BDR NEWS

The official newsletter of The Birth Defects Registry, Chennai

(Unit of Fetal Care Research Foundation)

Volume 1 **Issue 1, April 2001**

Fetal Care Research Foundation

In 1993, The Fetal Care Research Foundation (FCRF), a charitable trust was formed to do research in fetal malformations, prenatal diagnosis and fetal therapy. As an offshoot of FCRF, The Birth Defects Registry (BDR) was started in the year 1996. Over the last four years we have focussed on Down's syndrome and neural tube defects by establishing the maternal serum screening test apart from ultrasound evaluation. A registry for recording birth defects involving other centres in Chennai was initiated in January 2001.

What is BDR?

A Birth Defects Registry is a systematic mechanism for the collection, storage and reporting of congenital malformations.

Why is it needed?

About 2 - 4% of all pregnancies in India are likely to result in birth defects. Six percent of the population is affected by developmental disabilities. This alone accounts for 65 to 70 million people, (The Census 2001)which increases every year. Hence, there is a need for effective strategies to reduce the incidence of birth defects. The backbone of such strategies is an active birth defects registry. In order to be effective a model registry has to be created, which can be duplicated in various locations around the country, which will form the national birth defects registry.

Aims of the BDR

- To create awareness of the incidence and prevalence of birth defects in Chennai.
- To detect trends in the frequency of birth defects.
- To formulate and implement strategies to prevent occurrence of birth defects.
- To help medical personnel involved in antenatal care by providing statistical information about our own population, which will help in counseling.
- To serve as an impetus for further research in the field of fetal malformations and childhood disorders.
- Our final goal is to reduce the incidence of birth defects & help to plan effective strategies for disabled children by forming support groups etc.,

Defects to be monitored by Registry

1. Structural
2. Chromosomal
3. Metabolic
4. IUDs
5. Still borns
6. Abortions

Protocol for Data collection

This registry has an active data collection mechanism i.e. whenever a member is involved in the diagnosis or delivery of a fetus with birth/genetic defects, he/ she will report to the registry in the following manner:

- The details of the birth and the defects entered in a prescribed form provided by the registry.
- If the participant is not able to arrive at a diagnosis. One of the registry personnel will be available for help.

All members will be duly acknowledged in the periodical report, which will be published by the Registry.

Storing, Analysing and Reporting of Data:

- A Database is created for storing the records.
- To respect individual confidentiality personal information will not be entered in the data base.
- Separate files will be maintained for each member of the BDR.
- Members are requested to send their data once in two months, and the feedback regarding data collection for the same will be sent 15 days after receiving the data. Apart from the feedback a newsletter will be published once in 3 months and sent to the members.

Benefits of being a member of BDR:

- Easily retrievable data on birth defects available for members.
- Increase existing knowledge to identify and manage birth defects.
- Improve antenatal ultrasound skills.
- Entitled to take active Participation in ongoing research programs.
- To facilitate accurate counseling to the parents of malformed fetuses.

Proceedings of the first BDR meeting

The inaugural meeting of the Birth Defects Registry was held at Mediscan Systems, Chennai, on the 15th of March, 2001. Dr.Suresh, Director welcomed the gathering. The members assembled introduced themselves.

This was followed by a crisp introduction by Dr.Sujatha Jagadeesh, Dysmorphologist, of the Birth Defects Registry. She emphasized the need for the registry to create an understanding about the incidence and prevalence of Birth Defects in Chennai initially. "The ultimate goal is to reduce the incidence of birth defects in Chennai and to plan strategies for the best possible management of disabled children". This was followed by case presentation by Dr.Shivarajan, trainee in clinical dysmorphology to highlight the importance of karyotyping and perinatal autopsy in identifying the diagnosis and its role in risk prediction and management of subsequent pregnancies.

Mrs. Ranjini Parthasarathy, project coordinator explained the protocol for furnishing BDR statistics. It was decided that general delivery statistics will be collected from member hospitals once in two months for convenience. Members were requested to fill in the details about congenital anomalies detected, in BDR forms during that period. They were instructed to use the BDR code number assigned to their hospital in all their communications and forms. If there was any difficulty in dispatching the data, BDR personnel offered to collect the same. Feed back about the data collected would be given to them within 15 days.

Dr.G.Thangavel, Epidemiologist spoke about the methodology of storing and analysing the data collected. It was decided that the members will meet once in 3 months to sort out issues on BDR. After streamlining the process of data collection, members were requested to induct atleast one other member to strengthen the registry. A number of queries raised by the members regarding the timing and mode of sending the fetus for autopsy, samples for karyotyping and the charges incurred for the same were clarified. For members of the BDR, Mediscan systems offered,

- Obstetric scans & genetic counseling free of cost for all poor patients who are reported to the registry.
- In special situations, clinical photographs and examinations could be done.

The feasibility of taking simple photographs of the babies born with defects by the members for documentation was discussed. Dr.S.Suresh proposed vote of thanks and appreciated the good gesture and cooperation of all members in launching the birth defects registry.

Case reports (Presented by Dr.Shivarajan)

Case:1

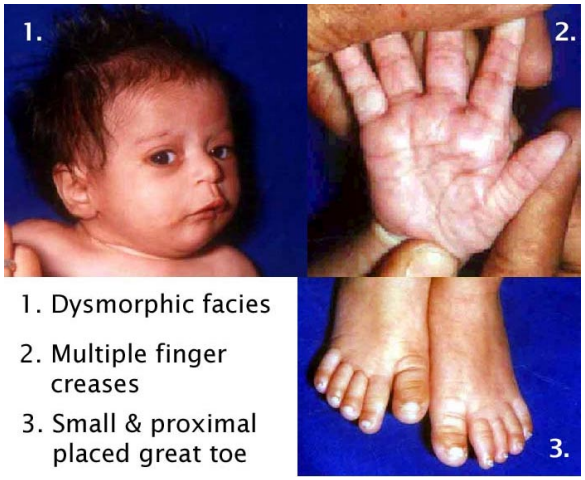
A 26 year old, gravida 4 brought her only living child for a karyotype and echocardiogram to our institution. Her first child was a male delivered at term which died immediately after birth. Documented features of the baby included cleft lip and palate, micropenis, undescended testes, hypospadias. No history of consanguinity. No other investigations were done. Her second pregnancy ended in a spontaneous abortion at 45 days.

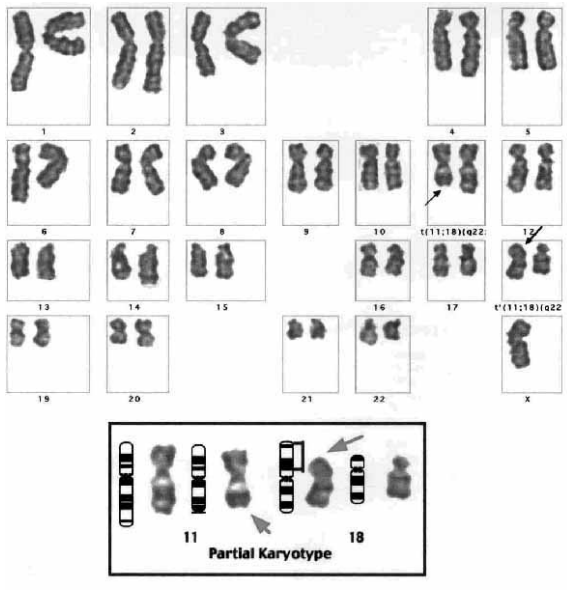
She was referred to us in the third pregnancy at 14 - 15 weeks for second opinion with suspected meningocele. Ultrasound revealed increased nuchal thickness with septations. Repeat scan and amniocentesis were suggested at 16 weeks and parental karyotyping in the meantime. Patient refused both. Repeat scan revealed persistent nuchal thickness, left sided pleural effusion & femur length below 5th centile. In view of progression of the USG findings, unfavourable prognosis was explained. The couple opted for termination. Autopsy revealed dysmorphic facies, posterior cleft palate, agenesis of both olfactory tracts, fusion of frontal lobes, lobar type of holoprosencephaly, agenesis of corpus callosum and abnormal cerebellum. Subsequently the parents opted for karyotyping which revealed balanced translocation between 11 & 18 in the father. Parents were counseled about the risk of recurrence and the need for serial scans and direct fetal sampling in every pregnancy.
In her 4th pregnancy scans done at Surat, revealed early onset IUGR. No prenatal diagnosis was offered. Patient delivered a 2 Kg female baby vaginally at term. No other details were available. Baby was referred to us for investigations. At 3 months of age, there was poor catch-up growth, frontal bossing, wide philtrum, prominent upper lip, micrognathia, short neck, multiple creases in the fingers, umbilical and inguinal herniae, presacral dimple, small and proximally placed great toes, no social smile / head control and normal echo. Karyotype of the baby revealed Partial Trisomy 11q.

Major diagnostic criteria reported for partial trisomy 11q

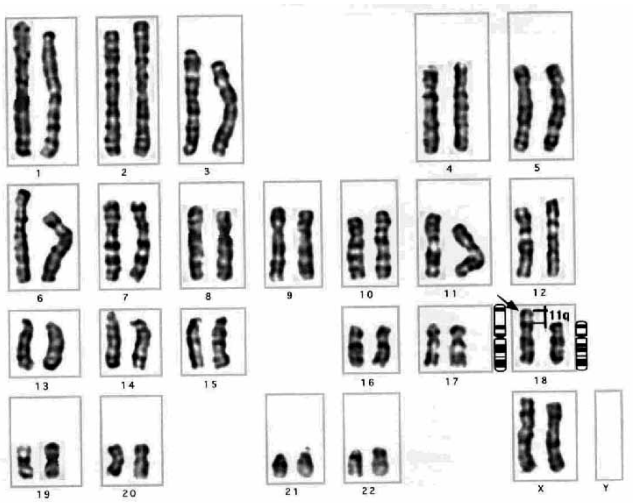
- Prenatal onset of IUGR
- Moderate to severe mental retardation
- Abnormal facies
- Abnormal creases
- CNS / CVS defects
- Survival is variable and the oldest reported is 16 years.

In conclusion, chromosomal anomalies should be suspected, when multisystem anomalies are detected on antenatal scan. If karyotype (KT) of the index case is not available, parental KT will be useful to rule out translocations in either of the parents. This will help us in the management of subsequent pregnancies.





Father's: 46 XY, t(11;18) (q22;pter)



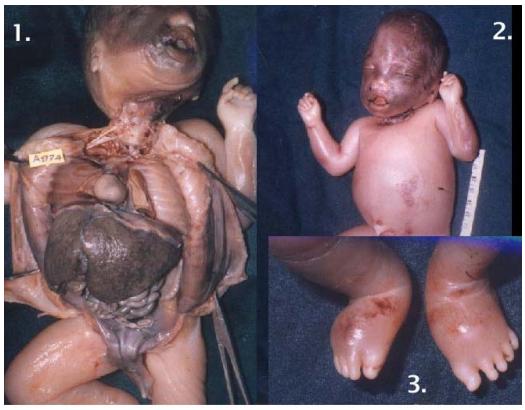
Child's : 46,XX, der(18), t(11:18)(q23;p11.32)

Case 2:

A 20 year old Rh negative mother, G3 P2L0, non consanguineous marriage was referred to us for an antenatal scan. Her first child was still born and her second child died soon after birth. The cause of deaths were not known. The mother was not given Anti-D after both deliveries.

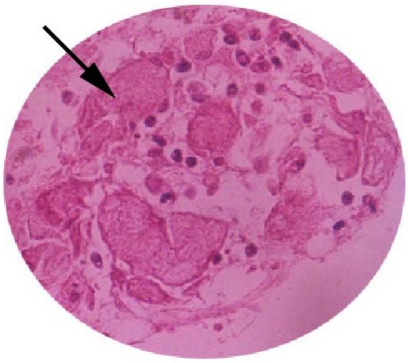
Ultrasound scan done at 21 – 22 weeks showed Hydrops with scalp edema. ICT in the mother was negative. After counseling, fetal blood sampling was done, which revealed marked Thrombocytopenia (Platelet count 26,000). DCT was negative. Fetal karyotype was normal. A diagnosis of NIH of unknown etiology was made. The fetus died in utero at 22 – 23 weeks and delivered vaginally after induction. Autopsy done on the fetus revealed icthyosis of the skin, dysmorphic facies, hypoplastic lungs and thymus and hepatosplenomegaly. HPE of the thymus, lungs, liver, spleen and adrenals revealed storage cells (Gaucher cells). The final diagnosis was Gauchers Type 2 B which is an autosomal recessive condition with 25% recurrence in every pregnancy. Thus, the importance of autopsy in explaining the cause of death in fetuses is illustrated in this case. It has enabled us to explain the NIH and recurrent neonatal deaths.

These are just a few among many more such instances where karyotyping and autopsy together help us in explaining fetal demise.



1. Hepatomegaly & hypoplastic lungs
2. Hydrops fetalis & dysmorphic face
3. Thick & abnormal skin

HPE of thymus showing Gaucher's cells



Our first meeting

Following is the list of Doctors who represented on their Hospitals at the first BDR meeting on 15th March 2001

Name of the Hospital	Participants	Code
Mediscan Prenatal Diagnosis & Fetal Therapy Centre	Dr. S. Suresh, Dr. Indrani Suresh, Dr. Sujatha Jagdeesh, Dr. Lathaa Bhat,Dr. Lata S, Dr. Gazala Jabeen,Dr.G. Thangavel, Dr.M.A.Shivarajan, Mrs. Ranjani-Pathasarathy, Mrs. Chandini Rajendran, Ms. Rehana	001
E V Kalyani Medical Centre	Dr. Arnab Basak	002
Sundaram Medical Foundation	Dr.Bhuvana	005
Vijaya Hospitals	Dr. Lalitha	006
Apollo Hospitals	Dr. Mini George	007
Corporation Hospital, Saidapet	Dr. Sheela Gopinath, Dr. V. Suganthi, Dr. Poongothai, Dr. N. Rajam	010
Public Health Centre, West Mambalam	Dr. Prabha Ganapathy, Dr. Karpaga Valli	011
CSI Rainy Hospital	Dr. Vijaya Lakshmi, Dr. V. Priya Menon	012
CSI Kalyani Hospital	Dr.Esther Gnanakumari	013
Nagamani Hospital	Dr. S. Premkumari	014

Other member hospitals

Name of the Hospital	Code
St. Isabel's Hospital	004
Sri Ramachandra Medical College Hospital	008
Andhra Mahila Sabha Hospital	009
GG Hospital	015

Issued four times in a year – January, April, July and October.
Published by Fetal Care Research Foundation, 203, Avvvai Shanmugam Salai, Royapettai, Chennai – 600014.