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# "A PRE- EXPERIMENTAL STUDY TO ASSESS THE EFFECTIVENESS OF INFORMATION BOOKLET REGARDING SELECTED GENETIC DISORDERS AMONG PEOPLE IN SELECTED AREAS OF PUNE CITY"

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## ABSTRACT

**Introduction**: A genetic disorder is a health problem caused by one or more abnormalities in the genome. It can be caused by a mutation in a single gene (monogenic) or multiple genes (polygenic) or by a chromosome abnormality. The mutation responsible can occur spontaneously before embryonic development, or it can be inherited from two parents who are carriers of a faulty gene (autosomal recessive inheritance) or from a parent with the disorder (autosomal dominant inheritance).

**Aims of the Study**: The aim of the study was to evaluate the effectiveness of an information booklet in improving knowledge about selected genetic disorders among people in selected areas of Pune city. It sought to compare pre- and post-intervention knowledge levels. The study also aimed to assess associations between knowledge and selected demographic variables.

**Methodology**: In this study, we used a quantitative approach method; where a pre-experimental one-group pretest-posttest design was used. The study consisted of 60 participants, who were selected using a non-probability purposive sampling technique. The reliability of the tool was tested on 6 participants and it was found to be reliable.

**Results**: The majority of participants (38.33%) were aged 26–30 years, with 65% males and 35% females. Most had secondary education (28.33%), and 40% were employed in the private sector. Around 21.67% had an annual income between ₹1,06,850 and ₹2,13,813. In the pre-test, 71.67% had poor knowledge of genetic disorders, while post-test results showed 81.67% had excellent knowledge. The information booklet significantly improved knowledge ($p = 0.00001$), and no significant association was found between knowledge and demographic variables.

**Conclusion**: Though most participants had poor initial knowledge of genetic disorders, post-intervention results showed significant improvement. This highlights the effectiveness of educational tools like information booklets in increasing awareness and the need for continued health education efforts.

**Keywords**: Genetic disorders, Information booklet, Knowledge assessment, Pre-experimental study, Pune city.

## INTRODUCTION

A genetic disorder is a disease caused in whole or in part by a change in the DNA sequence away from the normal sequence. Genetic disorders can be caused by a mutation in one gene (monogenic disorder), by mutations in multiple genes (multifactorial inheritance disorder), by a combination of gene mutations and environmental factors, or by damage to chromosomes

(changes in the number or structure of entire chromosomes, the structures that carry genes[1]. Some diseases are caused by mutations that are inherited from the parents and are present in an individual at birth, like sickle cell disease. Other diseases are caused by acquired mutations in a gene or group of genes that occur during a person's life. Such mutations are not inherited from a parent, but occur either randomly or due to some environmental exposure (such as cigarette smoke). These include many cancers, as well as some forms of neurofibromatosis.[2] There well over 6,000 known genetic disorders, and new genetic disorders are constantly being described in medical literature. More than 600 genetic disorders are treatable. Around 1 in 50 people are affected by a known single-gene disorder, while around 1 in 263 are affected by a chromosomal disorder. Around 65% of people have some kind of health problem as a result of congenital genetic mutations. Due to the significantly large number of genetic disorders, approximately 1 in 21 people are affected by a genetic disorder classified as rare [3].

Genetic disorders are due to alterations or abnormalities in the genome of an organism. A genetic disorder may be caused by a mutation in a single gene or multiple genes. Genes are the basic unit of heredity. Such disorders are known as genetic disorders [4].

**Need of the study:**

The 2021 Global Burden of Disease Survey found that 1.31 million people worldwide have severe thalassemia while thalassemia trait occurs in 358 million people 10 It is slightly more prevalent in males than females[5]. Those who have minor degrees of thalassemia, in common with those who have sickle-cell trait, have some protection against malaria, explaining why sickle-cell trait and thalassemia are historically more common in regions of the world where the risk of malaria is higher[6].

The present study provides a retrospective overview of a cohort of patients with rare genetic diseases identified at a tertiary genetic test centre in India.

Genetic disorders are a growing public health concern due to their lifelong impact on individuals and families. Despite advances in medical science, awareness and understanding of these conditions remain low among the general population, especially in urban communities like Pune city[7] Limited knowledge often leads to delayed diagnosis, inadequate prevention, and poor management, increasing the burden on healthcare systems and affected families[8]. As educational interventions have proven effective in improving health knowledge, the researcher identified the need to evaluate whether an information booklet could effectively enhance awareness about selected genetic disorders[9]. This study aims to bridge the knowledge gap, empower people with relevant information, and promote early detection and preventive measures, thereby contributing to better health outcomes in the community[10].

**AIM OF THE STUDY**

The aim of the study was to evaluate the effectiveness of an information booklet in improving knowledge about selected genetic disorders among people in selected areas of Pune city

**MATERIALS AND METHODS**

The present study utilized a quantitative research approach with a pre-experimental research design to assess the effectiveness of an information booklet regarding selected genetic disorders among people in selected urban areas of Pune city. The objectives of the study were to assess the pre-test knowledge score regarding selected genetic disorders, evaluate the post-test knowledge score after the intervention, determine the effectiveness of the information

booklet, and find out the association between pre-test knowledge and selected demographic variables. The target population included all individuals residing in the selected areas, while the accessible population consisted of people aged 18 to 40 years who were available during data collection. A total of 60 participants were selected through a non-probability purposive sampling technique to include individuals most relevant to the study. Inclusion criteria were adults aged 18 to 40 years, willing to participate, and able to understand either English or Marathi. Exclusion criteria included healthcare workers, individuals diagnosed with selected genetic disorders, and those with serious complications or unable to participate. The reliability of the tool was established using Karl Pearson's correlation coefficient (test-retest method), with an r-value of +0.9404 indicating high reliability. A pilot study was conducted on 6 samples to assess the feasibility of the study and the appropriateness of the statistical analysis plan.

## RESULTS

### Section-I: Description of samples of demographic attributes

According to Age majority of participants are aged between 26-30 years, representing 38.33% of the sample, followed by 31-35 years at 26.67%. Younger participants (18-25 years) make up 20% of the respondents, while 15% belong to the 36-40 years age group.

According to Gender A significant majority of the respondents were male (65%), followed by females (35%). There were no respondents identifying as transgender (0%).

According to Educational status majority of respondents have completed secondary education, accounting for 28.33%. This is followed by those with no formal education (21.67%) and those with higher education (18.33%). Graduates and those with primary education represent smaller segments, at 16.67% and 15%, respective

According to Income majority of participants earn between Rs. 1,06,850 and Rs. 2,13,813, comprising 21.67% of the sample. A significant portion of the sample falls in the Rs. 10,703 - Rs. 31,977 income bracket (18.33%), while the lowest income group (below Rs. 10,702) represents 10% of respondents. Other income ranges, such as Rs. 2,13,814 and above, Rs. 80,110 - Rs. 1,06,849, and Rs. 53,361 - Rs. 80,109, also contribute to the diversity, but in smaller proportions

According to Occupation In terms of occupation, the largest group works in the private sector (40%), followed by homemakers (21.67%), self-employed individuals (20%), and government employees (18.33%).

The demographic profile of the study participants reveals a diverse group in terms of age, gender, education, income, and occupation. Most participants were between 26 to 30 years old (38.33%), followed by those aged 31 to 35 years (26.67%), 18 to 25 years (20%), and 36 to 40 years (15%). In terms of gender, a majority were male (65%), while females accounted for 35%, with no participants identifying as transgender. Regarding education, the largest group had secondary education (28.33%), followed by no formal education (21.67%), higher education (18.33%), graduation (16.67%), and primary education (15%). Income distribution showed that most participants earned between Rs. 1,06,850 and Rs. 2,13,813 (21.67%), with others spread across various income brackets, including 18.33% earning Rs. 10,703 to Rs. 31,977 and 10% earning Rs. 10,702 or less. Occupation-wise, the highest proportion were private sector employees (40%), followed by homemakers (21.67%), government employees (18.33%), and self-employed individuals (20%). This demographic distribution indicates a

fairly balanced sample from different social and economic backgrounds, which may contribute to the generalizability of the study findings

**Section II: Analysis to assess the level of Knowledge score regarding selected Genetic disorders among people.**

**Table No. 1. Describes Pre-test level of Knowledge score regarding selected Genetic disorders among people.**

**N=60**

| LEVEL OF KNOWLEDGE PRE TEST | F | % | Mean | SD |
|---|---|---|---|---|
| POOR ( 1 - 9 ) | 43 | 71.67 | 8.53 | 3.32 |
| GOOD (10-18 ) | 15 | 25.00 | | |
| EXCELLENT ( 19-30 ) | 2 | 3.33 | | |

**Table No. 1. Describes Pre-test level of Knowledge score regarding selected Genetic disorders among people.**

The level of knowledge among participants during the pre-test revealed that the majority had poor knowledge regarding the selected genetic disorders. Specifically, 71.67% (43 participants) scored within the poor knowledge range (1-9), with a mean score of 8.53 and a standard deviation of 3.32, indicating limited awareness and considerable variability in their responses. Meanwhile, 25% (15 participants) demonstrated a good level of knowledge, scoring between 10 and 18. Only a small fraction, 3.33% (2 participants), had excellent knowledge, scoring between 19 and 30. These results highlight that prior to the educational intervention, most participants had insufficient understanding of genetic disorders, underlining the need for effective informational tools to enhance awareness in the community.

**Table No. 2: Post-test level of Knowledge score regarding selected Genetic disorders among people.**

**N=60**

| LEVEL OF KNOWLEDGE POST TEST | F | % | Mean | SD |
|---|---|---|---|---|
| POOR ( 1 - 9 ) | 0 | 0.00 | 24.5 | 4.17 |
| GOOD (10-18 ) | 11 | 18.33 | | |
| EXCELLENT ( 19-30 ) | 49 | 81.67 | | |

The post-test assessment of knowledge regarding selected genetic disorders showed a significant improvement among the participants. None of the participants scored in the poor knowledge range (1-9), indicating a complete elimination of low awareness after the intervention. About 18.33% (11 participants) demonstrated a good level of knowledge, scoring between 10 and 18. The majority, 81.67% (49 participants), achieved excellent knowledge levels, scoring between 19 and 30. The mean post-test score was 24.5 with a standard deviation of 4.17, reflecting a substantial increase in knowledge and relatively consistent understanding among the participants. These results suggest that the educational intervention, through the information booklet, was highly effective in enhancing the participants' knowledge about genetic disorders.

**Section III: Effectiveness of information booklet regarding selected Genetic disorders among people in selected areas of Pune city.**

**Table No. 3: Effectiveness of information booklet regarding selected Genetic disorders among people in selected areas of Pune city**

**N=60**

| EFFECTIVENESS OF INFORMATION BOOKLET ON KNOWLEDGE REGARDING SELECTED GENETIC DISORDERS AMONG PEOPLE | Mean | SD | DF | T test calcaluated value | P value | Remark |
|---|---|---|---|---|---|---|
| Pre test | 8.53 | 3.32 | 59 | 21.84 | 0.00001 | Significant |
| Post test | 24.5 | 4.17 | 59 | | | |

The effectiveness of the information booklet was assessed using a paired t-test, showing a significant improvement in knowledge scores from pre-test to post-test. The mean pre-test score was 8.53 (SD = 3.32), indicating poor initial knowledge, which increased to a mean post-test score of 24.5 (SD = 4.17) after the intervention. The calculated t-value of 21.84 with 59 degrees of freedom and a p-value of 0.00001 confirms that this improvement is statistically significant. These results demonstrate that the information booklet effectively enhanced participants' understanding of genetic disorders, including their causes and prevention. The improvement was consistent across various demographic groups, suggesting the booklet's broad applicability. This study highlights the value of structured educational materials in raising public awareness and knowledge about genetic health risks.

**Section IV**: **Finding related to an association between pre-test knowledge with selected demographic variables.**

The Chi-square test was used to assess the association between various demographic variables and the level of knowledge regarding selected genetic disorders among people in selected areas of Pune city. The analysis showed that there was no statistically significant association between any of the demographic variables and knowledge levels, as all p-values were greater than 0.05. For age, the Chi-square value was 12.592 with 6 degrees of freedom and a p-value of 0.165, indicating no significant association. For gender, the Chi-square value was 9.488 with 4 degrees of freedom and a p-value of 0.554, again showing no significance. The education status also showed no significant association, with a Chi-square value of 15.507, 8 degrees of freedom, and a p-value of 0.427. Similarly, income had a Chi-square value of 21.026 with 12 degrees of freedom and a p-value of 0.527, indicating no significant relationship. Lastly, occupation had a Chi-square value of 12.592 with 6 degrees of freedom and a p-value of 0.885. In conclusion, the Chi-square test results revealed no statistically significant association between demographic variables such as age, gender, education, income, and occupation with the level of knowledge regarding selected genetic disorders.

**DISCUSSION**

The current study and the study by Shivam Sharma and Rajesh Kumar Rawat (2024) both used a pre-experimental one-group pre-test post-test design to assess the effectiveness of an information booklet in enhancing knowledge about genetic conditions. While the current study

targeted the general population in selected areas of Pune city and focused on selected genetic disorders, Sharma and Rawat's study was conducted among antenatal women in Kanpur and included both genetic diseases and congenital anomalies. Both studies demonstrated a significant improvement in knowledge levels post-intervention, highlighting the effectiveness of the information booklet. In the current study, the mean pre-test score was 8.53 and the post-test score increased to 24.5, with a highly significant t-value of 21.84 ($p = 0.00001$). Although Sharma and Rawat did not report exact scores, they also found a marked improvement. The key difference lies in the population and content focus, but both studies support the value of educational tools in raising awareness about genetic health.

The current study and the study by Gayathri S. (2016) both emphasized the effectiveness of structured educational interventions in enhancing knowledge about genetic disorders. Gayathri's study focused on nursing students and used a Structured Teaching Programme (STP) Both followed a pre-experimental one-group pre-test post-test design. In Gayathri's research, 90% of students had inadequate knowledge in the pre-test, which improved significantly post-intervention with 88% achieving adequate knowledge. Similarly, in the current study, the mean knowledge score increased from 8.53 to 24.5, with a t-value of 21.84 and a p-value of 0.00001, indicating a significant improvement. While the former was more classroom-oriented, the latter was suited for community-based education, proving that both structured teaching sessions and self-learning tools like booklets can be effectively tailored to different target groups and learning environments [11]

## CONCLUSION

The study reveals that participants initially had limited knowledge about selected genetic disorders, with 71.67% scoring in the poor knowledge category during the pre-test. However, the use of an information booklet as an educational intervention led to a significant improvement in knowledge levels. Post-test results showed that 81.67% of participants achieved excellent knowledge scores, indicating a substantial gain. This improvement was statistically validated by a calculated t-value of 21.84 and a p-value of 0.00001, demonstrating the booklet's effectiveness in understanding about genetic disorders, including their causes, symptoms, inheritance, and prevention. Additionally, the study found no significant association between demographic variables—such as age, gender, education, income, and occupation—and knowledge improvement. This suggests that the information booklet was effective across diverse groups, highlighting the potential of such educational tools to reach broad populations regardless of background.

The findings emphasize the importance of accessible and in increasing public awareness about genetic disorders. Improved knowledge can empower individuals to recognize risk factors, seek timely healthcare, and adopt preventive measures. Overall, the study supports the use of information booklets as a practical and impactful method for community health education aimed at reducing the burden of genetic disorders through informed decision-making and early intervention.

## DECLARATION BY AUTHORS

**Ethical Approval:** The study was approved by the institutional ethics committee of Bharati Vidyapeeth (Deemed to be University), Pune. The study participants were briefed about the

purpose and nature of the study and written informed consent was obtained before data collection.

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**Conflict of Interest:** The authors certify that they have no involvement in any organization or entity with any financial interest in the subject matter or materials discussed in this paper

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