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# To Assess Effectiveness of Informational Booklet on Knowledge Regarding Bardet- Biedl syndrome among Nursing Students

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**Study Protocol**

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## ABSTRACT

**Background:** Biedl–Bardet syndrome (BBS) is an autosomal recessive disorder. It is classified as a "rare genetic disease " affecting about 1 in 250,000 persons worldwide. BBS is a disorder that affects many parts of the body and cause learning impairments, retinal degeneration, renal failure, obesity, post-axial polydactyly and hypogonadism. It is one of the most well-studied conditions in the family of diseases caused by defective cilia which is also known as ciliopathies. In this study, researcher provide an updated knowledge on clinical features, diagnostic developments and progress in the management of Bardet–Biedl syndrome. As Advancement occur in diagnostic technology including exome and whole genome studies and sequencing it is expanding the spectrum of patients who are diagnosed with this disorder and increasing the number of cases with diagnostic uncertainty.

**Material and Method:** A one-group pre-test-post-test research design was used to evaluate the effects of an information booklet on nursing students' understanding of Bardet-Biedl syndrome. Non probability purposive sampling technique is used. In this study a total number of 300 students who fulfilled the inclusion criteria were selected the setting of the study is selected nursing college Wardha.

**Expected result:** This study is planned to evaluate the effectiveness of information booklet on knowledge regarding Bardet- Biedl syndrome among nursing students, as a result, it is expected to determine the nursing student's degree of knowledge of the Bardet-Biedl Syndrome and to improve that knowledge.

*Keywords: Rare disease; bardet biedl syndrome; genetic therapies; nursing student.*

## 1. INTRODUCTION

A genetic disorder is a genetic problem caused by one or more abnormalities in the genome, especially a condition that is present from birth (congenital). Most genetic disorders are quite rare and affect one person in every several thousands or millions [1].

Biedl–Bardet syndrome (BBS) is an autosomal recessive disorder. It is classified as a "rare

genetic disease " affecting about 1 in 250,000 persons worldwide. BBS is a disorder that affects many parts of the body and cause learning impairments, retinal degeneration, renal failure, obesity, post-axial polydactyly and hypogonadism. Many small traits that are connected with BBS might benefit in diagnosis and are essential in therapeutic care [2]. According to population trends, there are approximately 3000 persons living with BBS in the United States and Canada and less than 15

cases documented from India. BBS prevalence varies greatly between communities ranging from 1:160 000 in northern European villages 1:13 500 to 1:17 5000 in segregated communities in the Kuwait and Newfoundland, where consanguineous marriage is more widespread [3].

Nurses are expected to be knowledgeable of genetic disorders such as Bardet Biedl syndrome that can be applied in medical practice. However, researchers are discovering that, despite the fact that student nurses will soon be working in hospitals, they are not knowledgeable or comfortable with clinical applications of genetics and genomics. The goal of this study was to assess nurse undergraduate students' knowledge of the Bardet-Biedl syndrome and to learn about their attitudes toward applying that knowledge in practice. Nurses were responsible for advising patients and family members about genetic counselling and allowing them to make an informed decision [4].

## 1.1 Background

Bardet–Biedl syndrome is a rare autosomal recessive disorder which affect multiple organ and cause defects in genes encoding for proteins that localize to the primary cilium/basal body complex. 21 disease-causing genes have been discovered to date. It is one of the most well-studied conditions in the family of diseases caused by defective cilia which is also known as ciliopathies. In this study, researcher provide an updated knowledge on clinical features, diagnostic developments and progress in the management of Bardet–Biedl syndrome. As advancement occur in diagnostic technology including exome and whole genome studies and sequencing it is expanding the spectrum of patients who are diagnosed with this disorder and increasing the number of cases with diagnostic uncertainty. As per result came of diagnostic improvements, a limited percentage of patients with only 1 or 2 clinical symptoms of BBS are now being identified. New interventions are developing are explored in this study which includes genetic therapeutics such as gene therapy, exon skipping therapy and gene editing. [5]

## 1.2 Need of the Study

Bardet Biedl syndrome Knowledge is needed by student nurse for the health care of their clients and their families. The goal of this study was to

evaluate nursing students' current genetic understanding of hereditary illnesses. This research enlightens knowledge of student nurses about this disorder. Nurse scientists around the world are increasingly integrating genomics into their programmers of research. Nurses require knowledge of Bardet Biedl syndrome for identification, support and guidance of clients either at the risk of genetic disorder as Bardet Biedl syndrome or inherited a genetic mutation. Through their focus on health and it's promotion, screening, prevention ,caring of the patient and family, community ties nurses have the chance to provide a unique viewpoint to genetic and also genomic healthcare [6].

## 1.3 Objectives

- 1) To assess the knowledge regarding Bardet- Biedl syndrome among nursing students.
- 2) To assess the effectiveness of informational booklet on Bardet- Biedl syndrome among nursing students.
- 3) To associate posttest knowledge score regarding Bardet-Biedl Syndrome among nursing students with their selected demographic variables.

## 2. METHODOLOGY

The research was conducted using an evaluative research approach with a one-group perest post-test research design. A non-probability and purposive sampling technique was utilised, with 300 samples taken from selected nursing college Wardha second, third, and fourth year B.B.Sc. student nurses.

### 2.1 Creteria of Study

#### 2.2.1 Inclusion criteria

Student nurses who: -

- Want to participate.
- Present on the day of data collection

#### 2.2.2 Exclusion criteria

Student nurses who: -

- Participated in similar type of study

### 2.2 Randomization

All participants were selected by sequentially numbered list at random.

### 2.3 Intervention

Data collection will be conducted in selected nursing college Wardha among B.Sc. nursing students. The permission for data collection taken from the principal of nursing college and class coordinator. By using purposive sampling technique 300 samples will be selected. Pertest will be conducted by using structured

questionnaire made by researcher for assessing the knowledge regarding Bardet Biedl syndrome among nursing students. Then informational booklet made by researcher were given to student nurses and after that post-test will be taken by using structured questionnaire method.

### 2.4 Statistical Analysis

Statistical analysis done by descriptive and inferential statistics.

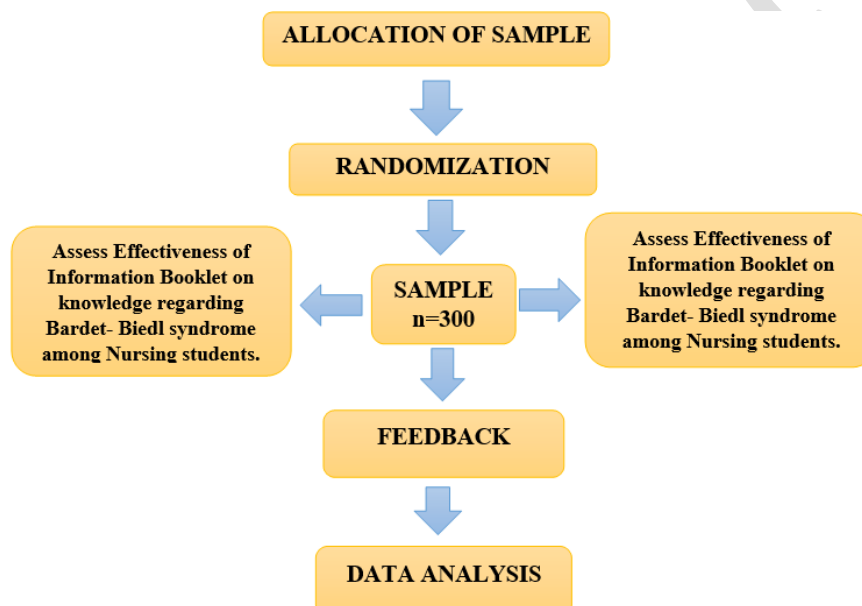


Fig. 1. Schematic diagram of study methodology

### 3. EXPECTED OUTCOME / RESULT

This study is planned to evaluate the effectiveness of information booklet on knowledge regarding Bardet- Biedl syndrome among nursing students Hence, It is expected that given informational booklet will help to improve the knowledge of the respondents.

### 4. DISCUSSION

It is clear that the nurses must expand their understanding of genetics as well as health and nursing. According to Kim's survey found a link between nurse's level of the knowledge and their interest in genetical disorders where they included by 969 nurses [7].

The study was conducted to assess the knowledge of students regarding genetics and its importance in nursing care. The results of the study researcher found that the majority of the

participant is having the average knowledge regarding the genetics and its importance in nursing care. The study also revealed that students need to emphasize on knowledge of genetics [8].

The study conducted on needs assessment of rare diseases education for nursing students in Poland from this study we found that although only approximately 1 /3rd of respondents declared having participated in university classes in rare diseases, a markedly higher number (~ 85% of nurses and ~ 75% of students) sees the need for such courses. Neither group feels well-prepared to deal with patients with rare diseases, 85% of nursing students expressing their concern in this respect. Both groups name the Internet as their major source of information [9].

The study conducted on nursing students' self-reported knowledge of genetics and genetic education we found that the majority of students

noted 'some' to 'minimal' knowledge of Mendelian inheritance and human chromosome abnormalities. In terms of awareness of genetic abnormalities and disorders, Students stated that they would like to receive more education related to genetic diseases and genetic counselling (93.9%). The study concluded that majority of students reported very little knowledge of genetic disorders. Most of students responded positively to all the education methods suggested in the questionnaire, adding that they would like more education about genetics [10].

## 5. CONCLUSION

Study finding will be drawn by using the statistical analysis.

## CONSENT

As per international standard or university standard, respondents' written consent has been collected and preserved by the author(s).

## ETHICAL APPROVAL

Datta Meghe Institute of Medical Sciences (Deemed to be University), DMIMS (DU/IEC/2021/303) and the study will be conducted in accordance with the ethical guidelines prescribed by institutional ethics committee on human research.

## COMPETING INTERESTS

Authors have declared that no competing interests exist.

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