 100ml of fluid was aspirated from the stomach. General condition of the baby was good with adequate peripheral perfusion, respiration and activity. Initial X- Ray showed a dilated duodenal loop. Postnatal USG revealed a dilated proximal bowel loop. A diagnosis of jejunal atresia was made and the baby was taken up for surgery.

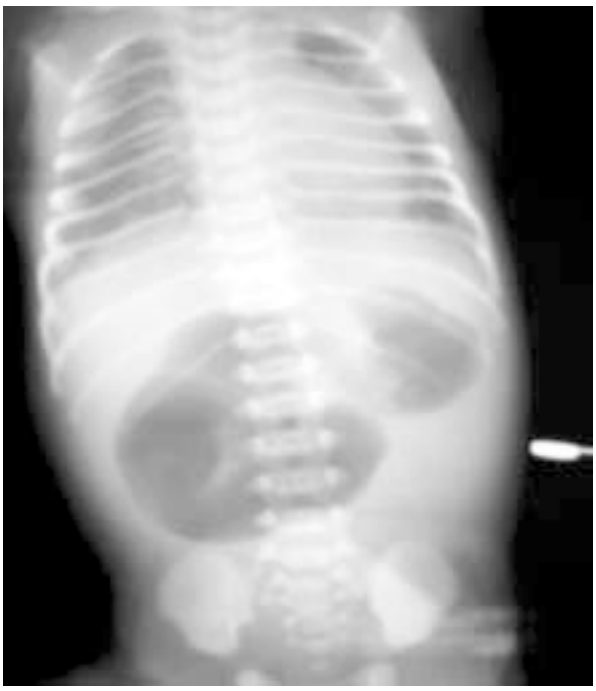


Fig. 2.3: X- Ray shows dilated duodenal loop

Intraoperative diagnosis showed dilated deuodenum, jejunal atresia type III (multiple, proximal, jejunal), inspissated meconium, precarious blood supply to small bowel and hypoplasia of small intestine.

During surgery Ladd's band and proximal jejunum (atretic segment) was excised and a side-to-side jejunoduodenal anastomosis was carried out. During the postoperative period, persistent gastric aspirate was done, partial parental nutrition was given and the baby passed stools on the fifteenth day. The baby was discharged on the seventeenth day and was fed with breast feeds alone.

The baby developed jaundice (cholestasis), anemia, bleeding tendency & hypocalcemic seizures in the first three months of postoperative period and was managed appropriately. Now at six months the baby has gained weight with normal calcium level and improved haemoglobin.

The most important questions that need to be addressed in this case are:

What counseling would we give if a dilated bowel was seen at 24 weeks scan?

Is the prospect of having a short bowel syndrome more of an intraoperative diagnosis than an antenatal one?

The answers to these questions were discussed in detail.

3. Femoral hypoplasia with unusual facies syndrome

Dr. Jayalakshmi, Fetal Care Research Foundation, Chennai

Case I:

Mrs. KF of 29 years (G2P1) with 3rd degree consanguinity and uncontrolled diabetes, came to us for routine antenatal ultrasound (USG)at 9-10 weeks. Both her parents were diabetics. Her first male child was 3.5 years old with dysmorphic features such as depressed chest, callosities

in both hands, absent transverse crease in left hand, absent crease in left thumb, bulbous finger tips, absent left fore arm bone, short and bent femur in both legs, wasting of muscles in both legs and right congenital talipes equinovarus (CTEV). Systemic screening showed all the systems as normal.

Hence, we advised her to come again at 16 weeks and she came to us at 14-15 weeks. USG showed increased nuchal translucency and skeletal dysplasia. Counseling was done regarding the recurrence of same previous problem. The pregnancy was terminated elsewhere and postmortem was not done.

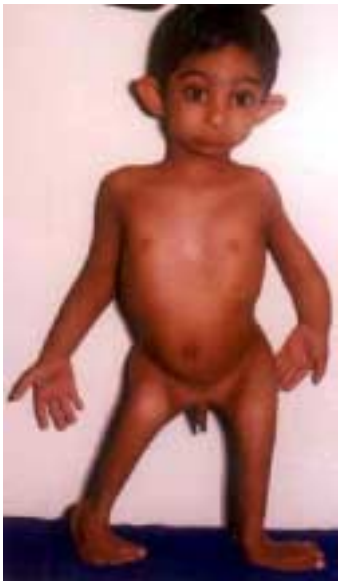


Fig. 4.1: Dysmorphic features of the First child

Case II:

A 27 years old Mrs.MK of G5P3A1L0 obstetric history, non-consanguinous, known diabetic and pregnancy induced hypertension (PIH) in previous pregnancies was scanned at 34-35 weeks. The USG showed skeletal anomaly with bilateral shortening of both femur with severely affected left femur. The rest of the other long bones fell in the 5th percentile for the corresponding weeks of gestation. Reduced mineralization of long bones and calvarium was seen. Abnormal facial profile (micrognathia) was noted. Subsequently the baby was delivered at 35 weeks and it died immediately after birth. Autopsy revealed a male baby weighing 2.5 kgs with dysmorphism, shortened lower limbs, bilateral club foot, short femur, posteriorly placed head and syndactyly in left foot. Internal organs and placenta were normal.

The possible theory of pathogenesis is that the femoral growth is inhibited during early organogenesis in diabetes complicating pregnancy. The mid diaphysis usually formed by subendochondral ossification is replaced by intramembranous ossification, thus inhibiting the growth of femur. The mode of inheritance may be autosomal dominant, maternal diabetes mellitus, sporadic or disruptions.

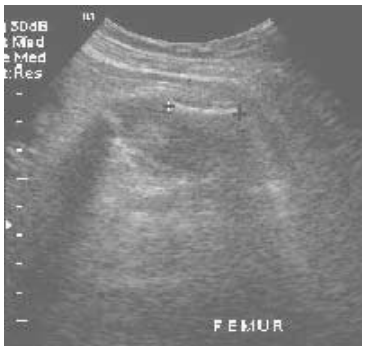


Fig. 3.2: USG shows short femur

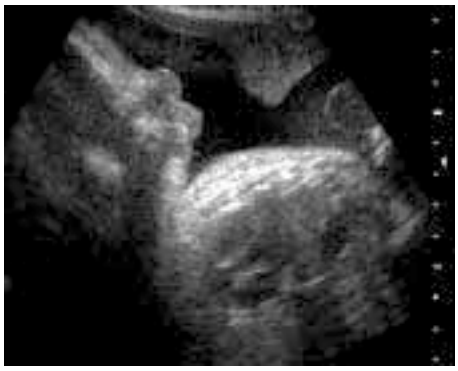


Fig. 3.3:Facial profile view showing micrognathia

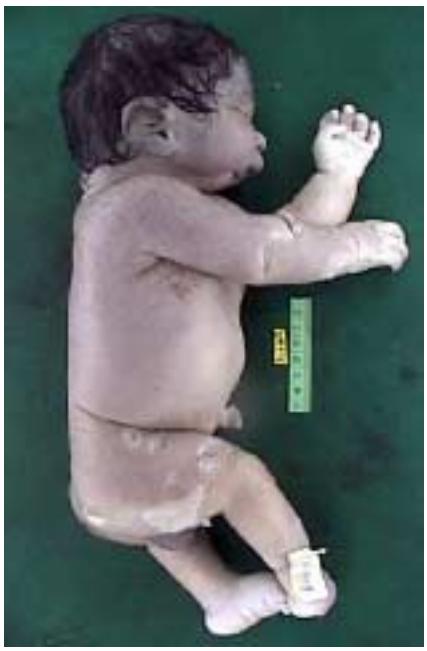


Fig. 3.4:Postnatal clinical picture shows micrognathia & short femur

4. Eventration of Diaphragm & Diaphragmatic Hernia

Dr. M. Jayasree, C.S.I. Multispeciality hospital, Chennai

Case-1:

Mrs. V. 27 years, G2A1, with no significant medical /family history was admitted as an unbooked case at 40 weeks. She was posted for emergency LSCS in view of oligohydramnios and cephalopelvic disproportion. She delivered a male child weighing 2.2 kg with APGAR score 9/10.

At 48 hours of life, baby developed tachypnoea. Chest X-Ray showed left hemidiaphragm at the level of 4th rib. Heart was on the right side and bowel loops were seen in the left hemithorax. A diagnosis of eventration of diaphragm was made and the baby was posted for surgery on the following day. At surgery, the diaphragm was found to be thin in the central portion and muscular at the periphery. The central portion was excised and approximated with 4-0 Prolene. Immediate chest X-Ray showed left hemi diaphragm in normal position. Postoperative period was uneventful and the baby was discharged on the 10th day.

Case-2:

Mrs.B.22 years, primigravida, was referred for a routine ultrasound (USG) at 18-20 weeks which showed a left sided congenital diaphragmatic hernia. After detailed counseling, the parents opted for continuation of pregnancy. A repeat USG at 34 weeks reconfirmed the diagnosis. The possibility of postnatal surgical correction was discussed with the parents. At 36 weeks, an emergency LSCS was done in view of CPD. A female child with abirthweight of 2 kgs was delivered with APGAR score of eight and three at 1 and 5 minutes respectively. Placenta was normal. Half an hour after birth, the baby had to be ventilated due to gasping respiration. At 24 hours, the baby was taken for surgery. Intraoperative findings showed stomach, small intestine, transverse & descending colon, spleen and left adrenal in left hemithorax. The defect was closed with a mesh. After an unevetful twodays, the baby developed pulmonary hypertension on the 3rd day and deteriorated rapidly. She died on the 4th day due to severe pulmonary hypertension.

Poor prognostic factors in this case were:

Early development of hernia
Early fetal distress (less than 2 hours)
Development of pulmonary hypertension.

5. Fetal wastage in assisted reproductive technology and how to reduce it.

Dr. Kamala Selvaraj, GG Hospital, Chennai

A retrospective analysis on 3720 ART procedures from 1995-2001 was presented. Among the 1096 (29.5%) pregnancies achieved, fetal wastage was 328 (29.9%). Among the 547 patients who delivered, 179 patients went into preterm labour and premature rupture of membrane (PROM) between 28-35 weeks of gestation. There were 122 twins and 57 triplets. Excellent antenatal and neonatal care resulted in 173 take home babies. The average weight of the babies was between 800-2200gms.

There were 34% missed abortions. Karyotyping of these missed abortions revealed triploidy as the commonest abnormality.·Increasing maternal age decreases the pregnancy rate and increases fetal wastage, which is due to aging oocyte and lowered receptivity of the endometrium.

Member hospitals

Name of the Hospital	Code
Mediscan Prenatal Diagnosis & Fetal Therapy Centre	001
EV Kalyani Medical Centre	002
Kanchi Kamakoti Child Trust Hospital	003
Sundaram Medical Foundation	005
Vijaya Hospitals	006
Apollo Hospitals	007
Sri Ramachandra Medical College Hospital	008
Durgabai Desmukh General Hospital	009
Corporation Hospital, Saidapet	010
Public Health Centre, West Mambalam	011
CSI Rainy Multi Specialty Hospital	012
CSI Kalyani Hospital	013
Nagamani Hospital	014
G.G. Hospital	015
Devaraj Manikchand Maternity Hospital	016
Ritherdon Nursing Home	017
Lakshmi Maternity Hospital	018

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