# B D R News

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

Volume 5 Issue 3: July 2005


**PROCEEDINGS OF THE BIRTH DEFECTS REGISTRY MEETING HELD ON 12/08/05**

The third Birth Defects Registry meeting of the year was held on the 12th of August 05 at Mediscan Systems Annexe, Chennai. Dr.S. Suresh welcomed the audience. He extended his invitation to all BDR members for the inauguration of Newborn Screening Program, launched as part of the birth defects prevention project on the 17th of September 05.He was glad to disclose that, a poster from our registry has been accepted for display at the II "International Conference on Birth Defects and Disabilities in the developing world" to be held in Beijing, China in September. The CME topic for the day was "Congenital Lymphangiomas". Dr. Suyashree Palkar spoke on the "Antenatal Presentation of Congenital Lymphangiomas" and Dr. S. Balagopal gave a presentation on the "Postnatal Management of Congenital Lymphagiomas". The excerpts of the presentations are given below.

**ANTENATAL PRESENTATION OF CONGENITAL LYMPHANGIOMA**


Dr. Suyashree Palkar, Fellow, Fetal Medicine,
Mediscan Systems.


**CONGENITAL LYMPHANGIOMAS** are hamartoma's of the lymphatic vessels representing 10% of all benign head & neck tumors.

***The causes of lymphangiomas*** can be

- Altered embryonic development,
- Abnormal embryonic sequestration of lymphatic tissues or
- Altered communication between the lymphatic and venous system.

They may be ***classified as***

1) Superficial: a)Lymphangioma Simplex eg. Found on Tongue & Genital. b) Lymphangioma Circumscriptum eg. Found on Face, Chest & Extremities.

2)Deep: a) Cavernous lymphangiomas which may be superficial and deep. b) Cystic lymphangioma. Eg. Nuchal cystic hygroma

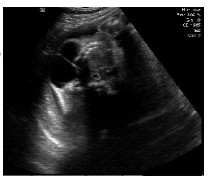
Nuchal Cystic Hygroma is nothing but a cystic lymphangioma which is traditionally labelled so when present in the neck.

***Lymphangiomas may be***

***1.Isolated*** - Sporadic / Associated Autosomal recessive trait.
***2.Associated with*** other structural abnormalities or
***3.Associated with*** Chromosomal abnormalities or Syndromes.

**LYMPHANGIOMA- SITES OF PRESENTATION**

***Isolated may be*** distributed in Head, Neck & Axilla - 75%,Trunk - 11%, Mediastinum - 1%, Extremities -11%,Abdomen & Genitalia 3%. These tumors can infiltrate into surrounding tissues & compress the adjacent organs due to their rapid growth.



50-80% of patients with nuchal cystic hygromas have chromosomal abnormalities and various malformation syndromes.

***Lymphangiomas may be associated with chromosomal abnormalities such as:***

a) Turner's sydrome (45XO-majority),
b)Trisomy21,c)Trisomy18 and d)Trisomy 13.

***Lymphangiomas may be associated with syndromes such as:***

a) Noonan Syndrome
b) Fetal Alcohol Syndrome
c) Klippel Trenaunay - Weber
d) Lethal Multiple Pterygium
e) Pentalogy of Cantrel
f) Tuberous Sclerosis
g) Congenital erythropoetic porphyria
h) Wolf - Hirshchorn syndrome

***Ultrasound features of Lymphangiomas:***

Since 50% of cystic lymphangiomas are present at birth and 90% are present by 2yrs of age, pre-natal diagnosis is important.

a) Cystic lymphangiomas appear as sharply defined unilocular or multilocular cystic masses with thin or thick walled septae.
b) Fluid within cysts appears anechoeic, enhanced through transmission.
C)They may have variable internal echoes or fluid filled levels due to bleeding and fibrin deposition.

***The protocol for diagnosis is as follows:***

Determine the size, extent of lesions and its proximity to vital structures, and the presence of vascular component or whether it is associated with other structural abnormalities. Monitoring the lesion is important for associated development of hydrops, polyhydramnios & skin edema.

***Regression of certain lymphangiomas are known to occur in Utero.***

***The limitations of USG are:***

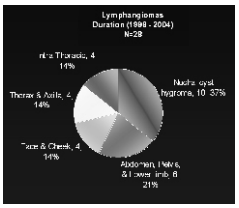
a) Operator dependency
b) Limited field of view.

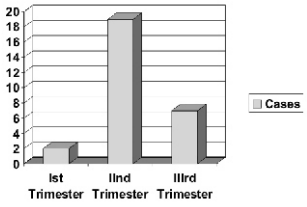
***Complementary imaging techniques used for diagnosis are:***

1)Colour doppler to detect vascularity within the lesions.
2)3D Ultrasound to render volume imaging &
3)MRI: which a) characterizes the lesion, b) shows better anatomical location, and c) accurately delineates the extent & limits of the mass and involvement of important structures.

**MEDISCAN EXPERIENCE:**

Between1998 - 2004, there were 28 cases of lymphangiomas diagnosed within 2 to 19 weeks of pregnancy. Their follow-up details are as follows:

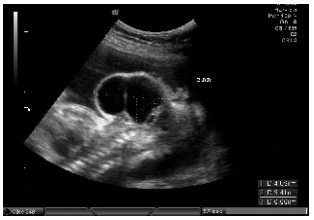


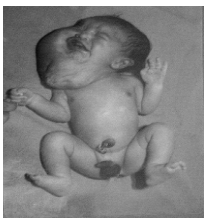




***Lymphangioma of face & cheek - n=6***

KT was done for these cases.

| *Case no.* | *Details* |
|---|---|
| 1. | Ongoing pregnancy being followed up |
| 2. | Twin Delievery - Baby A - Normal |
| 3. | Baby B - To review the cheek lymphangioma at 7 - 8yrs of age. |
| 4. | Lesion confirmed postnatally. No further details available |
| 5. | Pregnancy terminated |
| 6. | Lost to follow up |





***Intra thoracic tumour - n=4***

KT was done for these cases

| *Case no.* | *Details* |
|---|---|
| 1. | Solitary cyst regressed spontaneously |
| 2. | Prematurely born & dead |
| 3, 4 | Lost to follow up |

**CONCLUSION**

1) KT was advised all other lymphangiomas for all fetuses with nuchal cystic hygromas, in all other lymphangiomas without KT, all live borns were normal.

2)Nuchal cystic hygromas can be associated with other abnormalities.

3)Large lesions tend to extend and may progress to become Non- Immune Hydrops there by being associated with poor prognosis. Hence termination of pregnancy is warranted if IUD does not happen earlier.

**PROGNOSIS & MANAGEMENT**

1.Normal outcome rate < 10%

2.Prental diagnosis of Cystic lymphangiomas /Cystic hygromas depends on the size and extent of the lesion., careful assessment of the fetus and the presence of Non Immune Hydrops.Karyotyping is necessary for nuchal cystic hygromas.

3.In fetuses with normal KT & Isolated lesions, a)Spontaneous resolution may occur. b)In larger lesion with extension and involvement of important structures, the outcome may be less favorable with progression to NIH, polyhydramnios and lymphedema.c) Post-mortem examination should be encouraged giving vital information both for the current and future pregnancy.

***Nuchal cystic hygroma - n=10***

- KT done - 4 cases
  - -Normal KT -3
  - -Trisomy 18 - 1
- KT not done - 6 cases

Medically terminated - 7 cases
Surgically Corrected - 1
Lost to follow up - 2

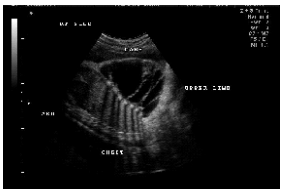
***Reasons for Termination (n= 7)***

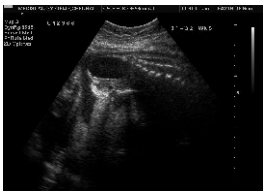
1. Dandy Walker malformation
2. Non-immune Hydrops
3. Teratoma on Histology
4. Multiple anomalies on autopsy
5. Trisomy - 18
6, 7.MTP by option

***Lymphangioma of Thorax - n=4***

- KT done - 2 cases
- KT not done - 2 cases

| *Case no.* | *Details* |
|---|---|
| 1. | Surgical correction done posnatally for the lesion extending axilla to abdomen. |
| 2. | Pregnancy terminated |
| 3, 4 | Lost to follow up |





***Lymphangioma of Abdomen & pelvis - n = 6***

| *Case no.* | *Details* |
|---|---|
| 1. | Posted for surgery in gluteal region |
| 2. | Terminated for multiple anomaliesl |
| 3. | Still born. |
| 4, 5, 6 | Lost to follow up |

**POSTNATAL MANAGEMENT OF CONGENITAL LYMPHANGIOMA**

Dr.S. Balagopal. Cons. Paediatric Surgeon,
Sri Ramachandra Medical College & Research Institute.

Dr. Balagopal started his speech by first describing the anatomy of the Lymph system.

The lymph system can be divided into 4 layers such as:

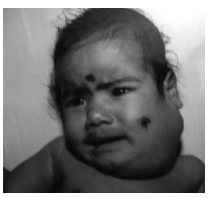
1) Superficial primary layer which are valveless
2) Sub dermal secondary layer which have valves
3) Deep layer on the muscular wall with valves and
4) Intramuscular layer which is independent.

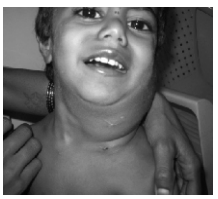
These join the main ducts:

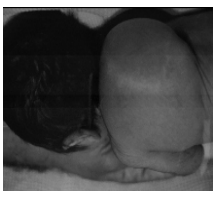
1) abdominal cisterna chyli & thoracic duct drain the abdomen, viscera and extremities and join the left jugular vein at the subclavian junction.

2)The right lymphatic trunk drains the right side of the head,neck, right arm & right chest joining the right jugular vein at the subclavian junction.

He elaborated on the postnatal surgical management of various lymphangiomas according to their site of presentation and morbidity level.







***Postnatal Management of Lymphangiomas:***
Cystic hygromas are multi loculated cystic spaces present in 1/12,000 births. Complications involved here would include pain, recurrent infection, cosmetic problems, Lymphatic leak/intralesional bleeding, pressure exerted on adjacent structures causing -Respiratory obstruction, Dysphagia, Nerve compression & Dental malocclusion.

Prenatal or nuchal post cervical cystic hygroma as already discussed has high mortality rate. Fetal KT is warranted to rule out chromosomal anomalies. Genetic counseling helps the family get an insight in to the problem and make appropriate decisions.

***Cystic hygroma is excised by***
***Surgery*** between 2-6months of age. Surgical complications include recurrence , fistula, infection, damage to vascular structures and nerves (7,9,10,11,12) cranial nerves, symphathetic system & branchial plexus). The mortality rate ranges from 2 to 6%.
***Incision & Drainage*** procedure for cystic hygroma involves the risk of infection and pressure symptoms. ***Sclerothrapy*** is another option using reagents such as Bleomycin ,OK.432, Doxycycline and Fibrin glue. Swelling is the complication of this procedure.

***Intra-abdominal lymphangiomas*** are usually detected in early infancy (90% before 2yrs) and they present as abdominal mass, intestinal obstruction, infection, bleeding , perforation, Volvulus and rupture. Ultrasound & Computerised Tomography are the diagnostic modalities. Postnatally complete excision of these tumors is advised.

***Capillary tumors are removed by***
a.Surgical excision b.Cryotherapy ( liquid $N_2$)
c. Electrocoagulation d.Laser ($CO_2$)

***Cavernous tumors are extricated by***
*a.* Surgical.Excision eg. Partial glossectomy
b.$CO_2$Laser - e.g. Head & Neck tumors impinging on airways.

***Dr. Balagopal concluded saying that,***
- There are no clear cut guidelines to terminate the pregnancy with isolated lymphangioma, when it is not compressing the other structures.
- There is no urgency to operate on a lymphangioma, except when it is present on the neck region and there is evidence of secondary complication.

**DISCUSSION**
There was an enlightening discussion between the audience and the speakers after the session. The details are as follow:

***1. What is the role of aspiration of lymphangioma in obstetric management?***
Aspiration need not be done in pregnancy for lymphangiomas. It may be warranted when it is huge in size and may compromise the process of delivery (either vaginal or Caesarian).

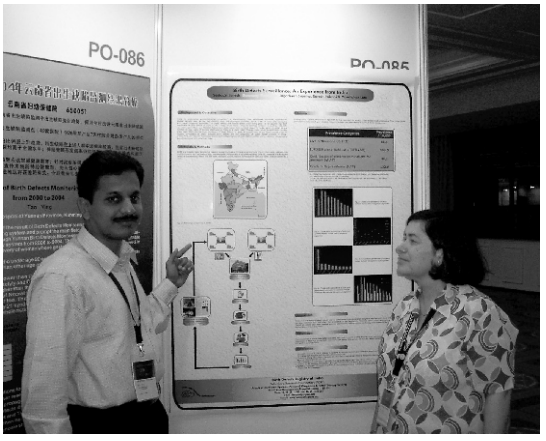
***2. Is calcification a feature of lymphangioma?***
Calcification may be a pointer to teratoma rather than a lymphangioma.

***3. Is AFP an indicator of malignant tumour?***
AFP level increase may be an indicator of malignant tumours. But standardized AFP values (MoM) are available only between 16 - 20 weeks of pregnancy and hence in the absence of standard values throughout gestation, it is not appropriate to use this as an index to diagnose malignancy.

**INTERNATIONAL CONFERENCE ON BIRTH DEFECTS WAS REPRESENTED BY BDRI**

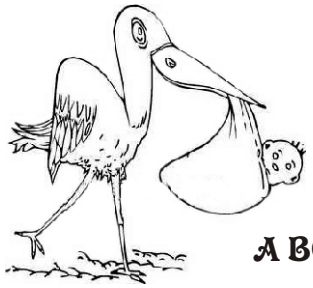
Dr.G.Thangavel, Epidemiologist, BDRI, participated in the ***SECOND INTERNATIONAL CONFERENCE ON BIRTH DEFECTS AND DISABILITIES IN THE DEVELOPING WORLD*** held at Beijing, China from 11th to 14th September 2005. The conference was sponsored by - Ministry of Health, China, Centre for Disease Control and Prevention (CDC), National Institutes of Health (NIH), March of Dimes Birth Defects Foundation - USA, World Health Organization (WHO), United Nations Children's Fund (UNICEF) and several other organizations. The prime objective of the conference was to identify steps to reduce gaps and disparities between developed and the less developed nations to reduce the impact of birth defects and genetic diseases on infant mortality. As part of the conference, participants signed a declaration affirming the importance of birth-defects prevention and health promotion for affected individuals. A large number of Epidemiologists, Clinical Geneticists and other professionals related to birth defects research both from the developed and developing nations across the globe participated. Apart from Dr. G. Thangavel, Prof. Ishwar C. Verma of Sir Gangaram Hospital, and Dr. Satyajeet Nanda, Demographer from New Delhi also participated in the conference.

Dr.G..Thangavel presented a poster titled "Birth defects surveillance - an experience from India", which summarized the data collected by BDRI over the past four years. He had an opportunity to meet eminent scientists involved in birth defects research. Notable among them were Dr. J David Erickson and Dr. Robert J Berry, Epidemiologists from CDC who were the main architects of China-US synergy on birth defects research. He held detailed discussion with Dr. Erickson about the pioneering role of BDRI in estimating the prevalence of birth defects in India and explored the possibility of collaborating with CDC, USA. He also met Prof. Pierpaolo Mastroiacovo, Director of International Clearing House for Birth defects and Dr. Eduardo E Castilla, from South American Birth Defects Registry (ECLAMAC), Dr. Mary-Elizabeth Reeve, Perinatal Epidemiologist, March of Dimes, who pledged their support to BDRI.

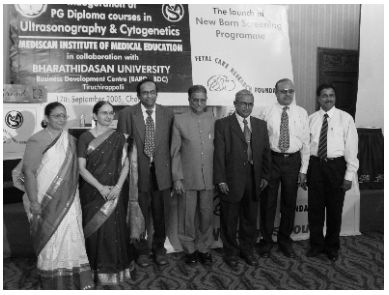



**BDRI & Rotary Madras Metro Partnership Program for Birth Defects Prevention**

**REPORTS ON INTERESTING EVENTS OF THE REGISTRY FOR OUR READERS**



## A BOLD STEP FORWARD




September 17, 2005 will go into the annals of Mediscan systems and Fetal Care Research Foundation (FCRF), Chennai as an important day for it was on that day twin "PG Diploma courses in Ultrasonography and Cytogenetics" were inaugurated and New Born Screening Program (NBS) for three relatively common metabolic disorders was launched.

The event organized by Mediscan Systems and FCRF took place in class and style attended by Obstetric, Paediatric Medical fraternity, Rotarians and the well wishers of FCRF. Dr. C. Thangamuthu ,Vice Chancellor, Bharathidasan University Business Development Centre, Thiruchirapalli, presided over the function. Dr. S.P. Thyagarajan, Vice Chancellor, Madras University was the Guest of honour.

Dr.Indrani Suresh welcomed the gathering followed by a video presentation of Mediscan Systems' quality services and achievements, in the past 23 years, and the mission of FCRF to serve the community at large. Dr. S.Suresh in his address said how Mediscan Systems found new vistas were opening up for their endeavours, in the field of medical education. Today with more than 40,000 Hi-tech Ultrasound Equipments in India, we lack adequately trained technicians to produce authentic reports using this diagnostic modality. Only a very few centres offer structured ultrasound training here. There is not only need but great demand for I level non-medical ultrasound technicians to ease the burden of the Radiologists and Obstetricians, who can concentrate on diagnostic ultrasound and invasive procedures. By designing and introducing PG Diploma course in ultrasound, Mediscan Institute of Medical Education not only prepares qualified I level ultrasound technicians but also provides job opportunities for science graduates. Training will be imparted by experienced Sonologists at Mediscan Systems keeping on par with international standards. As the technicians need continuous upgrading of knowledge , Mediscan Institute of Medical Education also offers recertification courses, he added.

Dr. C.Thangamuthu, in his inaugural address mentioned how he was impressed by the meticulous documentation of patient records for posterity at Mediscan Systems. He further added that documentation is important for any diagnostic institution which has a focus in clinical research. He said that the University would do the evaluation and certification of the candidates at the end of two years. He comprehended that Bharathidasan University is convinced and pleased to have academically associated with Mediscan Systems by accrediting its PG diploma courses, for Mediscan already enjoys the social and professional accreditation, and it is a win-win partnership for both the institutions concerned.

Dr. Sujatha Jagadeesh, Clinical Dysmorphologist, FCRF, highlighted the efficacy of New Born Screening as a public health care strategy. She enunciated that NBS should be made mandatory similar to developed countries. As there is treatment for many of the metabolic disorders, early detection and intervention would help the child have quality life. Dr. R.S.Shanmugasundaram,Consultant Neonatalogist, and Dr. P.M.Gopinath, Past President, OGSI gave their views on NBS in their respective perspective.

Prof. S.P.Thyagarajan after inaugurating the NBS Program said that Nano technology is going to change the current scenario of medicine, and it is better to acquaint ourselves with this knowledge and apply it in preventive healthcare programs such as newborn screening program.

Any community service project for its sustenance and success needs not only commitment but also social and economic support. Rotarian J.Raviraj, on behalf of Rotary Club of Madras Metro pledged commitment and continued support for the 'Prevention of Birth Defects' program on a long term basis. Two cheques for corrective surgeries were distributed by Rtn President, S.Prakash to 2 families of children affected by a metabolic disorder known as MPS. Later, Donors who have made significant contributions to FCRF were felicitated.

## *We Extend...*

*Hearty welcome to our new nodal member Dr. Bimal Sahani, Sonocare Diagnostic Centre and the participating members of Aurangabad .*

**AURANGABAD BIRTH DEFECTS REGISTRY INAUGURATED ON 25/09/05**

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