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A Study to Assess the Effectiveness of Information Booklet on Knowledge Regarding Genetic Diseases and Congenital Anomalies Among the Antenatal Woman of Selected Community Area of Kanpur, UP

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ABSTRACT

The health of newborns and their mothers is significantly impacted by genetic illnesses and congenital abnormalities. Although there are numerous prenatal screening tests available, moms' awareness has been demonstrated to determine uptake. This study aims to assess pregnant women's knowledge about birth defects and the factors associated with their desire for prenatal screening. [1]

A genetic ailment is a disease that has one or more abnormalities in its genome. A chromosomal abnormality, a mutation in one gene (monogenic), several genes (polygenic), or both may be the cause. Even though polygenic disorders are the most common, the phrase is usually used when discussing problems with a single genetic basis, either in a gene or a chromosome. The relevant mutation can either be inherited from one or both parents who have the disorder (autosomal dominant inheritance) or it might occur spontaneously before the embryo develops (a de novo mutation). Congenital abnormalities are defined as defects that arise during intrauterine life and are either structural or functional. These conditions, which are often referred to as congenital malformations, birth abnormalities, or anomalies, develop during pregnancy and can be identified before, during, or after delivery.[2] Due to the extremely high rates of illness and mortality, they are connected with, genetic disorders and congenital malformations are identified as public health hazards.

KEYWORDS: *genetic illness and congenital abnormalities, pre natal screening, antenal women, autosomal dominant inheritance*

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Every year, some 303,000 babies worldwide die as a result of birth defects, and 7.9 million are born with severe defects. The probability of physical, cognitive, and social problems for the children who make it through childhood grows with age, putting a heavy financial, emotional, psychological, and social load on their families. Because birth abnormalities are associated with miscarriages, early births, and stillbirths, research suggests that they pose a significant obstacle to the accomplishment of global health goals. Environmental factors account for the remaining birth issues, while chromosome abnormalities and single gene mutations account for the majority of birth deformities with recognized causes. However, this accounts for less than 25% of birth defects. Frequently, the underlying reasons are not well understood. It has been demonstrated that major birth abnormalities are more common and burdensome in low- and middle-income nations, with a corresponding 95% mortality rate among affected children. Several studies have reported on different birth deformity rates in South-West Nigeria: 6.9% for external congenital birth abnormalities, 0.7% for cervical ribs, and 0.5% for orofacial clefts. Mothers' ignorance about genetic disorders and congenital abnormalities has been blamed for the significant disparities in occurrence between developing and industrialized nations, at least in part. As of right now, these distinctions don't seem to have altered all that much historically. This is in addition to the socioeconomic conditions of those living in low- and middle-income countries.[3]

Premarital genotype screening, early detection and treatment of pre-existing medical conditions prior to pregnancy, folic acid supplementation prior to pregnancy, iodination of food items, rubella vaccination, improving mothers' nutritional status, and premarital genotype screening are a few interventions that can help prevent certain genetic diseases and birth defects. Knowing when to seek medical help and being aware of all these things requires an educated mother.[4]

According to research, many women over 35 who give birth in underdeveloped nations do so in the absence of community information, medical genetic screening and diagnostic facilities, and adequate antenatal care. A higher quality of life for both mother and child is more likely when there is sufficient knowledge, which has been proven to prevent genetic disorders and congenital anomalies as well as the physical, financial, and social ramifications of birth defects.[5]Both genetic disorders and congenital anomalies are considered public health hazards because of their high rates of morbidity and mortality. Roughly 303,000 newborns worldwide are estimated to die as a result of birth defects, and 8.9 million babies are born with severe congenital abnormalities. The children who make it through childhood are more likely to experience physical, cognitive, and social difficulties throughout their lives, which can cause significant emotional, financial, psychological, and social challenges for their families. According to research, birth abnormalities are a significant obstacle to achieving global health goals since they are linked to preterm deliveries, miscarriages, and stillbirths. Less than 25% of birth defects are due to environmental factors, with chromosomal abnormalities or single gene mutations accounting for the bulk of birth problems. The majority of birth abnormalities have unclear causes.[6] Research indicates that in underdeveloped countries, a large proportion of women over 37 give birth without access to competent prenatal care, diagnostic services, medical genetic screening, or community education. It has been demonstrated that substantial knowledge can prevent genetic illnesses and congenital abnormalities, as well as prevent the negative social, medical, and economic effects of birth defects. It can also increase the likelihood that both the mother and the child will have a higher quality of life. The role that nonspeaking prospective mothers' inadequate awareness of these illnesses plays in their decline is consistent with the study's goals, which examine prospective mothers' level of knowledge on genetic diseases and congenital defects.[7]

Need of the study:

An established method for lowering the prevalence of genetic illnesses includes prenatal diagnostics for serious genetic abnormalities and congenital deformities with a bad prognosis, as well as termination of the pregnancy if the fetus is affected. At-risk families are typically discovered following the birth of an affected child or the discovery of a detailed family history, and they are then provided with appropriate genetic counseling, including the possibility of a prenatal diagnostic for the ailment in question. However, a lot of genetic problems strike members of families that have never had a kid or person with the condition. The prevention of common genetic illnesses is now possible thanks to screening tests, which are now available and being provided to all pregnant women. The conditions that require population-based preventative efforts that have a high frequency in India are beta-thalassemia, down syndrome, and Neural tube defects.[8]

In India, screening and prenatal diagnosis for these illnesses are available through both the public and private sectors, and obstetricians and primary care doctors are becoming more aware of the problem. However, there is frequently a dearth of knowledge on the proper exam, the right time to order the test, and experience for pre-test and post-test counseling. Numerous screening options are available, each with a different detection rate and price, which contributes to inconsistent advice on the best screening choice. The screening for Down syndrome is governed by national rules in several nations, including Canada. There is no population-based government prenatal screening initiative in India. To help obstetricians and primary care clinicians prevent the birth of an afflicted child, we must develop prenatal screening guidelines based on the need for effective, evidence-based screening.[9]

In India, population-based screening for Down syndrome, beta-thalassemia, and NTDs is essential, and there are sufficient published statistics on their prevalence. Every year in India, around 21,400 children are born with Down syndrome, 9000 with beta-thalassemia, and 5200 with sickle cell disease6,7. However, it is difficult to adhere to a single standard screening protocol8 because to the variety of screening alternatives, variation in economic status, and heterogeneity of the widely used medical services in the nation. This page aims to teach readers about the wide range of screening tests that are available and the factors to be taken into account when selecting the right test, as well as information to help physicians decide whether to give the screening test to families.[10]

A developing nation like India faces a significant burden in managing uncommon diseases given the estimated 70 million individuals who suffer from them, the majority of whom go misdiagnosed. Unknown in India is the precise socioeconomic burden brought on by uncommon genetic illnesses. Interestingly, despite an overestimated prevalence caused by decreased case reporting, the societal effects of hemophilia have been adequately documented. According to other studies, government initiatives can lower patients' out-of-pocket expenses.

India has few resources dedicated to treating or understanding uncommon diseases, despite the fact that over 70 million people are affected by them. The Indian Rare Disease Registry was established by the Indian Council of Medical Research (ICMR) in recent years as a step towards bridging the gap between people with rare genetic illnesses and healthcare professionals. The register serves as a national data center for information on people with rare diseases.[11]

The overall prevalence of serious congenital abnormalities among 1822 births was 230.51 (170.99-310.11) per 10 000 births. The most frequently reported anomaly in the group was congenital heart defects, which had a prevalence of 65.86 (37.72-114.77) per 10,000 babies.

Down syndrome, thalassemia, sickle cell anemia, cystic fibrosis, and Tay-Sachs syndrome are a few of the genetic conditions that are frequently found in India. Trisomy 21 is another name for Down syndrome, a chromosomal condition in which an extra copy of chromosome 21 results from faulty cell division.[12]

Genetic illnesses and congenital abnormalities affect 2%–5% of all live births. In industrialized nations, these conditions can also account for up to 30% of pediatric hospital admissions, 50% of infant deaths, and up to 40% of pediatric hospital admissions. Additionally, these conditions affect roughly 60% of deaths among children under five. The meager demographic data currently available from several nations in the EMR suggests that, contrary to common opinion, congenital abnormalities and genetically determined illnesses are at least as important in the Region as they are in more industrialized parts of the world.[13]

Genetic and congenital diseases are major causes of parent and newborn death in the majority of the Region's countries. In terms of infant mortality, congenital anomalies currently rank top in the United Arab Emirates (UAE) and second in Bahrain, Kuwait, Oman, and Qatar. Reports from Mumbai, India state that in two hospitals, congenital abnormalities accounted for 25–35% of parent deaths. Furthermore, based on the available regional data on the involvement of congenital and genetic variables to the causes of disabilities, mental retardation, deafness, and blindness appear to have a major genetic component.[15]

One of the main causes of congenital malformations and mental retirement is chromosomal aberrations. The probability of having a child with Down syndrome increases with maternal age, rising from 1 in 600 births for moms under 30 to 1 in 50 births for moms over 40. In certain countries within the Region, mothers over 40 give birth to 50% of children with Down syndrome, based on data attainable through EMRs. There have been reports of differing Down syndrome prevalence rates among live births in the EMR, ranging from 2.5 per 1000 in Egypt to 1.15 per 1000 in the UAE [16]

OBJECTIVES: -

"A specific result that a person or system aims to achieve within a time frame and with available resources."[19]

"Research objective are specific accomplishment the researcher hopes to achieve by conducting the study."[20]

1. To determine pregnant women's level of knowledge about genetic diseases and congenital abnormalities
2. To assess how well pregnant women are informed about genetic diseases and congenital abnormalities through the use of information booklets.
3. To determine the relationship between the mothers' knowledge prior to the test and the socioeconomic variables that they have chosen in relation to genetic diseases and congenital abnormalities in pregnant women.

MATERIAL AND METHOD: -

RESEARCH APPROACH:-(Quantitative Evaluative Research Approach).

RESEARCH DESIGN: (Quasi Experimental One group Pre- test Post-test Research design).

STUDY SETTING:- (Kalyanpur, Kanpur, Uttar Pradesh).

ACCESSIBLE POPULATION:- (Antenatal Mothers of kalyanpur, Kanpur, Uttar Pradesh).

SAMPLE TECHNIQUES: Non - Probability Convenience Sampling Technique.

SAMPLE AND SAMPLE SIZE:- (60) Antenatal Mother.

Sample technique: -sampling technique was Non-Probability Convenience Sampling Technique.

CRITERIA FOR SAMPLING

Criteria sampling involves-

Inclusion Criteria:

- The study includes antenatal mother.
- Antenatal mother who will available at the time of data collection.

Exclusion Criteria: Antenatal mother who were not willing to participate.

Variables under study

"Attributes or characteristics that can have more than one value, such as height or weight. In other words, variable are qualities, quantities, or characteristics of people, thing, or situation that change or vary".[39]

A. Research variables:

"These are the qualities, properties, or characteristics, which are observed or measured in a natural setting without manipulating and establishing cause-and-effect relationship".[40]

Knowledge among the antenatal mothers of regarding genetic disorder & congenital anomalies was the research variable in the study.

B. Demographic variables:

"The characteristics and attributes of the study subjects are considered demographic variables."[41]

The demographic variables are age, education, no of pregnancy, monthly income, religion, dietary pattern, type of family, economic class, occupation, source of information.

DESCRIPTION OF THE TOOLS USED FOR THE STUDY; -

On the basis of development frame work to achieve the objective of the, the research was developed. Tool was developed by the investigator after an extensive review of primary and secondary source of literature and consultation with the help of the expert in the field nursing research. The tool was prepared in English language.

For conducting study, two tools were used.

TOOL 1 –Socio Demographic Variables

TOOL 2-Self Structured Knowledge Questionnaire

Questionnaire can be defined as "printed self -report form designed to elicit information that can be obtained through written or verbal response of the subject.

Tool: - 1 Socio Demographic Variables

It is mainly consisting of Socio Demographic information like age, education, no of pregnancy, monthly income, religion, dietary pattern, type of family, economic class, occupation, source of information.

Tool: -2 Self Structured Knowledge Questionnaire

This section includes 30 multiple choice questions regarding congenital anomalies & genetic disorder.

Structure of the Questionnaire.

Dimentions	Number of the Questions
Socio Demographic Variables	10
Self-Structured Knowledge Questionnaire	30

SCORING INTERPRETATION

Each question of the questionnaire will be scored one (1) for a correct response. There will be no negative marking.

$$\text{Formula used for scoring} = \frac{Obtained\ score}{Total\ Score} x\ 100$$

Table No. 3. Scoring according to their level of knowledge

S. No.	Level of Knowledge	Score	Range
1.	Inadequate Knowledge	0-10	0-30%
2.	Moderate Knowledge	11-20	30-70%
3.	Adequate Knowledge	21-30	70-100%

LIMITATIONS:-

- Knowledge regarding genetic disorder & congenital anomalies is measurable.

The investigator needed much cooperation from the antenatal mothers

DATA ANALYSIS & INTERPRITATION & DUSCISSION :-

This chapter deals with the statistical analysis and interpretation of the data collected from the samples while conducting the study.

Data refers to the knowledge collected during the course of the study. Analysis is the process of organizing and synthesizing the data so as to answer research questions and test hypotheses. Analysis of quantitative data with information collected during research study, which can be quantified, and statistical calculation can be computed. The purpose of analyzing the data is to describe the data in meaningful terms as the data collected from sample does not answer the research questions or test research hypothesis directly. The data used is to be systemically analyzed.

The data was collected from 60 Antenatal Mothers "To assess the effectiveness of Information Booklet on knowledge regarding genetic diseases and congenital anomalies among Antenatal Mothers."

THE COLLECTED DATA WAS ORGANIZED AND PRESENTED UNDER THE FOLLOWING HEADINGS: -

SECTION A: Frequency and Percentage wise description of socio Demographic Variables of antenatal mothers.

SECTION B: Frequency and Percentage wise distribution of antenatal mothers according to their pre-test and post-test level of knowledge regarding genetic diseases and congenital anomalies

SECTION C: Comparison of pre-test and post-test level of knowledge regarding genetic diseases and congenital anomalies among antenatal mothers.

SECTION D: Assess the effectiveness of Informational Booklet on knowledge regarding genetic diseases and congenital anomalies and it's benefits among antenatal mothers.

SECTION E: Association between the level of pre-test knowledge score with their selected Socio demographic variables of antenatal mothers.

CONCLUSION

From the present study it can be concluded that:-

1. **The percentage wise distribution of the patients according to their demographic variables.**

According to their age group it shows that highest percentage (40%) of mothers between 23-27 year of age groups. According to their educational status it shows that the highest percentage (33.3%) in higher secondary. According to their no of pregnancy it shows that the highest percentage(63.4%) 0f mother having one pregnancy. According to their monthly income of the family it shows that the highest (46.6%) percentage of family's monthly income is <5000/-. According to their religion it shows that 96.7% mothers are Hindu According to their diet pattern it shows that, 96.7% mothers are vegetarian. According to their family type it shows that 60% are living in joint family According to their economic status shows that, 70% in middle class According to their occupation shows that 83% are home maker According to their source of information it shows that 26.6% are getting knowledge from ASHA worker.

2. **Findings related to the effectiveness of an information booklet on knowledge regarding Genetic Disorder & Congenital Anomalies among the Antenatal Mothers**

The result shows comparison between pre-test and post-test level of knowledge. The pre-test is 12.5 and post-test is 16.5, mean percentage of pre-test is 44% and post-test is 56%; standard deviation of pre-test is 5.5 and post-test is 4.7; mean difference is 4. Thus it is showing that post-test mean score is higher than pre-test mean score.

3. **Findings related to Association between the level of pre-test knowledge score of Antenatal Mothers with their selected Socio demographic variables**

The study findings revealed that there was no significant association between pre test knowledge score and demographic -variables like monthly in-come, economic class of the family, occupation type and source of information at the level of 0.05. Except age, educational status, number of pregnancy, religion, diet pattern, type of family.

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