Birth defects- Act today!

Recommendations of the first national birth defects meeting PPHC2013 on Pregnancy loss, birth defects and genetic disorders in India: Epidemiology, social costs, health systems needs

Birth defects ---- an unrecognized public health problem in India

Birth defects represent a group of neglected childhood conditions where children are born with a physical or mental disability, or with a chronic medical condition like the hemoglobinopathies or bleeding disorders. Caused by genetic and environmental interactions, single gene disorders or chromosomal anomalies, birth defects have high social costs, as there are no government health services for patients affected with these conditions in India. As patients need lifelong care, families incur repeated out of pocket expenditure, which is frequently catastrophic in nature. Patients remain poorly treated, resulting in poor quality of life and premature mortality. Birth defects raise issues of equity, as the consequences of childhood disability are most profound amongst the poorest strata of society. Birth defects also have gender implications, as mothers are left to care for the disabled child with little knowledge or support.

Several reasons account for the low public health priority of birth defects in India and other developing countries. These include the low prevalence of birth defects, the genetic etiology that contributes to the chronic nature of these conditions and their resource intensive management.

India may harbour the largest global burden of birth defects

Globally, birth defects affect 2 to 3% of births. With 27 million births in 2010, 500 000 to 800 000 pregnancies may have been affected annually in India each year. Birth defects are responsible for pregnancy loss and early neonatal mortality. Their impact on early neonatal mortality indicators are unknown.

Pune Public Health Conference 2013 (PPHC2013)--- the first national birth defects Conference

The PPHC 2013, organized by the School of Health Sciences of the University of Pune, was convened to understand the available data on birth defects in India, risk factors for birth defects, and to deliberate on how a birth defects programme could be organized within existing health priorities of the country. The PPHC2013 also took the opportunity to sensitize a larger audience on birth defects, including key opinion leaders from the field of public health in Maharashtra. A few days before the Conference, the Rashtriya Bal Swastha Karyakram (RBSK) was launched, which added a new dimension to the discussion on birth defects prevention and care in India.

BRIEF REPORT ON PRESENTATIONS (11TH AND 12TH FEBRUARY,2013)

- Birth defects may affect 500 000 to 800 000 pregnancies each year in India. This estimate was based on global neonatal mortality estimates. The lack of data on the epidemiology, social costs and the health systems needs for addressing birth defects was described. The opportunity to use preconception care (PCC) for improving birth outcome, for reducing birth defects and as a point of entry for genetic services was discussed (Anita Kar).

- The framework for a birth defects programme in countries of the South East Asian region(Dr Neena Raina), and the current status of maternal and child health in India was discussed (Dr B S Garg).e

- The National Birth Defects Surveillance in the US was used an illustrative example for birth defects surveillance (Dr VijayaKancherla). The potential of the Mother Child Tracking System (Dr P PDoke) for monitoring congenital anomalies was presented.

- Indian data on risk factors for birth defects and pregnancy loss was discussed, which included adolescent pregnancy (Dr Shobha Rao, Dr Ashok Dyalchand,), micronutrient deficiencies (Dr Prema Ramachandran, Sadhana Bhagwat) and smokeless tobacco use (Dr P C Gupta). Role of nutrients in fetal programming was presented (Kumaran Kalyanaraman)

- Example of birth defects registration in Pune city (Dr Koumudi Godbole), magnitude and impact of genetic disorders (Dr R S Balgir, Dr Ajit Gorakshakar) and opportunities and challenges of newborn screening were presented (Dr Mamta Muranjan) .

- The opportunity for care for children with birth defects through the Rashtriya Bal Swastha Karyakram (RBSK) a new child health initiative of the Government of India was presented by Dr Manpreet Khurmi. The need for psychosocial support for parents (Supriya Phadnis) and the role of physiotherapists in addressing disability through public health services (Dr Kavitha Raja) was discussed.

- Poster sessions included four categories of presentations: pregnancy outcome, risk factors, genetic disorders and rehabilitation and care.

RECOMMENDATIONS OF THE CONFERENCE

It was recognized that

- Birth defects cause interminable suffering for patients and their families due to the disabling nature of these conditions and the need for lifelong medical or supportive care for patients.

- Repeated out of pocket and catastrophic expenditures for management of morbidity and disability drive families into impoverishment. Treatment is most often sub-optimal, leading to a poor quality of life.

- Low medical familiarity lead to patients remaining undiagnosed, further compromising the quality of life of the patients and the family members.

- Birth defects raise issues of equity, as the impact of caring for a child with disability is more severe on families from the lowest socio-economic strata.

- Birth defects have gendered consequences, as mothers may be deserted, stigmatized or blamed for the birth of the child. There is no formal system to educate caregivers of children with disability. Mothers endure the brunt of care giving, with little formal knowledge on management of the condition.

- Birth defects raise social issues, as often a child with a disability or chronic illness may result in stigmatization of the family. Social life of the family may be affected.

- Birth defects may influence maternal and child health indicators, as many birth defects result in pregnancy loss and early neonatal mortality.

- Globally, India has the largest annual birth cohort, suggesting that the largest number of children with birth defects in the world may be born each year in India.

The Conference deliberations lead to the following recommendations:

A. Development of a national policy on birth defects as a component of the NRHM RCH II programme

- In India, birth defects may affect 500 000 to 800 000 live births each year.
- Global estimates suggest that neonatal mortality due to congenital anomalies is 3.2 per 1000 live births in India, accounting for at least 72 000 neonatal deaths each year.

- In light of these facts, a national programme for the prevention and control of birth defects should be developed as a component of the NRHM RCH II programme.

- The birth defects program should include components of prevention, care, surveillance, advocacy, research and training.

- The birth defects programme should be incorporated and placed within existing maternal, neonatal and child morbidity and mortality reduction strategies.

B. Prevention

- As birth defects are disparate medical conditions whose management is multi-disciplinary and resource intensive, a programme for the prevention of birth defects should be immediately launched through a preconception care package.
- Preconception care (PCC) should be included as a part of the NRHM-RCHII programme.
- The PCC package can include
- **Pre-conception care services:**
    - Knowledge on healthy pregnancy habits and practices , folate supplementation: during adolescence and peri-conceptional period, correction of iron deficiency, ensuring optimum weight of mothers, family planning advice to prevent pregnancies in adolescent women and women of advanced age, rubella immunization wherever possible, or awareness about reporting to Medical Officer in case of rash during pregnancy , preventing consanguineous marriages, NCD (diabetes and obesity) prevention through healthy lifestyles promotion, screening and treatment

- **Reinforcing and improving antenatal care services:**
    - Advice on medications during first trimester and the risks of self-prescription, syphilis / HIV screening and management, prevention of exposure to tobacco and alcohol,prevention and management of diabetes and obesity during pregnancy, management of epilepsy, other maternal illness during pregnancy, antenatal screening and prenatal tests for BD
- **Improved perinatal care:**
    - Education of parents, family members on neonatal care, asphyxia prevention and management, neonatal screening for BD
- As birth defects and pregnancy loss share most risk factors, PCC will not only reduce birth defects, but will improve pregnancy outcome through reduction of pregnancy loss, stillbirths and early neonatal deaths.
- PCC is also a point for introduction of genetic services.
- The prevention package can be delivered by appropriately trained ASHA/ANM to newly married couples, and to women planning a next pregnancy. Family members and the community can be involved through public programmes on Village Health and Nutrition days.

C. Care

- The District Early Intervention Centres proposed under the RBSK should be established and functionalized on priority basis.
- Development of appropriate intervention should be immediately initiated, which should take into consideration the sensitivity, confidentiality and support needed for referral of affected families. .
- A pilot centre could be rolled out and tested.

- It is significant to keep in mind that people with disability have health access needs that are different from normal people, and that these concerns are greater for marginalised populations like women and children with disabilities. Children with disabilities have an ever-changing profile of disability and needs. Community based rehabilitation workers who will perform disability related services on the lines of the existing ASHAs can be considered.

- In case of congenital conditions, the quality of life of parents may be significantly affected. There is need for developing culturally appropriate psychosocial support interventions in the local language for parents.
- Parent-patient organizations should be encouraged, supported and partnered into the operation of the DEICs for providing constant education, support and handholding of parents.

D. Surveillance

- Screening under the RashtriyaBalSwasthyaKaryaram (RBSK) should be initiated with immediate effect.
- The DEIC data needs to be documented with care, as the magnitude of the selected congenital conditions can be estimated from this data.
- Birth defects reporting should be incorporated into the Mother Child Tracking System so that the burden of these conditions on the public health system, including their impact on early neonatal mortality can be estimated.

E. Genetic disorders

- Prevention of genetic disorders through genetic counselling is urgently required as these conditions are extremely traumatic to patients and families.
- Considering that medical and biotechnological competencies are widely available in India, prevention services need to be immediately launched in order to mitigate the suffering.
- Well-established and organized community based organizations (CBOs) (like the thalassemia, haemophilia and other societies) can be partnered into the process, so that the responsibility of education and psychosocial support can be continued by these organizations.
- A national level brainstorming is need to identify the public health mechanism of providing genetic services through preconception care as a part of the NRHM RCH II programme

F. Increasing visibility and awareness on birth defects and their prevention

- Widespread dissemination of messages through print and visual media should be initiated on priority basis.
- Training teams for disseminating knowledge on birth defects to medical colleges, medical associations, and professional organizations of nutritionists, geneticists, physiotherapists, social workers and others.
- CBOs should be engaged in disseminating the message of PCC.

G. Research

- The role of research for the characterization of this newly emerging field of birth defects is extremely important.
- Research data is needed in the following areas
    - (a) epidemiology including estimation of the magnitude of selected disorders,
    - (b) social costs, which could include case studies of families, studies on gender, economic aspects, equity,
    - (c) risk factor prevalence studies, in order to identify prevention programmes for addressing these conditions.
    - (d) operational research studies to determine the utilization, health systems cost and infrastructural needs for operationalization of the DEICs.

H. Ethical and regulatory issues

- Birth defects will raise a number of issues relating to regulation and ethics.
- Context specific, sensitive counselling material has to be developed for the selected conditions. Staff has to be rigorously trained.
- A committee should be set up to periodically monitor the process of diagnosis and referral of affected families to the DEIC and the subsequent process of management.
- Legal and ethical issues such as detection of anomalies in women presenting after the legislated period of pregnancy termination has to be resolved.
- Prenatal sonography for detection of congenital anomalies has to be strictly regulated in light of the ongoing activities targeted towards prevention of sex-selective abortion.

More photos at
https://plus.google.com/photos/111186982595975795326/albums/5849555165253558145?authkey=CIC81t-ww-zOzQE

School of Health Sciences

University of Pune, Pune 411007, India

91-20-25691758

Contact : Dr Anita Kar, akar@unipune.ac.in

Translating knowledge, making science work