BDR NEWS

The official newsletter of The Birth Defects Registry, Chennai

(Unit of Fetal Care Research Foundation)

Volume 1 **Issue 2, July 2001**

Proceedings of the second BDR meet

The second meeting of the "Birth Defects Registry" was held on the 5th of July 2001 at Mediscan Systems. Dr.S.Suresh, Director welcomed the gathering. The overwhelming support and cooperation extended by the member hospitals during this period was well appreciated.

Dr. Sujatha Jagadeesh presented the statistics, received from the hospitals during January, February and March in detail. Modifications were suggested for entering the data correctly in the BDR General Statistics Proforma, to maintain uniformity. Modified and new version of BDR General Statistics and BDR Proforma were also presented and views sought from the members. Suggestions given by the members were as follows:

(a) IUD (> 24 weeks) and still born to be classified under the same category.

(b) All abortions other than MTP (option / anomaly) to be classified as under spontaneous abortion.

(c) An additional field to be included after "final diagnosis" in the BDR Proforma to give details on whether the anomaly is correctable or non correctable and if correctable what was the postnatal management.

It was agreed to include all the suggestions in the new format. Members were instructed to use the new forms as a trial run for the next two months to perfect the present data entry system. When this system is perfected, all statistics will be fed into the master registry.

Dr.S.Suresh thanked all the members and assured them that the project will be pursued with all earnestness. The data will be useful for us to understand our population birth defects. Dr. Radha Rajagopalan, Medical superintendent of Apollo Hospitals, extended her invitation to have the next BDR meeting at Apollo Hospitals, which was gratefully accepted. It is scheduled to be held in September. Date will be announced to the members shortly.

Neural Tube Defect - Genetic counseling

Dr. Sujatha Jagadeesh, Dysmorphologist, FCRF

The incidence of neural tube defect is around 2-3/1000 live births. It includes spina bifida occulta, meningocoele, meningomyelocoele, encephalocele & anencephaly. Among these, anencephaly is lethal; long segment meningomyelocoele is associated with severe morbidity; spina bifida occulta & meningocoele may not be associated with gross neurological deficit; encephalocoele may have varied outcome depending on size, location operative feasibility etc.

Development of Neural tube defect:

Neural tube has an anterior neuropore which closes by 24 days of life and a posterior neuropore which closes by 28 days of life (Figure 1). Delayed closure of anterior neuropore results in anencephaly, delay in posterior neuropore closure leads to meningomyelocoele.

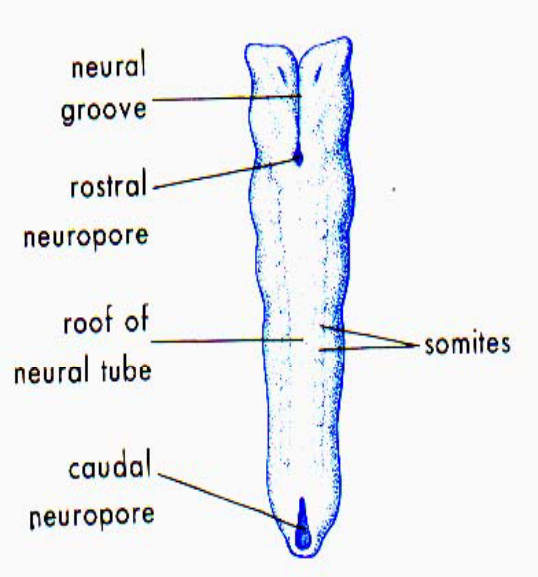


Fig. 1: Embryonic development of Neural tube

Who are at risk for neural tube defect?

a. Mother with previous child with NTD.
b. One of the parent with spina bifida.
c. The sibs of parent with a child with NTD.
d. The cousins of an affected child.
e. Any woman who is consuming folic acid deficient diet.
f. Women with uncontrolled diabetes mellitus.
g. Women on antiepileptic medication.

Case History 1:

25 years old, Primi Gravida had Medical Termination of Pregnancy at 24 weeks of gestation as antenatal ultrasound revealed Lumbar Meningomyelocele about a month ago. She has come to the clinic for prepregnancy counseling

What should be done?

1. Detailed history with family pedigree obtained.
2. Previous scan reports and investigations reviewed.
3. Contraception for about six months advised to help mother to recoup physical and mental health.
4. Periconceptional folic acid-4mgs/day at least 2mths prior to conception to be continued through the 1st trimester of next pregnancy.
5. As it is a mutifactorial condition (environmental factors altering genes) the recurrence is low-3-5% after one affected child or one affected parent, 10-15% risk after two affected children.
6. Maternal diabetes to be ruled out prior to conception as it is a potential teratogen.
7. Mothers on antiepileptic medications (esp.Valproate)should be put on either safer drugs or given the minimum possible dosage along with folic acid supplementation.
8. MSAFP is offered at 16-20 weeks and a targeted anomaly scan at 20 weeks in her subsequent pregnancy.
9. Parental reassurance is also given.
10. If there is a live affected child it is examined and suitable referrals made for the functional betterment of the disabled child.
11. Support groups are introduced so that parents learn about overcoming practical difficulties in bringing up such a child.

Case History 2:

23 years old, Primi gravida, non consanguineous marriage, married for 4 years. Conceived after follicular monitoring and had a course of Injection Profasi. USG at 9 weeks normal. Scan at 19 – 20 weeks revealed Arnold Chiari Malformation, Lumbosacral Meningomyelocele and bilateral Talipes Equinovorous.

How to Proceed?

1. Detailed history with family pedigree obtained
2. Associated problems if any (Arnold Chiari malformation, club feet, wasting of lower limbs, congenital dislocation of hips etc) are explained.
3. Associated anomalies are also looked for specifically ;Though rare, associated chromosomal anomalies, single gene disorders and sporadic syndromes are known to occur. Knowledge about this is important for counseling as recurrence risk varies depending on the associations
4. Specific information regarding maternal diabetis & teratogenic drug intake to be obtained.
5. Severity of the anomaly has to be explained to the couple with illustrative diagrams so that they understand well. Medical jargon is to be avoided during counseling.
6. Facts like: anencephaly is lethal, long segment spina bifida can be associated with neurological deficit, bladder bowel disturbance, surgical corrections are available, problems to be expected and low success rates in total rectification of the defect have to be explained. In an ideal situation a neurosurgeon may be requested to talk with the couple. The aim of the whole process is to ensure that the couple is maximally informed about the problem before they take a decision
7. The primary physician is also informed about the counseling information and a close follow up to be obtained.
8. If the parents decide to terminate the pregnancy the importance of perinatal autopsy is explained.
9. If the patient is willing for the same then patient is called back for pre pregnancy counseling at which time we proceed as above.

Case History 3:

32 year old Primi came for a second opinion for her antenatal ultrasound at 34 – 35 weeks of gestation. USG revealed Long segment Lumbo Sacral Meningomyelocele with Arnold Chiari Malformation.

What counseling should be offered?

Anomaly detected in late pregnancy:

1. A detailed history with family pedigree is taken
2. The anomaly is explained to the couple.
3. Parents may be given two options:
   a. Perinatal hospice – Baby just given feeds and parents get an opportunity to handle their baby as long as it lives.
   b. Active intervention to restore the baby's function to as normal as possible. This is achieved by referring the baby to appropriate specialists and introducing support groups to the family.
4. Pre pregnancy counseling offered prior to next pregnancy.

Case History 4 :

22 year old primi gravida, with history of Spina Bifida Occulta (Clinically tuft of hair seen and xray reveals Spina Bifida) was prescribed Folic acid from 6th week of gestation. Antenatal ultrasound at 20 weeks gestation

revealed gross lumbosacral myeloschisis and Arnold Chiari Malformation.

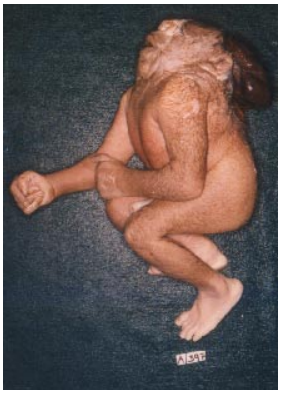
Is it all right to start Folic acid at 6 weeks of gestation? What would have been the proper methodology?

In this situation Folic acid should have been started at least one month prior to the pregnancy and continued through the first trimester. Protection given by this will be around 50 – 60% Neural Tube closure occurs by 28 days of gestation and any supplement given after that would not be beneficial.

Messages:

- MSAFP & targeted anomaly scan in all pregnancies will improve early detection of neural tube defects
- Periconceptional folic acid helps in preventing recurrence but is not useful if started after confirmation of pregnancy as neural tube closes at 28 days of life. It is said to reduce the recurrence risk in a woman with previous neural tube defect from 3-5% to 1%).
- Pre pregnancy counseling helps to prepare the couple well before the next pregnancy. Affected child's' uncles, aunts and cousins families may be offered periconceptional folic acid MSAFP at 16-20 weeks pregnancy and targeted scan at 20 weeks.
- Fortification of food (flour) with Folic Acid should be implemented on a national level. Public awareness should increase. The American Academy of Pediatrics endorses the US Public Health Service (USPHS) recommendation that all women capable of becoming pregnant consume 400 microgram of folic acid daily to prevent neural tube defects (NTDs). Studies have demonstrated that periconceptional folic acid supplementation can prevent 50% or more of NTDs such as spina bifida and anencephaly. For women who have previously had an NTD-affected pregnancy, the Centers for Disease Control and Prevention (CDC) recommends increasing the intake of folic acid to 4000 microgram per day beginning at least 1 month before conception and continuing through the first trimester. Implementation of these recommendations is essential for the primary prevention of these serious and disabling birth defects. Because fewer than 1 in 3 women consume the amount of folic acid recommended by the USPHS, the Academy notes that an urgent and effective NTD prevention campaign is the need of the hour.

Following figures show various types of NTD



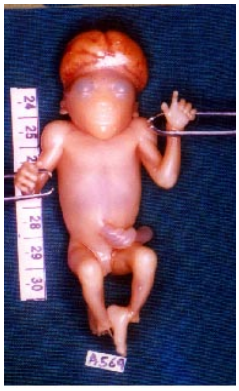


Fig. 2. Anencephaly *Fig. 3. Exencephaly*

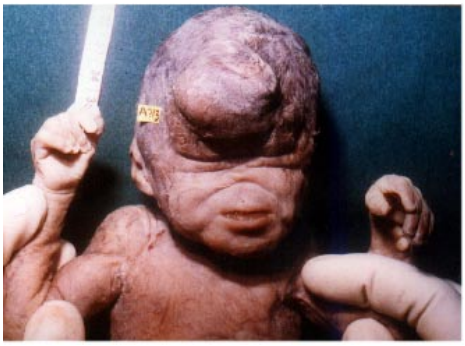


Fig. 4. Frontal Encephalocele

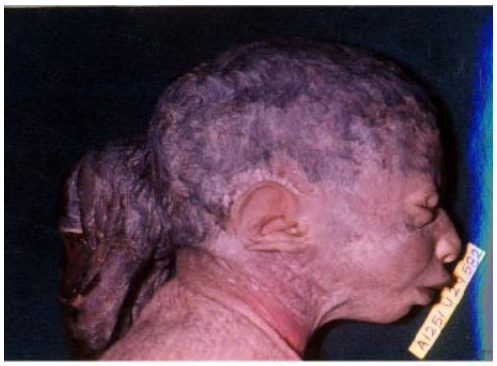


Fig. 5. Occipital Encephalocele

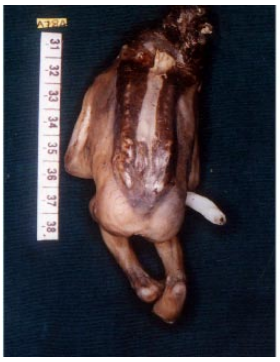


Fig. 6. Spinal Rachischisis

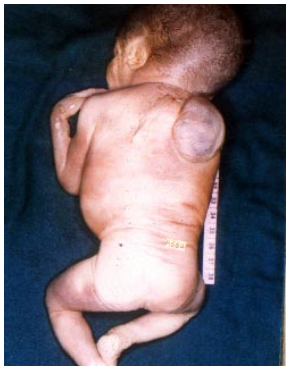


Fig. 7. Thoracic Meningomyelocele

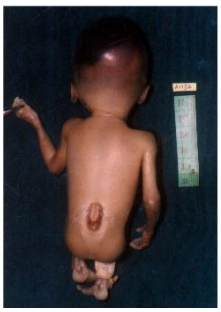


Fig. 8. Lumbar Meningomyelocele

Other member hospitals

Name of the Hospital	Code
St. Isabel's Hospital	004
C.S.I. Kalyani Hospital	013
Nagamani Hospital	014
G.G. Hospital	015

Following is the list of Doctors who represented their Hospitals at the second BDR meeting on 5th July 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. Mona	006
Apollo Hospitals	Dr. Radha Rajagopalan,Dr. Hari Shankar	007
Sri Ramachandra Medical College Hospital	Dr. S. Balagopal	008
Durgabai Desmukh General Hospital	Dr. Pethammal,Dr. Indira	009
Corporation Hospital, Saidapet	Dr. Sheela Gopinath, Dr. B. Shanthi, Dr. Rajeswari, Dr. N. Rajam,Dr. M. Raja Meenatchi	010
Public Health Centre, West Mambalam	Dr. Prabha Ganapathy,	011
CSI Rainy Multi Specialty Hospital	Dr. Vijaya Lakshmi, Dr. Swathi	012

BDR members are most welcome to publish their write ups and case presentations in the news letter.

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