# B D R News

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

Volume 6 Issue 2: April 2006


**Proceedings of the birth defects registry meeting held on 20/05/06**

The second meeting of the year was held on 20th May 2006 in the premises of Mediscan Systems Annexe, Chennai. Dr.S.Suresh extended a warm welcome to the audience. The Annual BDR Statistics (year 2004 -2005) was presented by Dr. G. Thangavel (Epidemiologist- BDRI). Before his talk, a film show on a metabolic disorder"Urea Cycle Defect"was shown to promote the concept & to facilitate the creation of support groups for various genetic conditions in India. *Following are some of the salient features of the report presented*

★ The prevalence of birth defects was found to be more in IUFD/ SB category than in live births (820 Vs 50).
★ Chennai BDR contributed maximum data to the Central Registry (26 %) followed by Erode & Sivakasi registries
★ 450/673 fetuses/babies had isolated anomalies and the rest had multisystem/single system associated anomalies. Central Nervous System (CNS) & Musculoskeletal anomalies constituted the majority of the defects reported
★ 98.3% had major anomalies and the rest had minor anomalies such as low set ears, hypospadias, malformed pinna, pre auricular skin tag etc.
★ Neural tube Defects (NTD) accounted for 74% of CNS anomalies.
★ Ventricular Septal Defects were found in 48% of Cardio Vascular System ( CVS) anomalies.
★ Cleft Lip & Palate ( CLCP) constituted the most common Gastro Intestinal Tract anomaly & Congenital Talipes Equino Varus (CTEV) deformity contributed to major musculoskeletal deformity.

**Birth defects registry of India - Annual Report - 2005**

*Introduction*

The prevalence of congenital malformation worldwide is about 2 - 3%. Although the nationwide prevalence estimate is not known in India, a few hospital-based studies indicate that it would be more than what meets the eye. However, no systematic surveillance program exists for birth defects in India. Having understood the lacunae in this area, Fetal Care Research Foundation (FCRF) established the Birth Defects Registry of India (BDRI) in 2001.

Following is the fifth successive annual statistical report of BDRI. It presents the prevalence of birth defects estimated from 12 regional registries; viz. Chennai, Erode, Trichy, Lalgudi, Nagercoil, Ramanathapuram, Coimbatore and Sivakasi in TamilNadu, Hyderabad and West Godavari in Andhra Pradesh, Bangalore in Karnataka and Vis Nagar of Mehsana district in Gujarat. Data from Trichy and Lalgudi were combined as they represent the same geographic area (Administrative district).

**Program description**

BDRI is a hospital-based descriptive surveillance programme, which passively collects (voluntarily reported) data on structural and chromosomal birth defects from hospitals of defined geographic areas. The data collected are checked by the Clinical Dysmorphologist before storing in the database. The diagnostic terms are then coded according to ICD 10. Finally statistical analysis is done and the annual report is presented to the members.

**Results**

During 2005 there were 69,514 births reported from member registries. Of which 97.9% were live born. (Table 1). There were 673 cases with birth defect(s). The over all crude birth prevalence is 96.8 / 10,000. High prevalence was reported from Hyderabad and low prevalence reported from West Godavari (Fig 1). Though over all CNS anomalies were high, musculoskeletal anomalies was the leading group of defects in Chennai & Hyderabad (Fig. 2). Tables 2A to 2J show the detailed anomaly specific and system specific crude birth prevalence across all registries.

**Limitations of the data**

Though the overall crude birth prevalence is 96.8 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 12% of total births are covered by the programme in Chennai)
**b.** Data collection is not active,
**c.** Most of the minor anomalies may not have been reported,
**d.** Only a few cases from Neonatologists and Pediatricians were reported which is not sufficient .
**e.** 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 1: Frequency of birth categories.**

| Categories | N | % |
|---|---|---|
| Live birth | 68021 | 97.9 |
| Intrauterine fetal death /still birth | 1268 | 1.8 |
| MTP for anomaly | 228 | 0.3 |
| ***Total births*** | 69514 | 100 |

*Fig 1: Crude birth prevalenceof all anomalies and folic acid preventable anomalies across the registries*

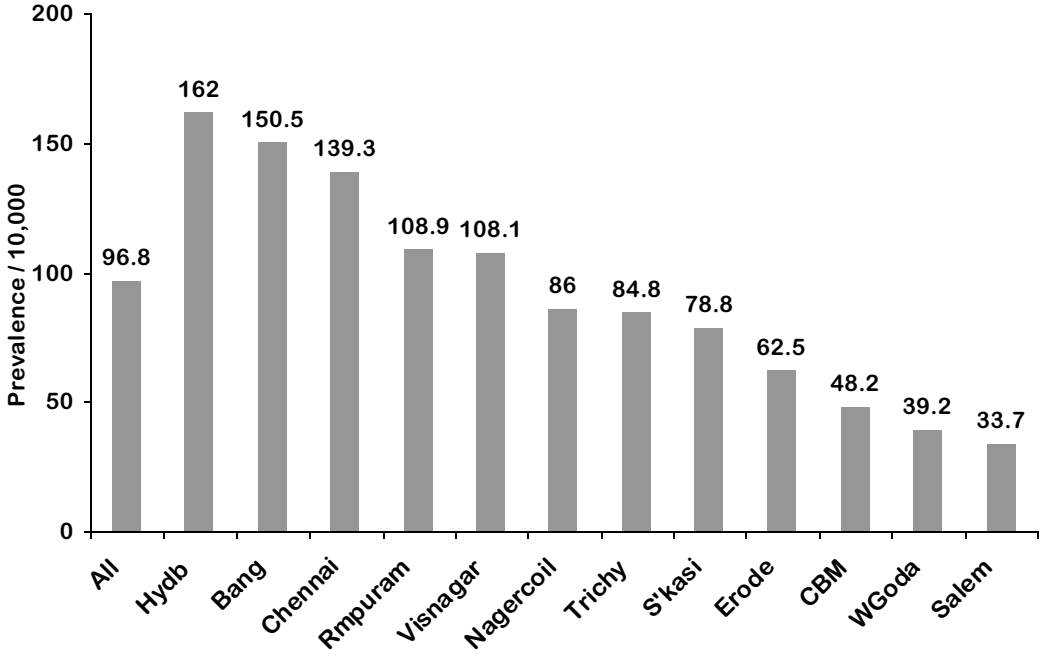
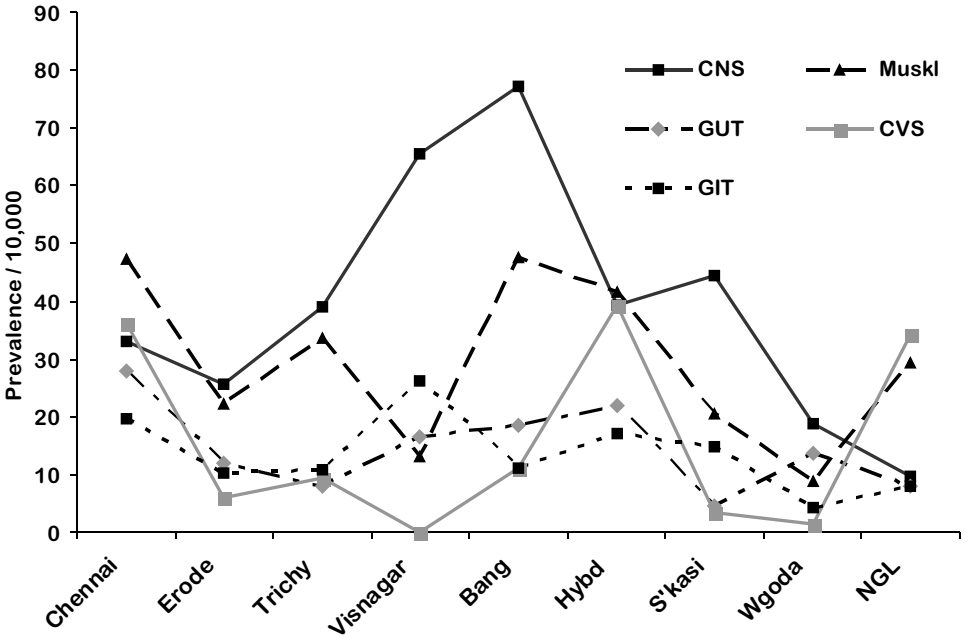



*Fig 2: Crude prevalance of selected system anomalies across all registries*




***Dr.Thangavel while concluding his presentation summarized the analysis as per Geographic variations***

★ Prevalence of NTD was more in Vis Nagar followed by Bangalore & Salem.
★ VSD was found more in Chennai, Vis Nagar & Trichy.
★ CLCP was reported more in Sivakasi & Bangalore
★ CTEV was reported high in Bangalore, Chennai & Hyderabad
★ Abdominal wall defects were common in Salem & Hyderabad
★ Limb reduction defects were found more in Salem & Trichy
★ Anorectal anomalies were reported more in Sivakasi & Visnagar
★ Cystic Renal Dysplasia was high in Salem, Chennai & Bangalore.
★ Renal agenesis was reported more in Chennai

*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)** | **228** | **32.8** |
| A01 Anencephaly (Q00.0) **(Incl. Acrania, Exencephaly, Iniencephaly)** | 84 | 12.1 |
| A02 Encephalocele (Q01.0 – Q01.9) **(Incl. Frontal & Occipital Encephalocele/ Meningocele)** | 14 | 2.0 |
| **A03 Microcephaly (Q02)** | 5 | 0.7 |
| A04 Congenital Hydrocephalus without Spina bifida (Q03.0 – Q03.9) **(Incl. Dandy – Walker malformation, Ventriculomegaly)** | 40 | 5.8 |
| A05 Spina bifida without anencephaly (Q05.0 – Q05.9) **(Incl. Meningocele, Meningomyelocele, Myelocele, Rachischisis, excluding Spina bifida occulta)** | 89 | 12.8 |
| A06 Holoprosencephaly (Q04.2) | 6 | 0.9 |
| A07 All other congenital malformations of brain, spinal cord & nervous system (Q04 & Q06) (Incl. Agenesis of corpus callosum, absence of nerves, cerebral cysts and cerebellar malformations, etc.) | 16 | 2.3 |

| Diagnostic Grouping | Number of cases | Prevalence / 10,000 |
|---|---|---|
| **B. Congenital Anomalies of Eye, Ear, Face & Neck (Q10 – Q18)** | **41** | **5.9** |
| B01 Anophthalmos / Microphthalmos / Macrophthalmos (Q11.0 – Q11.9) | 4 | 0.6 |
| B02 Absent external auditory meatus (Q16.1) | 2 | 0.3 |
| B03 Low set ears (Q17.4) | 19 | 2.7 |
| B04 All other congenital anomalies of Eye, Ear, Face & Neck (Q10 – Q18) | 22 | 3.2 |

| Diagnostic Grouping | Number of cases | Prevalence / 10,000 |
|---|---|---|
| **C. Congenital Anomalies of the Circulatory System (Q20 – Q28)** | **120** | **17.3** |
| C01 Common Truncus / Persistent Truncus arteriosus (Q20.0) | 3 | 0.4 |
| C02 Double outlet right ventricle (Q20.1) | 6 | 0.9 |
| C03 Transposed Great vessels (Q20.3) | 7 | 1.0 |
| C04 Ventricular Septal Defect (Q21.0) | 41 | 5.9 |
| C05 Atrial Septal Defect / Patent or persistent foramen ovale (Q21.1) | 32 | 5.2 |
| C06 Atrioventricular septal defect / Endocardial Cushion Defect / Ostium primum (Q21.2) | 5 | 0.7 |
| C07 Tetrology of Fallot (Q21.3) | 7 | 1.0 |
| C08 Pulmonary valve Atresia (Q22.0) | 0 | 0.0 |
| C09 Ebstein's anomaly (Q22.5) | 1 | 0.1 |
| C10 Hypoplastic right heart syndrome (Q22.6) | 1 | 0.1 |
| C11 Other tricuspid valve abnormalities (Q22.8) | 0 | 0.0 |
| C12 Bicuspid aortic valve (Q23.1) | 0 | 0.0 |
| C13 Hypoplastic left heart syndrome (Q23.4) | 11 | 1.6 |
| C14 Dextrocardia (Q24.0) | 1 | 0.1 |

| | | |
|---|---|---|
| C15 Patent ductus arteriosus (Q25.0) | 17 | 2.4 |
| C16 Anomalies of arch of Aorta (Q25.1 & 25.4) | 6 | 0.9 |
| C17 Anomalies of pulmonary artery (Q25.5 – 25.7) | 8 | 1.2 |
| C18 Persistent left superior vena cava (Q26.1) | 2 | 0.3 |
| C19 Single umbilical artery (Q27.0) | 21 | 3.0 |
| C20 Other specified and unspecified congenital heart anomalies (Q20.2, Q20.8, Q22.4, Q23.0, Q23.2, Q24.8, Q24.9 & Q25.8, Q26.2) | 5 | 0.7 |

| Diagnostic Grouping | Number of cases | Prevalence / 10,000 |
|---|---|---|
| **D. Congenital anomalies of the Respiratory system (Q30 – Q34)** | **9** | **1.3** |
| D01 Congenital cystic adenomatoid malformation of lung (Q30.0) | 2 | 0.3 |
| D02 Absence / Malformation of nose (Q30.1 – Q30.9) | 3 | 0.4 |
| D03 Laryngeal atresia (Q31.8) | 0 | 0.0 |
| D04 Tracheal atresia (Q32.1) | 0 | 0.0 |
| D05 Agenesis of lung (Q33.6, Q33.8) | 4 | 0.6 |

| Diagnostic Grouping | Number of cases | Prevalence / 10,000 |
|---|---|---|
| **E. Congenital anomalies of the Gastrointestinal tract (Q35 – Q45)** | **96** | **13.8** |
| E01 Cleft palate (Q35.0 – Q35.9) | 4 | 0.6 |
| E02 Cleft lip (Q36.0 – Q36.9) | 6 | 0.9 |
| E03 Cleft palate & cleft lip (Q37.0 – Q37.9) | 28 | 4.0 |
| E04 High arched palate (Q38.5) | 3 | 0.4 |
| E05 Other congenital malformations of tongue and mouth (Q38.2, Q38.3) | 3 | 0.4 |
| E06 Atresia of esophagus without fistula (Q39.0) | 7 | 1.0 |
| E07 Tracheoesophageal fistula with atresia (Q39.1) | 3 | 0.4 |
| E08 Tracheoesophageal fistula without atresia (Q39.2) | 4 | 0.6 |
| E09 Gastric outlet obstruction (Q40.0) | 1 | 0.1 |
| E10 Tubular Stomach (Q40.2) | 2 | 0.3 |
| E11 Absence, atresia and stenosis of small intestine (Q41.0 – Q41.9) | 8 | 1.2 |
| E12 Imperforate anus (Q42.3) | 20 | 2.9 |
| E13 Other Congenital malformations of large intestines (Q42.1) | 3 | 0.4 |
| E14 Meckel's diverticulam (Q43.0) | 0 | 0.0 |
| E15 Anomalies of liver and gall bladder (Q44.0 – Q44.9) | 2 | 0.3 |
| E16 Abscent pancreas (Q45.0) | 2 | 0.3 |
| E17 Anovestibular fistula / Rectovestibular fistula (Q64.7) | 2 | 0.3 |
| E18 Other specified and unspecified gastriointestinal tract (Q43.0 – 43.9) | 7 | 1.0 |

| Diagnostic Grouping | Number of cases | Prevalence / 10,000 |
|---|---|---|
| **F. Congenital Anomalies of the Genital and Urinary Systems (Q50 – Q64)** | **113** | **16.3** |
| F01 Congenital malformation female genital organs (Q50.0 – Q52.9) | 5 | 0.7 |
| F02 Undescended testis (Q53.0 – Q53.9) | 6 | 0.9 |
| F03 Hypospadias (Q54.0 – Q54.9) | 18 | 2.6 |
| F04 Other congenital malformations of male genital organs (Q55.0 – Q55.9) | 7 | 1.0 |
| F05 Indeterminate sex (Q56.4) | 15 | 2.2 |
| F06 Renal agenesis (Q60.0 – Q60.6) | 11 | 1.5 |
| F07 Cystic kidney disease (Q61.0 – Q61.9)<br>(Incl. Infantile or Adult polycystic kidney and Multicystic dysplasia) | 19 | 1.3 |
| F08 Congenital hydronephrosis (Q62.0) | 14 | 2.0 |
| F09 Pelviureteric junction obstruction (Q62.1) | 1 | 0.1 |
| F10 Other ureter anomaly (Q62.4 – Q62.8) | 3 | 0.4 |
| F11 Other congenital malformations of kidney (Q63.0 - Q63.9)<br>(Incl. Fused / Horseshoe kidney) | 9 | 1.3 |
| F12 Ectopia vesicae / Bladder exstrophy (Q64.1) | 3 | 0.4 |
| F13 Congenital posterior urethral valve (Q64.2) | 11 | 1.5 |
| F14 Other congenital malformations of bladder & urethra (Q64.3, Q64.8) | 6 | 0.9 |

| Diagnostic Grouping | Number of cases | Prevalence / 10,000 |
|---|---|---|
| **G. Congenital Anomalies of the Musculoskeletal System (Q65 – Q79)** | **220** | **31.6** |
| G01 Congenital dislocation of hip (Q65.0) | 1 | 0.1 |
| G02 Talipes equinovarus (Q66.0) | 60 | 8.6 |
| G03 Other Congenital malformations of feet (Q66.1- Q66.9)<br>(Incl. Rocker bottom foot) | 7 | 1.0 |
| G04 Congenital Musculoskeletal deformities of head, face, spine & chest (Q67.0 – Q67.9)<br>Incl. Dysmorphic face (Q67.0) | 16 | 2.3 |
| G05 Congenital deformities of knee (Q68.2)<br>Genu recurvatum | 1 | 0.1 |
| G06 Polydactyly (Q69.0 – Q69.9) | 20 | 2.9 |
| G07 Syndactyly and polysyndactyly (Q70.0 – Q70.9) | 11 | 1.5 |
| G08 Upper limbs - reduction defects / shortening (Q71.0 – Q71.9) | 13 | 1.9 |
| G09 Lower limbs - reduction defects / shortening (Q72.0- Q72.9) | 10 | 1.4 |
| G10 Unspecified limbs - reduction defects / shortening (Q73.0 – Q73.8) | 7 | 1.0 |
| G11 Arthrogryposis (Q74.3) | 4 | 0.6 |
| G12 Other congenital malformations of limbs (Q74.8 & Q74.9) | 7 | 1.0 |
| G13 Hypertelorism (Q75.2) | 9 | 1.3 |
| G14 Other congenital malformations of skull & face bones (Q75.0– 75.9) | 18 | 2.6 |
| G15 Spina bifida occulta (Q76.9) | 0 | 0.0 |

| | | |
|---|---|---|
| G16 Other congenital malformations of bony thorax and spine (Q76.0 – Q76.8) (Incl. Scoliosis, Hemivertebre etc) | 35 | 5.0 |
| G17 Osteochondrodysplasia with defects of growth of tubular bones & spine (Q77.0 – Q77.9) | 24 | 3.5 |
| G18 Osteogenesis imperfecta (Q78.0) | 2 | 0.3 |
| G19 Diaphragmatic Hernia (Q79.0) | 15 | 2.2 |
| G20 Absence / Eventration of diaphragm (Q79.1) | 1 | 0.1 |
| G21 Exomphalos / Omphalocele (Q79.2) | 12 | 1.7 |
| G22 Gastroschisis (Q79.3) | 9 | 1.3 |
| G23 Thanatophoric Dysplasia (Q77.1) | 4 | 0.6 |
| G24 Other congenital malformations of abdominal wall ( Q79.5, Q79.8) (Incl. Limb body wall complex, Cloacal anomaly) | 5 | 0.7 |
| G25 Other specified and unspecified congenital malformations of musculoskeletal system | 4 | 0.6 |

| Diagnostic Grouping | Number of cases | Prevalence / 10,000 |
|---|---|---|
| **H. Other Congenital Anomalies (Q80 – Q86 & Q89)** | **10** | **1.4** |
| H01 Icthyosis (Q80.8) | 1 | 0.1 |
| H02 Congenital hypothyroidism (Q89.2) | 0 | 0.0 |
| H03 Simian crease (82.8) | 5 | 0.7 |
| H04 All other congenital malformations not elsewhere classified (Q82.3, Q89.4, Q89.9) | 5 | 0.7 |

| Diagnostic Grouping | Number of cases | Prevalence / 10,000 |
|---|---|---|
| **I. Multisystem Anomalies / Syndromes** | **15** | **1.2** |
| I01 Meckel Gruber Syndrome (Q61.9) | 0 | 0.1 |
| I02 Pierre Robin syndrome (Q87.0) | 3 | 0.4 |
| I03 Sirenomelia sequence (Q87.2) | 2 | 0.3 |
| I04 VACTREL (Q87.2) | 1 | 0.1 |
| I05 Frydman Cohen Syndrome (Q87.8) | 2 | 0.3 |
| I06 Other Syndromes (Q75.1, Q87.3, Q87.5, Q87.9) | 3 | 0.4 |

| Diagnostic Grouping | Number of cases | Prevalence / 10,000 |
|---|---|---|
| **J. Chromosomal Anomalies (Q90)** | **23** | **3.3** |
| J01 Down's Syndrome (Q90.0 – Q90.9) | 15 | 2.2 |
| J02 Edwards' Syndrome (Q91.3) | 4 | 0.6 |
| J03 Pautau's Syndrome (Q91.7) | 1 | 0.1 |
| J04 Other Syndrome (Q96.0, Q99.1, Q99.8) | 3 | 0.4 |

**WELL DONE PUNE BDR ! KEEP IT UP !!**

Pune BDR recently joined hands with BDRI. Deenanath Mangeshkar Hospital, Pune has been steering the activities of the registry under the guidance of Dr. Koumudi Godbole, Dr Pratibha Nanak Kulkarni & the project coordinator Ms. Vidyullata Deshpande.

They have conducted 2 meetings which were well attended by the members.

***The proceedings of the meetings are as follow:***

**I BDR Meeting - 25th March 2006**

3 cases were presented. Dr. Anand Railkar of Railkar Hospital, Loni-Kalbhor presented a case of oesophageal atresia, imperforate anus, absent radius & abnormal posteriorly rotated low set ears. The differential diagnoses discussed for this were Townes - Brocks Syndrome, VACTERL association & Chromosomal Anomaly (Karyotype not available).The other 2 cases presented were Complex Congenital Heart Disease by Jehangir Hospital, Pune. The role of fetal echo in these cases were discussed. Dr. Jyotisha singh from Deenanath Mangeshkar Hospital spoke on Fetal MRI.

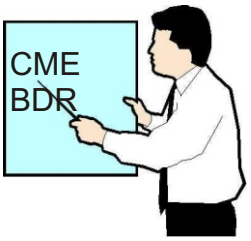
**II BDR Meeting-20th May 2006**

During the second meeting 2 cases were presented. Dr. Manisha Doiphode from Swaminathan hospital, Pimpri, Pune, presented a case which was prenatally diagnosed to be Tuberous Sclerosis . Dr Usha Pratap from Deenanath Hospital Performed fetal ECHO for this case & found multiple cardiac tumors in the late III trimester. Postnatally, the baby was diagnosed to have cortical tubers as well and the baby was shifted to NICU for cardiac failure and related problems. There are no skin lesions so far. The baby is being followed up.

The other case presentation was from Deenanath Hospital where a pregnancy was terminated at 16 weeks as the fetus had Sacrococcygeal Terotoma and was sent for autopsy. Postmortem study revealed Adrenal Cytopathy which is commonly found in fetuses with Beckwith - Weidemann syndrome which is known to have a higher incidence of embryonal tumors. Though this syndrome was not ruled out in the fetus, autopsy facilitated prenatal counseling of the parents concerned.

Dr. Koumudi Godbole, Consulting Geneticist, Deenanath Mangeshkar Hospital also presented a summary of total cases & malformations reported so far and discussed the logistics of Pune BDR with the members at the meeting.

BDRI invites other member registries to follow Pune BDR and conduct CME in their regions. The proceedings may be sent across for publishing in the News Letter. This two way communication would facilitate enrichment of knowledge on Birth defects.






**BDRI & Rotary Madras Metro Partnership Program for Birth Defects Prevention**


# CLINICAL ULTRASONOGRAPHY IN PRACTICE

# CUSP 2006

## New Frontiers - New Challenges

**Kamaraj Memorial Auditorium, Chennai**

**22, 23 & 24 September 2006**

This news letter is available online at http://www.mediscansystems.org. Issued four times in a year - January, April, July and October. Published by Fetal Care Research Founation, 203, Avvai Shanmugam Salai, Royapettah, Chennai - 600 014. For Private circulation. Printed at The Print Shoppe (Print Supplies), Ayanavaram, Chennai - 600 023.