B D R News

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

Volume 8 Issue 2: April 2008

Proceedings of the birth defects registry meeting held on 09.05.2008

The second BDR meeting of the current year was held on 09/05/08 at the premises of Mediscan Systems in Chennai. Dr.Indrani Suresh extended a warm welcomed to the audience who had gathered in appreciable number braving the scorching summer heat. She stressed the need to have a meeting for annual statistical reporting of birth defects and said that unless we arrive at a conclusive picture of birth defects in our population, we cannot evolve strategies to prevent them. Incidentally the other two programs that followed the annual report, emphasized the effective awareness campaign towards prevention of the two most common birth defects Neural Tube Defect (NTD) & Down syndrome. She welcomed the guest speaker Dr.Rekha Ramachandran, President, Down syndrome Association of Tamilnadu who is widely known for her yeoman services to children affected with Down syndrome. She was there to show the documentary she has produced for a project conceived towards prevention of Down syndrome.

During her brief address, she appealed to the obstetricians who were the primary consultants of expectant mothers to make use of screening /diagnostic tests antenatally available so that less number of children would be born with this problem in future. She wished that God had not created this disorder when she counseled parents with affected children. It is a long lasting agony for the parents to bring up & protect these children once they are born. She wondered that despite the availability of prenatal diagnosis for more than a decade, she encounterd quite a number of cases every day. Lack of awareness among the public & the inability of the masses to undertake the tests suggested due the high cost involved, motivated her to conceive a project to work towards prevention. She was thankful to the National Rural Health Mission, Government of Tamilnadu for sponsoring the cost of confirmatory procedure (ChorionicVilli Sampling/Amniocentesis) for those cases identified to be screen positive for Down syndrome. She also thanked Dr. Suresh for being a part of the project by agreeing to do the confirmatory procedure for mothers at her request. Her mass awareness campaign she hoped would bring down the incidence so that it minimized the burden of postnatal care for those involved. The movie titled “Today's Dreams, Tomorrow's Hopes” portrayed the touching & thought provoking efforts of the people behind its creation & it is sure to fetch a positive response both from the public & the medical fraternity across the state.

Dr. S. Suresh while presenting the strategies he has planned towards prevention of Neural Tube Defects in the population, lauded Dr. Rekha's contributions to the largest support group in the country. He appreciated the Government for collaborating with a private organization to support a cause of this kind. He also recollected the first time way back in 1997 when he started the II trimester screening, there were hardly any takers. But he continued to offer the test for its benefit. Since now the first trimester screening facility has a detection rate of 90% compared to 60% in II trimester screening, he appealed to medical community to make use of it for all pregnant mothers and not only for selective high risk cases. Any screening test is population based he added. He was happy that now the procedure cost is funded, more number of deserving cases could be benefitted by Dr. Rekha's project in the state.

Dr. Suresh addressed the need for urgent actions to prevent open neural tube defects. With the advent of Ultrasound since 20 years, it is pathetic to see cases even today with NTD in late last trimester. It is the most commonly missed out anomaly leading to severe postnatal morbidity& long-term disability in children. It also poses a great economic & emotional burden on the family & society. As the age old proverb goes- Prevention is better than cure, especially when there are easy preventive measures available!

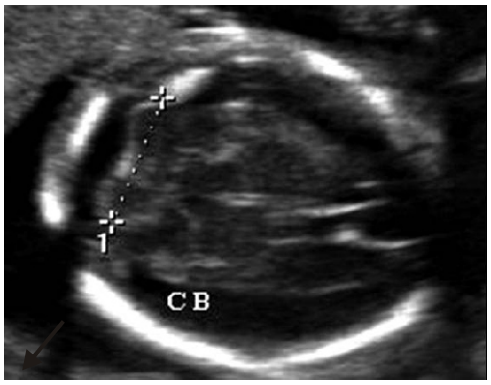
He grouped the strategies into primary & secondary preventive measures. **Primary prevention** comprises of

1. **C**reating public awareness about peri conceptional folic acid &
2. **R**enaming Folic acid as THE ENGAGEMENT PILL so that it attracts more attention from the public!

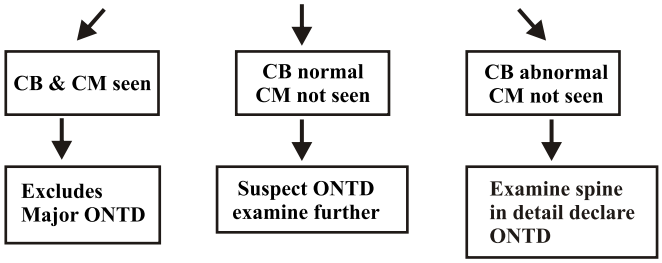
He even coined slogans for campaigns such as “A pill a day will keep NTD away/at bay “etc to make it sound more interesting! While explaining the strategies for **Secondary prevention** he requested the Sonologists to get tuned to detecting open NTD as early as 14 weeks of gestational age. One should be aware of the simple pattern recognition of NTD and **l**earn by comparing standard USG pictures with the abnormal ones. Repeated audit of the pictures is a must to get reassured of one's diagnostic skills. All II Trimester scans should have a**t**least one documented picture of posterior fossa. He gave a simple checklist as given overleaf to diagnose or rule out NTD.

He also emphasized the need for periconceptional folic acid in high-risk mothers having Diabetes / Epilepsy. They need a special focus on the fetal spine & it is better to go in for second opinion once a problem is suspected. He informed that the ongoing ICMR- NTD project would throw light on the need to administer Vitamin B_{12} along with Folic acid to prevent NTD. Dr. Suresh concluded saying that he has a realistic goal of reducing incidence of NTD by 50% in the next 2 years & by 80% in the next 5 years!

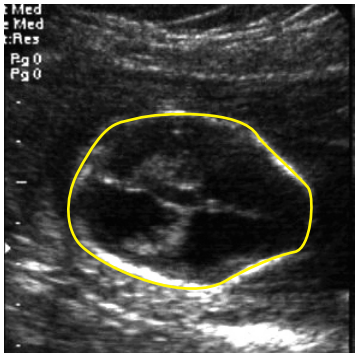
Imaging posterior fossa



CB - Cerebellum, CM - Cisterna magna, ONTD - Open Neural Tube Defects

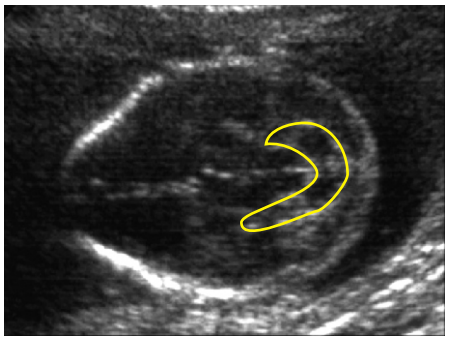


Cranial USG findings - NTD



Lemon sign

Banana sign



Annual Statistics Report of Birth Defects - Year 2007

Ms. V. Jayanthi, Statistician , MediScan

Introduction

The prevalence of congenital malformation worldwide is about 2 -3%. In India, though nation wide prevalence is not known, few small hospital-based studies indicate that it would be high. However, there is no systematic surveillance for birth defects across the nation. Having understood the lacunae in this area, Fetal Care Research Foundation (FCRF) established the Birth Defects Registry of India (BDRI) in 2001. Given below is the seventh successive annual statistical report of BDRI.

It presents the prevalence of birth defects estimated from 18 regional registries; viz. Chennai, Erode, Trichy, Lalgudi, Madurai, Nagercoil, Ramanathapuram, Dindigul, and Sivakasi in TamilNadu, Hyderabad and West Godavari in Andhra Pradesh, Bangalore and Belgaum in Karnataka, Mumbai, Pune, Akola, Jalgaon, and Aurangabad in Maharashtra and Vis Nagar of Mehsana district in Gujarat. Data from Trichy and Lalgudi were combined as they represent the same geographic area (Administrative district).

Programme description

BDRI is a hospital-based descriptive surveillance programme, 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 being stored in the database. The diagnostic terms are then coded according to ICD10 version. Finally statistical analysis is done and the yearly annual report is presented to the members.

Results

During 2007 there were 1,30,565 births reported from the member registries, of which 97.8% were live born. (Table 1). There were 1252 cases with birth defect(s). The over all crude birth prevalence is 95.9/10,000. High prevalence was reported from Hyderabad and low prevalence reported from West Godavari and Ramanathapuram (Fig1). CNS anomalies were found to be high (Fig. 2) across all registries, except Hyderabad and Nagercoil, Tables 2A to J show the detailed anomaly specific and system specific crude birth prevalence across all registries.

Limitations of the data

Though overall crude birth prevalence is 95.9 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 23% of total births are covered by the programme 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 were used, only a few cases from neonatologists and pediatricians were reported. Although the estimated prevalence may not reflect the true population prevalence in the regions, it shows the pattern and type of congenital malformations most likely to occur in those areas.

Table 1: Frequency of birth categories.

Categories	N	%
Live birth	127677	97.8
Intrauterine fetal death /still birth	2534	1.9
MTP for anomaly	354	0.3
Total births	130565	100

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

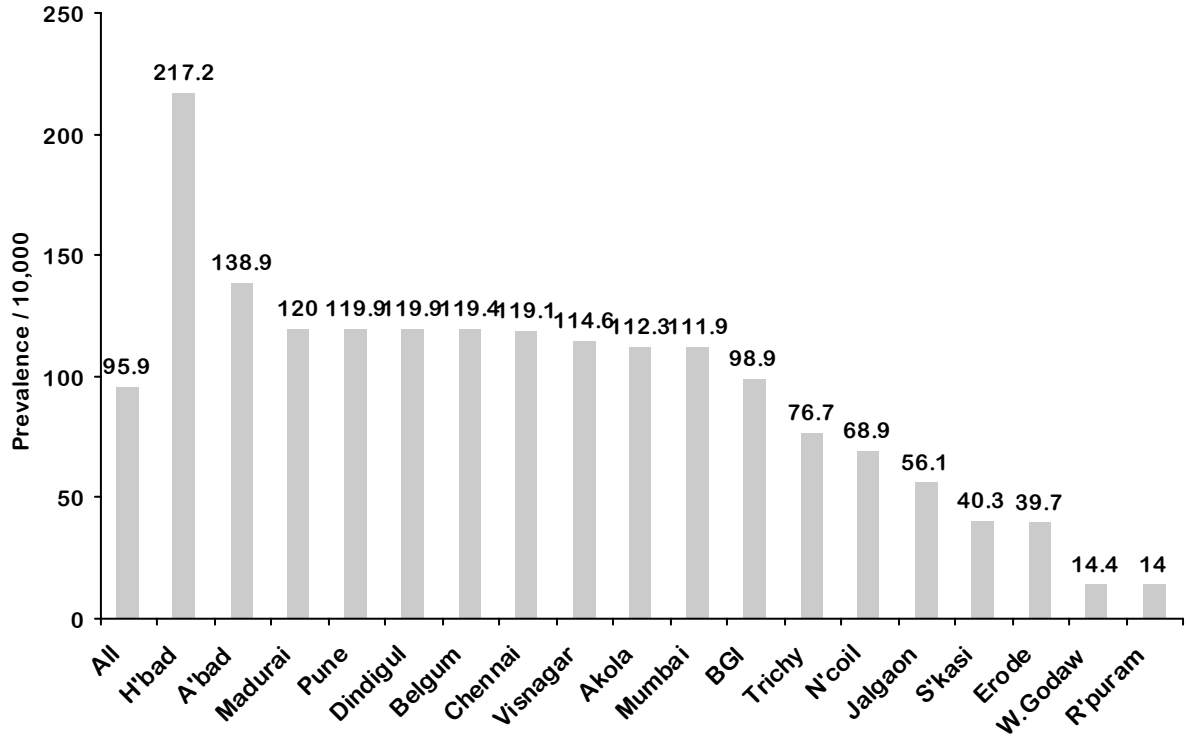


Fig 2: Crude birth prevalence of selected system anomalies across all registries

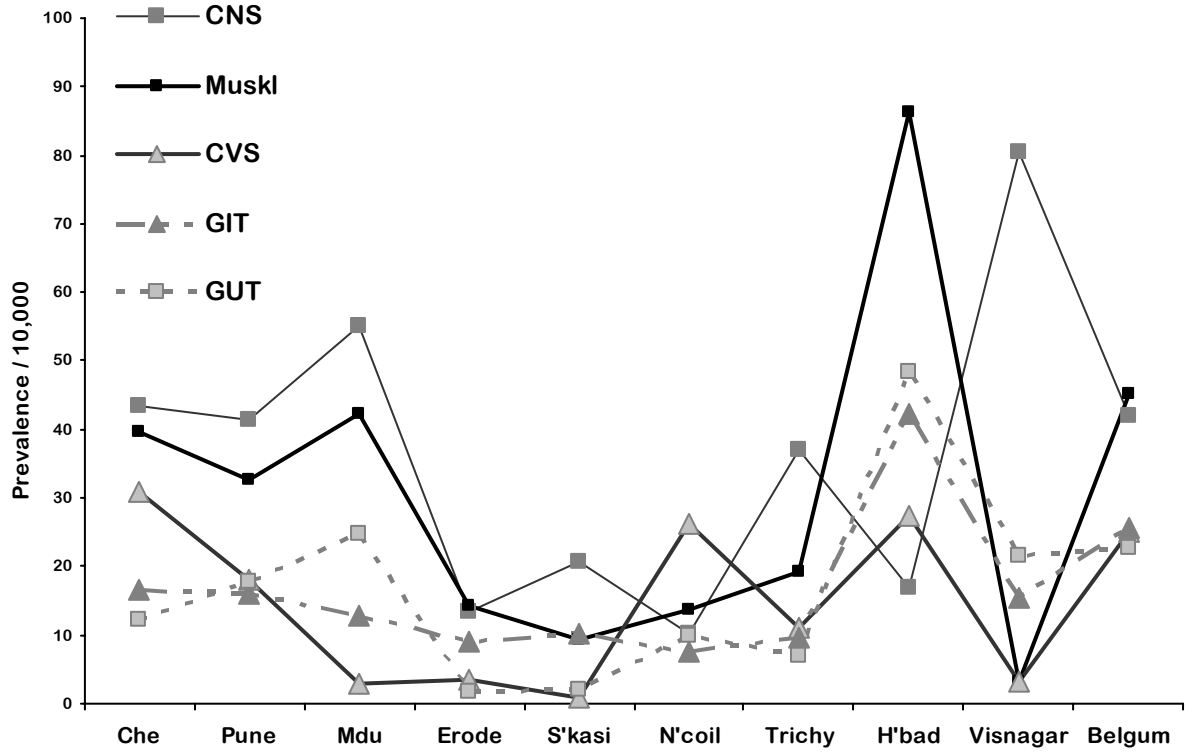


Table 2A-J: Anomaly specific and system specific crude birth prevalence of congenital malformations across all registries

Diagnostic Grouping	Number of cases	Prevalence / 10,000
A. Congenital Anomalies of the Central Nervous System (Q00 – Q07)	**460**	**35.2**
A01 Anencephaly (Q00.0) **(Incl. Acrania, Exencephaly, Iniencephaly)**	159	12.2
A02 Encephalocele (Q01.0 – Q01.9) **(Incl. Frontal & Occipital Encephalocele/ Meningocele)**	50	3.8
A03 Microcephaly (Q02)	9	0.7
A04 Congenital Hydrocephalus without Spina bifida (Q03.0 – Q03.9) **(Incl. Dandy – Walker malformation, Ventriculomegaly)**	88	6.7
A05 Spina bifida without anencephaly (Q05.0 – Q05.9) **(Incl. Meningocele, Meningomyelocele, Myelocele, Rachischisis, excluding Spina bifida occulta)**	136	10.4
A06 Holoprosencephaly (Q04.2)	8	0.6
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.)	15	1.1

Diagnostic Grouping	Number of cases	Prevalence / 10,000
B. Congenital Anomalies of Eye, Ear, Face & Neck (Q10 – Q18)	**99**	**7.6**
B01 Anophthalmos / Microphthalmos / Macrophthalmos (Q11.0 – Q11.9)	11	0.8
B02 Absent external auditory meatus (Q16.1)	9	0.7
B03 Low set ears (Q17.4)	49	3.8
B04 All other congenital anomalies of Eye, Ear, Face & Neck (Q10 – Q18)	37	2.8

Diagnostic Grouping	Number of cases	Prevalence / 10,000
C. Congenital Anomalies of the Circulatory System (Q20 – Q28)	**197**	**15.1**
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)	4	0.3
C04 Ventricular Septal Defect (Q21.0)	38	2.9
C05 Atrial Septal Defect / Patent or persistent foramen ovale (Q21.1)	21	1.6
C06 Atrioventricular septal defect / Endocardial Cushion Defect / Ostium primum (Q21.2)	15	1.1
C07 Tetrology of Fallot (Q21.3)	1	0.1
C08 Pulmonary valve Atresia (Q22.0)	0	0
C09 Ebstein's anomaly (Q22.5)	2	0.2
C10 Hypoplastic right heart syndrome (Q22.6)	9	0.7
C11 Other tricuspid valve abnormalities (Q22.8)	0	0
C12 Bicuspid aortic valve (Q23.1)	1	0.1
C13 Hypoplastic left heart syndrome (Q23.4)	16	1.2

C14 Dextrocardia (Q24.0)	2	0.2
C15 Patent ductus arteriosus (Q25.0)	25	1.9
C16 Anomalies of arch of Aorta (Q25.1 & 25.4)	8	0.6
C17 Anomalies of pulmonary artery (Q25.5 – 25.7)	5	0.4
C18 Persistent left superior vena cava (Q26.1)	3	0.2
C19 Single umbilical artery (Q27.0)	32	2.4
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)	22	1.7

Diagnostic Grouping	Number of cases	Prevalence / 10,000
D. Congenital anomalies of the Respiratory system (Q30 – Q34)	**9**	**0.7**
D01 Congenital cystic adenomatoid malformation of lung (Q30.0)	7	0.5
D02 Absence / Malformation of nose (Q30.1 – Q30.9)	3	0.2
D03 Laryngeal atresia (Q31.8)	1	0.1
D04 Tracheal atresia (Q32.1)	0	0
D05 Agenesis of lung (Q33.6, Q33.8)	1	0.1
D06 Other Respiratory anomalies	1	0.1

Diagnostic Grouping	Number of cases	Prevalence / 10,000
E. Congenital anomalies of the Gastrointestinal tract (Q35 – Q45)	**182**	**13.9**
E01 Cleft palate (Q35.0 – Q35.9)	21	1.6
E02 Cleft lip (Q36.0 – Q36.9)	17	1.3
E03 Cleft palate & cleft lip (Q37.0 – Q37.9)	53	4.0
E04 High arched palate (Q38.5)	15	1.1
E05 Other congenital malformations of tongue and mouth (Q38.2, Q38.3)	1	0.1
E06 Atresia of esophagus without fistula (Q39.0)	16	1.2
E07 Tracheoesophageal fistula with atresia (Q39.1)	0	0
E08 Tracheoesophageal fistula without atresia (Q39.2)	12	0.9
E09 Gastric outlet obstruction (Q40.0)	0	0
E10 Tubular Stomach (Q40.2)	0	0
E11 Absence, atresia and stenosis of small intestine (Q41.0 – Q41.9)	15	1.1
E12 Imperforate anus (Q42.3)	31	2.4
E13 Other Congenital malformations of large intestines (Q42.1)	1	0.1
E14 Meckel's diverticulam (Q43.0)	0	0
E15 Anomalies of liver and gall bladder (Q44.0 – Q44.9)	0	0
E16 Abscent pancreas (Q45.0)	1	0.1
E17 Anovestibular fistula / Rectovestibular fistula (Q64.7)	1	0.1
E18 Other specified and unspecified gastriointestinal tract (Q43.1 – 43.9)	9	0.7

Diagnostic Grouping	Number of cases	Prevalence / 10,000
F. Congenital Anomalies of the Genital and Urinary Systems (Q50 – Q64)	**166**	**12.7**
F01 Congenital malformation female genital organs (Q50.0 – Q52.9)	3	0.2
F02 Undescended testis (Q53.0 – Q53.9)	20	1.5
F03 Hypospadias (Q54.0 – Q54.9)	24	1.8
F04 Other congenital malformations of male genital organs (Q55.0 – Q55.9)	8	0.6
F05 Indeterminate sex (Q56.4)	22	1.7
F06 Renal agenesis (Q60.0 – Q60.6)	26	2.0
F07 Cystic kidney disease (Q61.0 – Q61.9) (Incl. Infantile or Adult polycystic kidney and Multicystic dysplasia)	43	3.3
F08 Congenital hydronephrosis (Q62.0)	16	1.2
F09 Pelviureteric junction obstruction (Q62.1)	7	0.5
F10 Other ureter anomaly (Q62.4 – Q62.8)	0	0
F11 Other congenital malformations of kidney (Q63.0 - Q63.9) (Incl. Fused / Horseshoe kidney)	3	0.2
F12 Ectopia vesicae / Bladder exstrophy (Q64.1)	1	0.1
F13 Congenital posterior urethral valve (Q64.2)	5	0.4
F14 Other congenital malformations of bladder & urethra (Q64.3, Q64.8)	6	0.5

Diagnostic Grouping	Number of cases	Prevalence / 10,000
G. Congenital Anomalies of the Musculoskeletal System (Q65 – Q79)	**374**	**28.6**
G01 Congenital dislocation of hip (Q65.0, Q65.1)	3	0.2
G02 Talipes equinovarus (Q66.0)	107	8.2
G03 Other Congenital malformations of feet (Q66.1- Q66.9) (Incl. Rocker bottom foot)	6	0.5
G04 Congenital Musculoskeletal deformities of head, face, spine & chest (Q67.0 – Q67.9) Incl. Dysmorphic face (Q67.0)	34	2.6
G05 Congenital deformities of knee (Q68.2) Genu recurvatum	6	0.5
G06 Polydactyly (Q69.0 – Q69.9)	36	2.8
G07 Syndactyly and polysyndactyly (Q70.0 – Q70.9)	17	1.3
G08 Upper limbs - reduction defects / shortening (Q71.0 – Q71.9)	22	1.7
G09 Lower limbs - reduction defects / shortening (Q72.0- Q72.9)	5	0.4
G10 Unspecified limbs - reduction defects / shortening (Q73.0 – Q73.8)	6	0.5
G11 Arthrogryposis (Q74.3)	12	0.9
G12 Other congenital malformations of limbs (Q74.8 & Q74.9)	5	0.4
G13 Hypertelorism (Q75.2)	22	1.7
G14 Other congenital malformations of skull & face bones (Q75.0– 75.9)	41	3.1
G15 Spina bifida occulta (Q76.9)	1	0.1

G16 Other congenital malformations of bony thorax and spine (Q76.0 – Q76.8) (Incl. Scoliosis, Hemivertebre etc)	30	2.3
G17 Osteochondrodysplasia with defects of growth of tubular bones & spine (Q77.0 – Q77.9)	29	2.2
G18 Osteogenesis imperfecta (Q78.0)	4	0.3
G19 Diaphragmatic Hernia (Q79.0)	25	1.9
G20 Absence / Eventration of diaphragm (Q79.1)	1	0.1
G21 Exomphalos / Omphalocele (Q79.2)	29	2.2
G22 Gastroschisis (Q79.3)	6	0.5
G23 Thanatophoric Dysplasia (Q77.1)	3	0.2
G24 Other congenital malformations of abdominal wall (Q79.5, Q79.6, Q79.8) (Incl. Limb body wall complex, Cloacal anomaly)	11	0.8
G25 Other specified and unspecified congenital malformations of musculoskeletal system	2	0.2

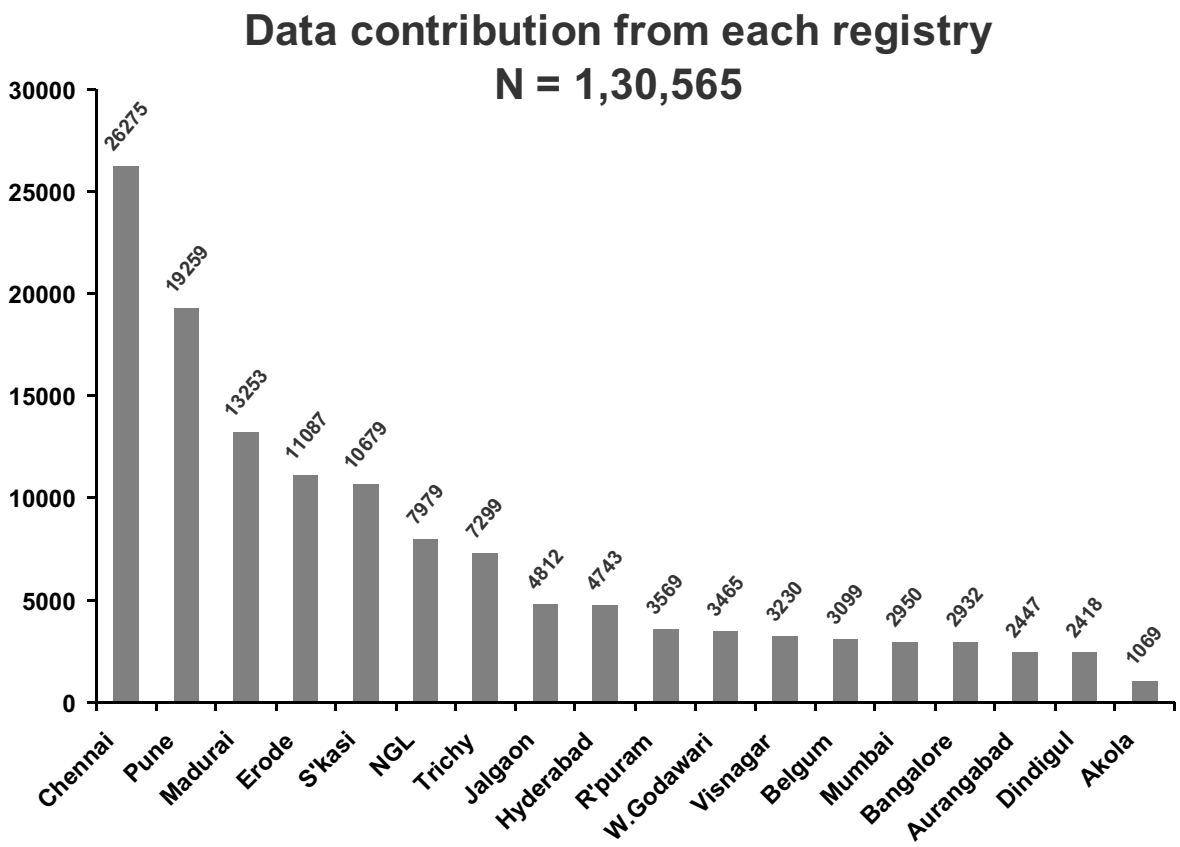
Diagnostic Grouping	Number of cases	Prevalence / 10,000
H. Other Congenital Anomalies (Q80 – Q86 & Q89)	**14**	**1.1**
H01 Icthyosis (Q80.8)	1	0.1
H02 Simian crease (82.8)	8	0.6
H03 All other congenital malformations not elsewhere classified (Q81.8, Q82.3, Q84.8, Q89.4, Q89.9)	5	0.4

Diagnostic Grouping	Number of cases	Prevalence / 10,000
I. Multisystem Anomalies / Syndromes	**18**	**1.4**
I01 Meckel Gruber Syndrome (Q61.9)	5	0.4
I02 Pierre Robin syndrome (Q87.0)	4	0.3
I03 Sirenomelia sequence (Q87.2)	4	0.3
I04 VACTREL (Q87.2)	0	0
I06 Other Syndromes (Q75.1, Q87.1, Q87.3, Q87.5, Q87.8, Q87.9)	5	0.4

Diagnostic Grouping	Number of cases	Prevalence / 10,000
J. Chromosomal Anomalies (Q90)	**19**	**1.5**
J01 Down's Syndrome (Q90.0 – Q90.9)	13	1.0
J02 Edwards' Syndrome (Q91.3)	4	0.3
J03 Pautau's Syndrome (Q91.7)	0	0
J04 Other Syndrome (Q96.0, Q98.4, Q99.1, Q99.8)	2	0.2

Salient features of the BDRI report for the year 2007

- Number of active registries contributing data increased from 1 in 2001 to 18 by 2007. Following table gives a glimpse of the data contributed by each registry



- Number of births analyzed increased from 14,161 to 1,30,565 over a period of 7 years.
- CNS anomalies stood at 26.4/ 10000 followed by CTEV and the least reported anomaly was renal agenesis.
- Anencephaly was the commonest reported CNS defect followed by Spina bifida & Hydrocephalus/ Dandy walker malformation.
- Among the Cardiac defects reported, Septal defects (37%) constituted the most followed by defects of the Great arteries & veins (20%)
- Visnagar BDR had the highest reporting of NTD as in the previous years followed by Madurai & Trichy BDR
- Musculoskeletal anomalies & Cardiac defects were reported high in Hyderabad when compared to CNS anomalies, probably due to practice of non termination policy by the Nodal centre which contributes the major data to the registry.
- CVS anomalies were reported high in Nagercoil & Hyderabad registries which may be due to more involvement of Paediatricians in these registries.
- Another conspicuous finding in the present report was the overall reduction in crude birth prevalence of birth defects when compared to previous years-i.e 131.3/10000 in 2001 & 95.9/10000 in 2007. This according to our Epidemiologist may be due to declining enthusiasm among the members over a period & non- reporting of minor defects in the long run. The BDRI team appeals to all members to revive their zeal & continue to contribute their valuable data until we reach our goal. Let us work towards expanding the BDR network further & contribute information with sustained fervor & enthusiasm for a national cause!

BDRI extends a hearty welcome to the nodal - Dr. Shah Maternity Nursing Home, and the participating members of Ahmedabad

BDRI

&

Rotary Madras Metro

Partnership Program
for Birth Defects Prevention

Dt 3230

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