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RESEARCH ARTICLE

RISK FACTORS FOR CONGENITAL ANOMALY: ANALYSIS AT SBMCH.

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Abstract

Introduction:-

Congenital anomalies are a leading cause of infant death and disability and their incidence varies between ethnic group.. Congenital anomalies affect a remarkable proportion of newborn population and contribute significantly to the childhood mortality and hospital admissions . It is are a global health problem. Every year an estimated 7.9 million children are born with a serious birth defect, 3.3 million children (under five years) die from birth defects, and 3.2 million who survive may develop a disability later in the life . They are the leading cause of prenatal mortality and childhood morbidity and disability in many countries . The wide range of causes of birth defects means that a portfolio of prevention approaches is needed. The prevention of these disorders is available in 60% of cases . This needs however epidemiological information.

Prevalence studies of congenital anomalies are useful to establish baseline rates, to document changes over time, and to identify clues to the etiology. Many of developed countries monitor the prevalence of birth defects through registration or surveillance system of fetuses and infants. In addition, international organizations have been established to conduct worldwide surveillance and research into the occurrence and possible causes of congenital anomalies and to establish prevention strategies.

Congenital anomalies are the most common causes of death in children (1–59 months) in India. however, are mostly limited to particular type of defects . Without comprehensive data on congenital anomalies, it is difficult to evaluate possible teratogens and to implement effective prevention and care services. This information is also important for planning and performing antenatal screening for congenital anomalies, particularly in high risk populations. Nevertheless, published comprehensive data about the prevalence of birth defects are scarce in developing countries .The aim of this study was to determine the epidemiological features of congenital anomalies .

Material and Methods:-

We obtained questionnaire data from the mothers of children with one or more anomalies , a prospective birth cohort study of 60 babies and their families in which recruitment was undertaken between 2016 and 2017. Details of anomalies were prospectively reported to the study and we cross checked these details against medical records

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Disscussion:-

Definition:-

Congenital anomalies are also known as birth defects, congenital disorders or congenital malformations. Congenital anomalies can be defined as structural or functional anomalies (for example, metabolic disorders) that occur during intrauterine life and can be identified prenatally, at birth, or sometimes may only be detected later in infancy, such as hearing defects.

In simple terms, congenital refers to the existence at or before birth.

Congenital anomalies are important causes of infant and childhood deaths, chronic illness and disability. Through the resolution on birth defects of the Sitxty-third World Health Assembly (2010), Member States agreed to promote primary prevention and improve the health of children with congenital anomalies by:

1. Developing and strengthening registration and surveillance systems
2. Developing expertise and building capacity
3. Strengthening research and studies on etiology, diagnosis and prevention

Key facts:-

1. An estimated 303 000 newborns die within 4 weeks of birth every year, worldwide, due to congenital anomalies.
2. Congenital anomalies can contribute to long-term disability, which may have significant impacts on individuals, families, health-care systems, and societies.
3. The most common, severe congenital anomalies are heart defects, neural tube defects and Down syndrome.
4. Although congenital anomalies may be the result of one or more genetic, infectious, nutritional or environmental factors, it is often difficult to identify the exact causes.
5. Some congenital anomalies can be prevented. Vaccination, adequate intake of folic acid or iodine through fortification of staple foods or supplementation, and adequate antenatal care are just 3 examples of prevention methods.

Causes and risk factors:-

Although approximately 50% of all congenital anomalies cannot be linked to a specific cause, there are some known genetic, environmental and other causes or risk factors.

Genetic factors:-

Genes play an important role in many congenital anomalies. This might be through inherited genes that code for an anomaly, or resulting from sudden changes in genes known as mutations.

Consanguinity (when parents are related by blood) also increases the prevalence of rare genetic congenital anomalies and nearly doubles the risk for neonatal and childhood death, intellectual disability and other anomalies.

Some ethnic communities (such as Ashkenazi Jews or Finns) have a comparatively high prevalence of rare genetic mutations such as Cystic Fibrosis and Haemophilia C.

Socioeconomic and demographic factors:-

Low-income may be an indirect determinant of congenital anomalies, with a higher frequency among resource-constrained families and countries. It is estimated that about 94% of severe congenital anomalies occur in low- and middle-income countries. An indirect determinant, this higher risk relates to a possible lack of access to sufficient, nutritious foods by pregnant women, an increased exposure to agents or factors such as infection and alcohol, or poorer access to healthcare and screening.

Maternal age is also a risk factor for abnormal intrauterine fetal development. Advanced maternal age increases the risk of chromosomal abnormalities, including Down syndrome.

Environmental factors:-

Maternal exposure to certain pesticides and other chemicals, as well as certain medications, alcohol, tobacco and radiation during pregnancy, may increase the risk of having a fetus or neonate affected by congenital anomalies.

Working or living near, or in, waste sites, smelters or mines may also be a risk factor, particularly if the mother is exposed to other environmental risk factors or nutritional deficiencies.

Maternal nutritional status:-
Maternal folate insufficiency increases the risk of having a baby with a neural tube defect while excessive vitamin A intake may affect the normal development of an embryo or fetus.

Prevention:-
Preventive public health measures work to decrease the frequency of certain congenital anomalies through the removal of risk factors or the reinforcement of protective factors. Important interventions and efforts include:

1. ensuring adolescent girls and mothers have a healthy diet including a wide variety of vegetables and fruit, and maintain a healthy weight;
2. ensuring an adequate dietary intake of vitamins and minerals, and particularly folic acid in adolescent girls and mothers;
3. ensuring mothers avoid harmful substances, particularly alcohol and tobacco;
4. avoidance of travel by pregnant women (and sometimes women of child-bearing age) to regions experiencing outbreaks of infections known to be associated with congenital anomalies;
5. reducing or eliminating environmental exposure to hazardous substances (such as heavy metals or pesticides) during pregnancy;
6. controlling diabetes prior to and during pregnancy through counselling, weight management, diet and administration of insulin when required;
7. ensuring that any exposure of pregnant women to medications or medical radiation (such as imaging rays) is justified and based on careful health risk–benefit analysis;
8. vaccination, especially against the rubella virus, for children and women;
9. increasing and strengthening education of health staff and others involved in promoting prevention of congenital anomalies;
10. screening for infections, especially rubella, varicella, and syphilis, and consideration of treatment.

Detection:-
Health care before and around the time of conception (preconception and peri-conception) includes basic reproductive health practices, as well as medical genetic screening and counselling. Screening can be conducted during the 3 periods listed:

1. Preconception screening can be useful to identify those at risk for specific disorders or at risk of passing a disorder onto their children. Screening includes obtaining family histories and carrier screening, and is particularly valuable in countries where consanguineous marriage is common.
2. Per-conception screening: maternal characteristics may increase risk, and screening results should be used to offer appropriate care, according to risk. This may include screening for young or advanced maternal age, as well as screening for use of alcohol, tobacco or other risks. Ultrasound can be used to screen for Down syndrome and major structural abnormalities during the first trimester, and for severe fetal anomalies during the second trimester. Maternal blood can be screened for placental markers to aid in prediction of risk of chromosomal abnormalities or neural tube defects, or for free fetal DNA to screen for many chromosomal abnormalities. Diagnostic tests such as chorionic villus sampling and amniocentesis can be used to diagnose chromosomal abnormalities and infections in women at high risk.
3. Neonatal screening includes clinical examination and screening for disorders of the blood, metabolism and hormone production. Screening for deafness and heart defects, as well as early detection of congenital anomalies, can facilitate life-saving treatments and prevent progression towards some physical, intellectual, visual, or auditory disabilities. In some countries, babies are routinely screened for abnormalities of the thyroid or adrenal glands before discharge from the maternity unit.

Treatment and care:-
Many structural congenital anomalies can be corrected with paediatric surgery and early treatment can be administered to children with functional problems such as thalassaemia (inherited recessive blood disorders), sickle cell disorders, and congenital hypothyroidism (reduced function of the thyroid).

WHO response:-

The report accompanying the resolution of the Sixty-third World Health Assembly (2010) on congenital anomalies describes the basic components for creating a national programme for the surveillance, prevention and care of congenital anomalies before and after birth. It also recommends priorities for the international community to assist in establishing and strengthening these national programmes.

1. World Health Assembly report on birth defects
2. World Health Assembly resolution WHA63.17 on birth defects
3. Global Strategy for Women's, Children's and Adolescents' Health, 2016-2030

WHO develops normative tools, including guidelines and a global plan of action, to strengthen medical care and rehabilitation services to support the implementation of the United Nations Convention on the Rights of Persons with Disabilities. Similarly, WHO supports countries to integrate medical care and rehabilitation services into overall primary health care, supports the development of community-based rehabilitation programmes, and facilitates the strengthening of specialized rehabilitation centres and their links with community-based rehabilitation.

United Nations Convention on the Rights of Persons with Disabilities:-

The WHO Department of Public Health and Environment focuses on a number of activities, and defines interventions, to address the environmental and social determinants of child development. These include children's unique vulnerabilities to polluted indoor and outdoor air, contaminated water, lack of sanitation, toxicants, heavy metals, waste components and radiation; combined exposures with social, occupational and nutrition factors; and the settings in which children dwell (home, school).

The current Zika virus outbreaks and their association with an increase in microcephaly and other congenital malformations have raised great concern across the world, particularly in the Americas. In 2016, WHO declared a Public Health Emergency of International Concern .

Results:-

Among the 60 congenital babies ,Consanguinity is a major risk factor for 58 % of the congenital anomalies , the other contributing factor would be nutritional such as deficiency of folic acid ,which lead to major congenital anomalies such as neural tube defects, which accounts for 34% .Iatrogenic factors accounted for 8% of the anomaly . Most of the major congenital anomalies were found around second trimester in well informed and educated groups .10% of the congenital anomaly were found only in late second and early third trimesters. Neural tube defects were detected much earlier when compared to other anomalies .

Conclusion:-

Our findings will be valuable in health promotion and public health, and to those commissioning antenatal, paediatric, and clinical genetic services. Sensitive advice about the risks should be provided to communities at increased risk, and to couples in consanguineous unions, to assist in reproductive decision making.

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