Report of Workshop on "Addressing Congenital Anomalies in Indian Infants"

Date: 6th September 2014

Organized by:

Rapporteur:

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http://www.venturecenter.co.in/campaigns/mch/events.php

A workshop on "Addressing Congenital Anomalies in Indian Infants" was organized by Bioincubator at Venture Center (Bioincubator at Venture Center is supported by BIRAC, Government of India) on Saturday, 6th September 2014.

This report contains:

Key Lessons from the workshop

1. Need for data on birth defects

- To understand the exact needs of patients with birth defects, needs assessment studies with data mining exercises. Some birth defects according to the burden in priority could be: congenital heart disease, neural tube defects, Down's syndrome, orofacial clefts, sickle cell disease, rhesus D haemolytic disease of the newborn, congenital hypothyroidism and thalassaemias.
- Identifying individuals who may benefit from genetic services: Screening tools for suspecting a case; Rapid detection kits for identification of the condition
- Surveillance of Birth Defects by devising a basic monitoring system to give Baseline and representative Indian data, starting with common birth defects like Neural Tube Defects.
- Building robust documentation systems which understands all complexities of the Indian health care delivery system (primary, secondary and tertiary level delivery system) and ensures that data is not lost to follow up.
- Technology based applications for documenting and diagnosing patients at the community level to send data from community to the expert.

2. Training modules for health professionals and community

- Recognizing historical and physical features of common genetic conditions by education of health professionals
- Monitoring the health of individuals with a genetic disorder by education of health professionals
- Providing basic genetics information to patients and families by generating easy to understand literature
- Appropriately referring patients by informing doctors about these services
- Training of professionals in counseling, ultrasound, invasive tests and fetal therapy

- Improve awareness amongst the medical fraternity and the patient community regarding the availability of diagnostic tests.
- Education or creating awareness amongst general public, medical fraternity, basic sciences fraternity and need for interdisciplinary coordination.
- Training related to pre and post test counseling

3. Nutritional interventions

- Nutritional interventions: Macronutrients and micronutrients like iron, folate, iodine
    a. Lifestyle: Addictions and non-prescription therapeutic drug use
    b. Maternal health: Bad obstetric history, diabetes, TB, epilepsy
    c. Limiting exposures: Environmental, Infectious
- Nutritional interventions from pre-conception stage. Eg: Folic acid and B12 intervention pre-conceptionally for preventing NTDs and congenital heart defects. Need for food fortification innovations

4. Provision of comprehensive services – counseling, diagnostics, therapeutics

- Providing a medical home, a set of comprehensive care services
- Developing psychosocial support services
- Possessing knowledge of how to access the full range of genetics services from which patients might benefit and facilitating use of such services by creating lists of available services
- Need of support groups for various birth defects, financial support and resource allocation for caring the affected child.
- Effective public health programs for birth defects
- Production of cheaper therapeutics for managing birth defects. Eg: Cheaper recombinant drugs for managing hemophilia bleeds.
- Innovation in transfusion related problems related to devices and diagnostics related to blood transfusion

- Innovation in manufacturing several blood products from donated blood as per requirement of patients.

5. Need for point of care diagnostics for genetic disorders

- Commercialization of available laboratory methods – making them affordable to the patients
- Enzyme analysis and molecular diagnosis for genetic disorders should be available commercially and at affordable prices at more laboratory locations.
- Screening for neonatal disorders can be given priority: congenital hypothyroidism, congenital adrenal hyperplasia, galactosemia, phenylketonuria, Glucose-6-phosphate deficiency, biotinidase deficiency, Medium-chain acyl-CoA dehydrogenase (MCAD) deficiency, Maple syrup urine disease (MSUD), homocystinuria, aminoacidopathies, hemoglobinopathies and cystic fibrosis.
- Newborn and Prenatal screening and Diagnostic services
- Indigenization of majority of diagnostic platforms which are mostly imported
- Indigenization of several molecular investigations which are still outsourced
- Reducing Turn Around Time (TAT) of diagnostic tests

6. Need to handle ethico-legal aspects of diagnostic tests

- Stringency of PCPNDT act, devise easy way of maintenance of records for submission
- Ethical and regulatory implications of diagnostic tests

Detailed record of the workshop

A workshop on "Addressing Congenital Anomalies in Indian Infants" was organized by Bioincubator at Venture Center (Bioincubator at Venture Center is supported by BIRAC, Government of India) on Saturday, 6th September 2014.

The aim of the workshop was to network all stakeholders to help initiate useful projects, start-ups etc for addressing Child health related issues prevalent in India.

Workshop aimed to:

- Provide an update of key issues relating to birth defects in infants in India.
- Explore potential industry-academia/research partnerships in technology development and advancement.
- Foster networks with industry/academic experts/doctors/NGOs.
- Introduce some recent funding opportunities

The audience was well represented by participants from the academia, NGOs, technocrats, entrepreneurs and medical professionals. The workshop began with a brief welcome by Dr. Premnath introducing the various activities of Venture Center. A brief background of the workshop was given by Pradnya Aradhye.

A quick overview regarding Congenital anomalies in India highlighting the current status, key problems and needs and opportunities to address these issues was given by Mugdha Lele. According to WHO, birth defects are defined as structural or functional anomalies, which are present at the time of birth. There are several complex causes for the causation of birth defects. In approximately 50% cases there is no known specific cause. The known causes can be divided according to the stage at which these originate. Preconception stage causes could be genetic or partially genetic, origninating mostly before conception. These include chromosomal abnormalities, single-gene defects etc. Post-conception stage causes develop after conception

but before birth. These primarily include non-genetic causes, which are teratogens like radiations, environmental pollutants, maternal illness or maternal infections or drugs taken by the mother during pregnancy. The burden due to birth defects is being discussed in the background of the "Health Transition" which has set in India. In developed countries, due to this health transition, there is a decline in infant and under-5 mortality due to infectious diseases and malnutrition, but the mortality from birth defects remains constant. It has been observed that as countries develop, birth defects form a greater proportional cause of infant mortality. This effect will be seen in India, in the coming few years.

Some vital statistics in India were shared in this session:

- Neonatal mortality rate for India (2012) – 31/1000 live births
- IMR – 44/1000 live births
- Under-5 Mortality Rate – 56/1000 live births vs MDG target for 2015: 42/1000 live births

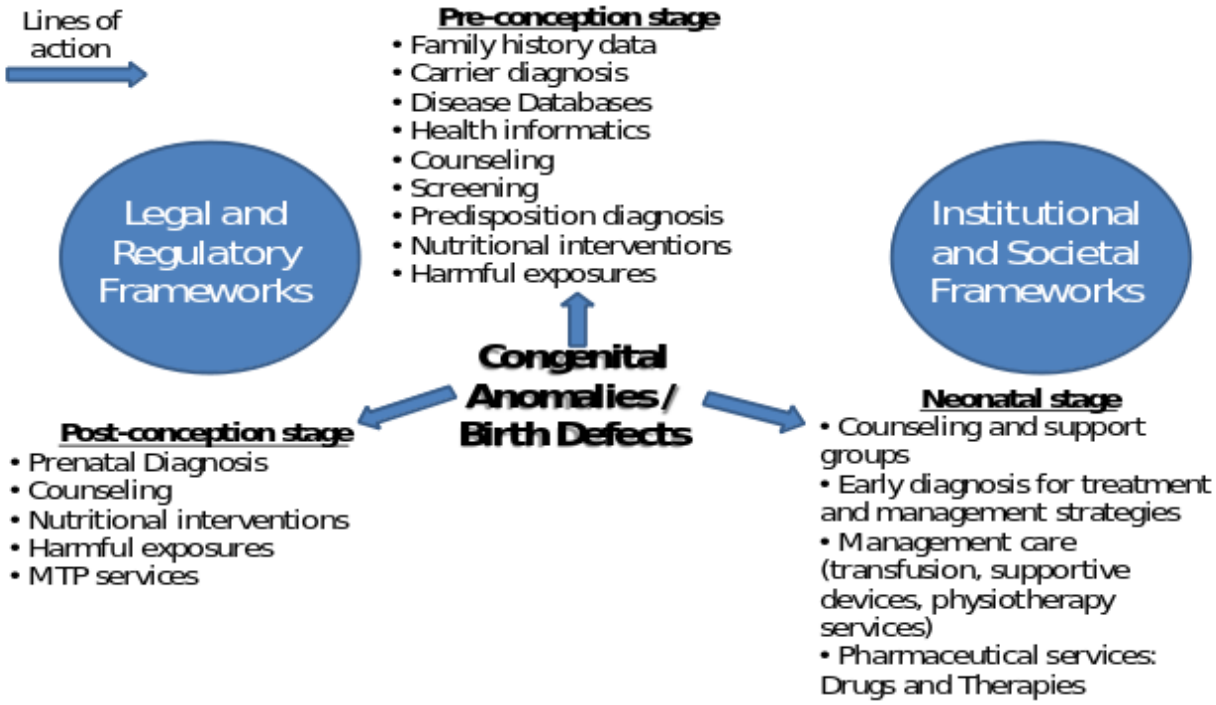
Amongst neonatal deaths, congenital anomalies contribute to 8.8% deaths according to a paper by Liu *et al* (2012). The firs report regarding the burden of birth defects is the March of Dimes report (2006) which highlights that up to 70% of birth defects would either be prevented or, with proper care, managed for better quality of life.

Some priorities of interventions were also highlighted:

- Development of affordable and easy-to-use diagnostics for identifying birth defects in the prenatal and neonatal stages to ensure early diagnosis.
- Development of non-Invasive and/or least Invasive technologies
- Affordable and Point of care screening tests/products for newborns for detecting prevalent disorders
- Innovate pre-conception and prenatal diagnostic techniques
- Innovative technologies / products for newborn care
- Improving the specifications of monitoring and life-support systems to suit Indian requirements

- Innovate child health services to care for infants with birth defects.
- Conducting research on unknown causes of congenital anomalies and still births

Probable "lines of action" targeting the "pre-conception", "post-conception" and "neonatal" stages were suggested.



Some industries which could be involved in making an impact on the birth defects burden were suggested:

- Mechanical / Electronics : Devices
- Biotechnology : Diagnostics
- Health Care
- Pharmaceutical
- Nutraceutical
- ICT

The next talk was delivered by Dr. Anita Kar on "Public health perspective: Overview of disease burden of birth defects in India" which highlighted the lack of public health services for birth

defects in the Indian scenario. She also pointed out the confusion in the terminologies related to birth defects and defined birth defects as the defect in structure, function or metabolism, which presents at birth and results in physical or mental disability or an incurable medical condition. Birth defects include congenital anomalies, chromosomal disorders and single gene disorders. These are considered to be low prevalence conditions and are wholly or partly genetic. Birth defects could be accounted for in spontaneous abortions and stillbirths, children (and adults) with impairment in cognition, hearing, speech, vision and locomotor disability and children (and adults) with life-long medical conditions arising due to single gene disorders.

She highlighted the fact that birth defects are unrecognized medical conditions due to lack of data. There numerous genetic studies which are however, restricted to the laboratory. There are no field trials no studies on the cost-benefit analysis, no data on number of cases, no studies on needs of patients and practitioners.

To understand the exact needs of these patients, needs assessment studies need to be done, which involve exercises like data mining, as collection of data is a major problem in this field.
To give a glimpse of the public health burden of birth defects, she highlighted that nearly 32% neonatal deaths in the South-East Asian region are due to birth defects. In India alone, near 76,000 deaths per year are occurring due to birth defects.

Further Dr. Kar summarized the burden of congenital anomalies in India and its impact on child mortality. In South East Asian (SEA) Region, according to the Lancet study by Liu *et al*, the burden is 3/1000. The estimated neonatal mortality due to congenital anomalies in the SEA region is 37%. In India, 2 to 3 % births are due to congenital anomalies, giving 540,000 to 810,000 affected children with congenital anomalies. She also presented the estimates of the incidence and prevalence of specific birth defects in India which includes congenital heart disease in the first place for disease-specific annual live births, followed by neural tube defects, Down's syndrome, orofacial clefts, sickle cell disease, rhesus D haemolytic disease of the newborn, congenital hypothyroidism and thalassaemias. Data regarding type of disability

associated with birth defects indicates that 84% mental retardation as the disability, followed by mental illness, hearing, speech, locomotor, visual and other disabilities. According to one estimate in 2002 there are 5.5 million disabled due to birth defects.

Referring to the magnitude of genetic disorders in the Indian population, Dr. Kar used the model of haemophilia, where an adult patient with this disorder is estimated to incur Rs.3,00,000/- towards treatment costs, annually. Other impacts of the disease include progressive disability, loss of school/work days and physical and psychosocial trauma to the patients and the care givers, mostly mothers of patients. India harbours nearly 10% of the global haemophilia population and in the next 5 years, the number of these patients would cross 1 lakh.

The reason behind being concerned about the public health impact of birth defects is epidemiological transition where there is replacement of communicable diseases by non-communicable diseases due to expansion of public health programmes. In a developing country like India, due to the epidemiological transition, there is decrease in mortality, increase in life expectancy, decrease in infectious diseases, mortality in the 0-4 year age group has also decreased; there is increased mortality in adult and middle ages, increase in morbidity and mortality due to infectious diseases in the older ages, increased burden due to the onset of non-communicable diseases in younger ages. Causes of neonatal deaths are Low birth weight, preterm births, neonatal sepsis and intrapartum causes like birth asphyxia and birth trauma. Neonatal mortality rate is 33/1000 live births (ranges from 19 in urban to 36 in rural areas) and early neonatal mortality rate is 25/1000 live births (ranges from 28 in rural areas to 15 in urban areas).

The prevention of birth defects is the mainstay of the action against birth defects. These could be through preconception and antenatal interventions: These include

- Nutritional interventions: Macronutrients and micronutrients like iron, folate, iodine
- Lifestyle: Addictions and non-prescription therapeutic drug use

- Maternal health: Bad obstetric history, diabetes, TB, epilepsy
- Limiting exposures: Environmental, Infectious

Action suggestions:

Needs	Actions suggested
Identifying individuals who may benefit from genetic services	Screening tools for suspecting a case; Rapid detection kits for identification of the condition
Recognizing historical and physical features of common genetic conditions	Education for medical professionals
Monitoring the health of individuals with a genetic disorder	Education for medical professionals
Providing basic genetics information to patients and families	Education for families
Providing a medical home	A set of comprehensive care services
Recognizing the special psychosocial issues	Developing psychosocial support services
Possessing knowledge of how to access the full range of genetics services from which patients might benefit	Creating lists of available services
Appropriately referring patients	Informing doctors about these services
Facilitating the use of genetics services	Generating referral lists

This includes advising referred patients or relatives (usually parents) on the risk of an nature of the disorder, the probability of developing or transmitting it, and the options open to them in management and in making reproductive decisions.

- Genetic diagnosis (the estimation of risk where linkage analysis can be used and mutation testing) and
- Psycho-social support

However, for congenital anomalies no prenatal tests can be designed.

Dr. Kar's talk thus highlighted the public health perspectives of birth defects and what could be the possible points of intervention to bring down the burden of birth defects in India.

The next talk was by Dr. Manisha Doiphode on, "Prenatal diagnostic test: What are the limitations?" These diagnostic tests are of two types: Non-invasive (ultrasound, antenatal screening and Maternal blood for fetal DNA – MBFD test) and invasive procedures (amniocentesis and chorionic villus biopsy). The limitations of all these tests were highlighted, so as to bring forth the specific needs for technological innovations.

Limitations of some diagnostic methods:

Method	Limitations
Ultrasonography	⦿ Not 100% accurate: Operator dependant, hence manual operated and dependent on knowledge and training skills of the operator ⦿ Technology is expensive, there are different machines for different purposes and the resolution is variable ⦿ Structural anomalies may not be picked up, due to wrong position of the fetus or due to twin pregnancies ⦿ Organ functioning cannot be analyzed
Antenatal screening	⦿ Test is not a diagnostic, it gives "risk" ⦿ Dependant on maternal factors. Eg: hypothyroidism may give false positive results ⦿ Being software dependant, there could be technical errors, errors in hormone estimations, data entry errors, errors in ultrasound ⦿ Required accreditation from Fetal Medicine Foundation, UK
Maternal Blood Fetal DNA (MBFD)	⦿ Samples sent out of India for analysis. Hence delays in reporting time ⦿ QF – PCR available only for chromosomes 13,18,21,X,Y. This technique is not available for analysis of other autosomes. ⦿ It is extremely expensive procedure.

Amniocentesis and Chorionic Villus Biopsy	⦿ Invasive tests ⦿ Only skilled personnel can conduct these tests ⦿ Available only at approved centers, hence access limited ⦿ Stringent time limit during pregnancy term to conduct these tests according to PCPNDT Act. Otherwise tests become illegal and are punishable. ⦿ Risk of miscarriage ⦿ Problem of maternal cell contamination, may not give 100% accurate results for molecular diagnosis ⦿ Costly ⦿ In karyotype analysis, microdeletions or point mutations may be missed out ⦿ In FIST technique, few chromosomes are studied, not all genetic conditions covered ⦿ In linkage analysis method, 100% diagnosis may not be possible due to lack of informativity of the marker used

Some focus areas highlighted were like requirement of training of professionals in counseling, ultrasound, invasive tests and fetal therapy. There was a gap in the laboratory methods available and lack of commercial availability for these methods, the equipments required are expensive making it unaffordable to the end user or the patient. Stringency of the PCPNDT act, maintenance of records to be submitted, training and registration required under this act as deterrents for establishment of such services and hence these services are available at a select few centers. There is also a lack of knowledge amongst the medical fraternity and the patient community regarding the availability of these tests. She also highlighted the need of support groups, financial support and resource allocation for caring the affected child.

In his talk on “Diagnosis And Management Of Genetic Disorders And Congenital Anomalies: The Indian Scenario”, Dr. Gambhir pointed out that diagnosis of genetic disorders is not given due

importance in clinical practice in India because of the misconceptions that genetic disorders are rare, they are not important from the burden point of view and they too complicated to be managed. However, nearly 17% of all conceptions are genetic in origin. Every 40 seconds, a child with a genetic disorder is born world-wide. Birth defects include congenital malformations and congenital anomalies. Congenital anomalies are classified as: malformation, deformation, disruption, dysplasia, sequence and association depending on the timing of the insult, etiology, risk of recurrence and prevention.

Malformation can be defined as a primary structural defect in an organ caused by inherent abnormality in development. In this, the normal development gets arrested or misdirected and it shows polygenic multifactorial inheritance. In disruption there is interruption in a normally formed organ. Due to some external factor there is interruption in the blood supply. Since it is mechanical in occurrence it is least likely to recur. In deformation, normally developing organ is distorted by mechanical forces. It could be intrinsic factors resulting in muscle weakness or extrinsic factors such as overcrowding, multiple pregnancies, uterine malformation, oligohydramnios (deficiency of amniotic fluid) and tight abdominal musculature. Recurrence of deformation depends on the cause of occurrence. For example, if deformation is caused due to oligohydramnios, then these are non-recurrent. Dysplasia is disorder in organisation of cells. It may show single gene or multiple gene involvement. If it is polygenic, the risk of recurrence is 3%. It shows Mendelian inheritance pattern.

Some examples of birth defects were discussed, where the preventing strategies included peri-conceptional vitamin supplementation amongst others. Effective public health programs for birth defects also need to be in place, where several countries like US, UK, Chile and China have shown decreased incidence of neural tube defects by folic acid fortification interventions.

At the clinical level, while approaching a genetic problem, more time must be spent on taking clinical and family history. The approach should be pre-test counseling, diagnosis, post-test counseling and then followed by prenatal screening, prenatal diagnosis and newborn screening

for the condition in question. Some limitations in diagnosis and management were also highlighted. Very few laboratories are offering enzyme analysis. Also molecular diagnosis for genetic disorders should be available commercially and at affordable prices.

Screening can also be the approach for some birth defects. Screening is the search for unrecognized disease by means of a rapidly applied test. Screening is used to detect early and asymptomatic disease in apparently healthy individuals. Which diseases should be screened? The disease should be a public health problem. Effective intervention should be available. The disease should have an asymptomatic stage. The outcome should be significantly better if treatment is started in asymptomatic stage.

Some disorders for which screening is possible in neonatal period are: congenital hypothyroidism, congenital adrenal hyperplasia, galactosemia, phenylketonuria, Glucose-6-phosphate deficiency, biotinidase deficiency, Medium-chain acyl-CoA dehydrogenase (MCAD) deficiency, Maple syrup urine disease (MSUD), homocystinuria, aminoacidopathies, hemoglobinopathies and cystic fibrosis.

Some needs in this area were: Surveillance of Birth Defects, Newborn and Prenatal screening, Diagnostic services, Baseline and representative Indian data, Indigenization of majority of diagnostic platforms which are mostly imported, Indigenization of several molecular investigations which are still outsourced. Other requirements were education or creating awareness amongst general public, medical fraternity, basic sciences fraternity and need for interdisciplinary coordination.

Dr. Bhan's talk on, “Ethical and regulatory issues in genetic testing” focussed in detail on the nuances of the PCPNDT Act. He also discussed about issues related to the deployment of new tests such as the ethics behind standardizing new tests, availability, accessibility and affordability of these tests. Also he highlighted the need to return the research results to the study participants as a part of ethical obligation on part of the researcher. There is also a need

to understand the implications of genetic testing not only on the patient, but also on the immediate and extended family. The new marketing strategies related to direct to consumer marketing for genetic testing, though, has improved access to these tests, has also brought to the front the need for doing pre and post test counseling by trained genetic counselors. As more and more companies get involved in starting newer diagnostics, more attention needs to be given to the ethical and regulatory implications of these diagnostic tests. He discussed several case studies to elaborate these issues in the Indian scenario.

The panel discussion conducted by Dr. Premnath (VP), with Dr. Gambhir (PG), Dr. Doiphode (MD), Dr. Bhan (AB), and Dr. Sunil Lohade (SL: Paediatrician trained in haemophilia care) as the panel members. The point of discussion was the priority areas for each of the panelists which need to be addressed in this field of birth defects in the Indian scenario. PG highlighted the need for a basic monitoring system which could detect all the birth defects in India. Initially it could be a small scale system starting with a small part of Pune city and then could be scaled up to cover the wider population. The current birth defects registry is a hospital-based system, which is not documenting all the cases of birth defects. According to PG, unless such a robust monitoring and surveillance system is in place, baseline data about birth defects would not be available. These data would them also help in determining the causative factors for various genetic conditions.

Was any such surveillance system in place and how did it work? PG told that a national level surveillance program is in place in UK. MD further added that the data in such a surveillance program could give out interesting genotype phenotype correlations and several other interesting interactions.

For which birth defects such a program should be initiated in India? Neural Tube Defects can be taken as a priority, as it is commonly occurring clinically. Also rpimary prevention for NTDs is possible. The genes involved have been largely studied abroad, but not in India. So, family studies for NTDs can be initiated based on this surveillance data.

MD gave the details about the Birth Defects Registry of India (BDRI) started by Dr. Suresh, where several hospitals have registered with the BDRI and regularly report to this registry any birth defects encountered in their hospitals. This will however, skew the data, as only those hospitals which are registered with BDRI report such births. This is the limitation of this registry.

AB brought forth the point that screening would be a huge and challenging task due to the sheer numbers in the Indian population. In India the situation is very unique, where there is lack of health care facilities, amongst the available health care facilities available, there are levels like primary, secondary and tertiary. Also there is a difference between the urban and rural areas. During monitoring and surveillance there is an issue about loss to follow up. Hence, innovations are required in builiding robust documentation systems which understands all these complexities and ensures that data is not lost to follow up.

VP stated that there are several dimensions to these data. We need to understand the mechanisms to create data, there are institutional mechanisms involved, data mining is another issue and how to manage such a huge data. Since these data is related to health aspects there a several legal, ethical and regulatory issues involved. But to get these data related to birth defects, does India have good screening and diagnostic tools, is the detection fool proof was the question.

AB admitted that there are several good diagnostic tests available, but turn around time (TAT) is too high and hence there is loss to follow up. So other innovations could be to reduce this TAT. Also indegenizing these tests, instead of out sourcing would bring down the price and would make these tests affordable to the Indian population. AB also stated that as there is no cure for genetic conditions, people do not want to know the diagnosis. However, if some clinical intervention is possible for a genetic condition, then may be there would be a demand for these diagnostics. However, the role of clinician here in convincing the family is very crucial.

PG told that it is very important to deliver the services to the entire family and not just the affected individual. The community should be involved in any effort related to initiating any registry to nake it workable. Also this should be complemented by creating public awareness about the need for such services.

When SL's views were asked from the point of view of quality fo life of haemophilia patients, he expressed the need for manufacturing cheaper recombinant drugs for managing haemophilia bleeds, as the need of the hour. Another problem is identification and documentation of patients. An entrepreneur in the audience, was working on a Cloud based technology which could be harnessed to address this problem. This technology could be used for creating and capturing data on haemophilia patients. Also, information could be collected in the regional language via mobile SMS based applications.

MD pushed the need for creating awareness in the general the community and doctors via media. If simple symptoms identifying the common birth defects could be flashed in the media, probably several more patients could be picked up. The doctor can upload the data regarding these patients using this cloud technology, patient gets quick diagnosis and is thus benefitted. At the grassroot levels ASHAs or dais conducting deliveries could collect these data which is then analyzed by the expert.

However, the question is what after diagnosis. Here the need for coordination of resources was discussed. Fruitful networks amogst doctors, patients, pharma companies, suppliers and other stakeholders needs to be established. In all these networks ethical and regulatory aspects have to be in place. To improve awareness, clear and positive messages need to be given.

VP asked other than need for data which could be IT-based innovation, what could be the other innovations? Are innovations in care management, school frameworks, physiotherapy, support group formation, nutritional interventions required. PG told that folic acid and B12 intervention pre-conceptionally is known to reduce NTDs and congenital heart defects. So innovations

related to food fortification can be undertaken. AB highlighted that malnutrition leading to low birth weight babies resulting in babies with developmental problems. To tackle malnutrition, people have tried packaged food, which has not been very successful, as it is not acceptable to the community. So how to offer food in an acceptable manner needs inter-sectoral, inter-ministry collaboration.

SL also proposed the need for innovation in transfusion related problems for haemophilia patients, who are exposed to the risk of HIV and HBV. Also to maintain the hemoglobin levels of haemophilia patients, they are given iron folic acid tablets regularly.

VP proposed the innovations related to devices and diagnostics related to blood transfusion. SL told that from one blood sample 10 other products could be made and given to patients according to the need. Hence there is a need for making such blood products according to the need of the patients.

The panel discussion ended by summary of the innovations related to antenatal and newborn screening, prenatal diagnostics with low TAT, cost effective tests which will improve take up for these tests and thus eventually result in bringing down the burden of birth defects in India.

The workshop concluded by a Skype call with Sonia Gandhi the Program Manager for the SPARSH scheme from BIRAC-DBT. A call for proposals has been given for “Reproductive, Maternal, Newborn and Child Health”. Further details are available on the BIRAC website.

Appendix 1

Workshop Outline		
0915-0930	Registration	
0930-0945	Welcome to Venture Center BioIncubator. Introduction to the workshop.	Premnath V Pradnya Aradhye
0945-1000	Quick Overview: Congenital anomalies in India – Current status, key problems and needs, and opportunities to address the issues	Mugdha Lele
1000-1045	Public health perspective: Overview of disease burden of birth defects in India:	Anita Kar
1045-1100	Networking and Tea	
1100-1145	Prenatal diagnostic tests: What are the limitations?	Manisha Doiphode
1145-1230	Diagnosis and management of genetic abnormalities and congenital anomalies: The Indian scenario	Prakash Gambhir
1230-1330	Lunch	
1330-1500	Panel discussion • Key issues related to Congenital anomalies. Priority areas. • Genetic Counseling: Problems and limitations in the clinical scenario in India • Current approaches to addressing the issues and challenges faced • Potential areas for technology intervention. Other approaches to addressing the issues.	**Moderator:** Premnath V **Panelists:** Anant Bhan, Manisha Doiphode, Prakash Gambhir, Sunil Lohade **Rapporteur:** Mugdha Lele
1500-1545	Ethical and regulatory issues in genetic testing	Anant Bhan

1545-1615	An overview of emerging funding opportunities: SPARSH from BIRAC focused on Reproductive, Maternal, Newborn and Child Health (Deadline: 30 Sept 2014)	Pradnya Aradhye
1615-1700	Q&A for SPARSH funding scheme (over SKYPE)	BIRAC: Sonia Gandhi
1700-1730	Closing comments and closure of the event	

 PRADNYA ARADHYE	Pradnya Aradhye is currently Associate, Bioincubator, Venture Center. She has done her M.Tech in Biological Sciences and Bioengineering from IIT Kanpur. Currently she is handling all BioIncubator activites at Venture Center. She is responsible for creating a pipeline of potential and signing-up incubates for the Bioincubator. Contribute to building scientific support systems and resources for VC incubates including specific expertise. Discussions with scientists to understand their competencies.
 ANANT BHAN	Dr Anant Bhan is an independent bioethics and global health researcher. He is a medical doctor by profession and completed his medicine degree from Bangalore Medical College and Research Institute. Further he did PGDMLE, Medical Law and Ethics from National Law School India University. He went to University of Toronto to do MHSc in Bioethics. He has various research publications on public health and bioethics to his credit.
 MANISHA DOIPHODE	Dr. Manisha Doiphode is a consultant in prenatal genetics and fetal medicine. She has completed her postgraduate diploma from University College London. She has been practising as genetic counsellor and fetal medicine consultant since 2005. She is visiting faculty at Pune University and Maharashtra University of Health Sciences, Nashik. She has interest in research and is involved in clinical and community research projects. She has publications in peer reviewed journals to her credit.
 PRAKASH GAMBHIR	Dr. Prakash Gambhir is presently Associate professor of paediatrics and Genetic Clinic in charge at B.J.Medical college and Sassoon General Hospitals Pune. He has also his own well equipped Genetics Laboratory and Genetic Clinic in Pune. He has graduated with Gold Medal for M.D Pediatrics.He has worked in various ICMR DBT and DST task force projects in Human Genetics and also obtained training in genetics in Germany. He has published more than 25 papers in reputed international and national journals. These include many rare case syndromes as well as novel hypothesis. His current research interests include Prenatal screening, Newborn screening, fetal dysmorphology and rare disorders.
SONIA GANDHI 	Sonia Gandhi is currently assisting the Investment group of BIRAC, New Delhi as Project Manager and is responsible for proper functioning of the grants management system, ensuring compliance to regulations, evaluation and management of projects as per guidelines. Previous to this she worked with BIRAP as Program Manager for Healthcare with responsibility of Techno-commercial guidance and support for projects in identified areas of Healthcare Industry. Prior to this, she was in Quality Management Systems of Reliance Life Sciences and is involved in ensuring the compliance of various processes of the product development to applicable regulatory guidelines. She is double Masters in Biotechnology and Quality Management and also a Certified Quality Management Professional from Birla Institute of Technology and Science and Reliance Institute of Life Sciences. She also visited the EMA, MHRA, HTA and NICE offices at UK to understand the regulatory scenario impacting Medical Devices, Clinical

	Trials, Vaccines and Stem Cells development.
 ANITA KAR	Dr Anita Kar is Director at Institute of Health Sciences at University of Pune. She has done her PhD from University of Calcutta in Zoology and Cytogenetics and completed her Post-doctoral training at University of Zoology, Massachusetts Institute of Technology, USA. She has numerous national and international publications and academic and research awards to her credit. Dr Kar is member of various academic and research committees for Public Health Curriculum Development, Advisory boards, Institutional Bio-safety Committee and Ethics Committees.
 MUGDHA LELE	Dr Mugdha Lele is a Scientific Advisor at Venture Center, NCL Innovation Park, Pune. She was previously an Assistant Professor in Dept of Genetics, Immunology and Biochemistry at Maharashtra University of Health Sciences. Her research work is related to epidemiological profiling and development of molecular diagnostics for Genetic disorders. She has a few national and international publications to her credit.
 SUNIL LOHADE	Dr. Sunil Lohade is a Pediatrician at Lohade Hospital, Chinchwad, Pune. He is trained as a haemophilia specialist at the Royal Free hospital in UK and is associated with the Haemophilia Federation of India, Pune Chapter from its inception in 1991. Currently he is Vice-president of the Pune Chapter.
 PREMNATH VENUGOPALAN	Dr. V. Premnath Founding Director – Venture Center and Head, NCL Innovations. He holds a B.Tech. from the Indian Institute of Technology - Bombay and a Ph.D. from the Massachusetts Institute of Technology, USA. He has also been a Chevening Technology Enterprise Fellow with the Centre for Scientific Enterprises, London Business School and Cambridge University, UK. He brings with him considerable experience in technology development and commercialization, working with start-up companies (in Cambridge-UK and India) and engaging with large corporations on research and consulting projects as project leader.

Appendix 2

Photos

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