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Interventional Programs for Genetically Disabled People Through Evidence-Based Advocacy

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Abstract

Gene alterations that are essentially present in every cell in the body cause many hereditary diseases. As a result, these ailments frequently impact many bodily systems, and the majority of them are incurable. To treat or manage some of the accompanying symptoms, there might be methods available. When monitored up to the age of 25, it is estimated that 5.3% of babies will experience a genetic disease. In order to build a modified intervention program for people with genetic disorders, this paper reviewed existing programs and interventions for people with geneticdisorders. It did this by using information about sickle cell disease. Data was gathered at the sicklecell clinic at the State Hospital, Adeoyo, Ibadan, Oyo state, where all sickle cell patients receive care. 53.3% of the survey participants were male (n=81), whereas 46.7% of the participants werefemale (n=71). In terms of age, 59.2% (n=90) of the respondents are under 20 years old, 27.6% (n=42) are between 21 and 26 years old, 9.9% (n=15) are between 26 and 30 years old, 1.3% (n=2)are between 31 and 35 years old, and 0.7% (n=1) of the respondents are each in the age brackets of 36 to 40, 41 to 45, and over 45. This demonstrates that the age range between 0 and 30 years old has the highest percentage of respondents, whereas people older than 30 are infrequently found.The respondents' marital status was also taken into account; 91.4% (n=139) of them are single, 7.2% (n=11) are married, and 1.3% (n=2) are separated.

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Introduction

According to Taylor [1], a genetic illness is a disease condition brought on by mutationsthat may be deadly or inflict varied degrees of harm. Gene mutation alters the structure of the geneand results in abrupt and spontaneous phenotypic changes [2]. It happens as a result of a modification to the DNA molecules' nucleotide sequence in a specific chromosomal location. Mutant genes are the ones that have undergone change. Bases in the gene can be lost, added, or rearranged as a result of gene mutation. According to Odunlade [3], the mutation might occur as a duplication, insertion, deletion, inversion, or substitution of nucleotides. Sickle cell disease, anemia, cystic fibrosis, phenylketonuria, and hemophilia are a few conditions that can develop as a result of substitution mutation. A common chromosomal anomaly called Down syndrome (DS) is typically caused by an extra copy of the 21st gene. According to research, Down syndrome affects 1 in 865 live births in Nigeria, with cases among young mothers being more common [5]. With an estimated 6,000 live births per year, it is the most prevalent neurogenetic

condition linked to intellectual disability [4]. According to Presson [6], 8 out of every 10,000 Americans are thought to have Down syndrome, a rise in prevalence of more than 31% since 1979 (Shin et al., 2009).

Nigeria has a high prevalence of sickle cell disease (SCD), a genetically based hemologic illness. Dr. JB Herrick discovered it for the first time in the blood of a West Indian medical student who was anemic around 1904. It is unclear precisely when, how, or where the sickle cell gene mutation took place. Nevertheless, it is hypothesized that it evolved among the Veddoids in the Arabian Peninsula's Middle East. From then, the sickle cell gene moved to Southern Europe, India, and Africa [7]. Similar to sickle cell disease (SCD), glucose-6-phosphate dehydrogenase (G6PD) deficiency is common in malaria-endemic countries including Africa (particularly sub-Saharan Africa), Asia, and the Mediterranean regions [8]. This has been related to the fact that this illness offers some protection against severe malaria, giving sufferers who live in malaria-endemic areas a survival edge. G6PD deficiency is the most prevalent enzymopathy in the world, affecting up to 400 million people (Frank, 2005). It is inherited in an X-linked recessive manner.

Gene alterations, which are essentially present in every cell in the body, are the cause of numerous genetic illnesses. These illnesses frequently impact several bodily systems as a result, and the majority of them have no known cures. To cure or control some of the accompanying signs and symptoms, however, there may be methods available. When followed up until the age of 25, 5.3% of babies are predicted to have a genetic disease. Hemoglobinopathies, glucose-6-phosphate dehydrogenase deficiency, and autosomal recessive diseases are more common in developing nations compared to western nations because of a higher proportion of consanguineous marriages and severe falciparum malaria in the distant past. Gene alterations that are seen in almost every cell in the body cause a number of hereditary diseases. As a result, these ailments frequently impact several bodily systems, and the majority of them cannot be treated. The related indications and symptoms may, however, be amenable to treatment or management. If a newborn is tracked up until the age of 25, it is predicted that 5.3% of them will have a genetic disease. Due to historically severe falciparum malaria, developing nations experience higher rates of hemoglobinopathies and glucose-6-phosphate dehydrogenase deficiency than western nations, and autosomal recessive disorders are more common due to a higher proportion of consanguineous marriages. In order to build a modified intervention program for people with genetic disorders, this paper reviewed existing programs and interventions for people with genetic disorders. It did this by using information about sickle cell disease.

Related articles

John Langdon Down, who documented many of its characteristics in "Observations on ethnic classification of idiots" in 1866, gave the condition, also known as trisomy 21, its name. However, Esquirol identified a person with Down syndrome for the first time in 1838. The relationship between the syndrome and congenital heart problems was originally noted by Garrod in 1894, and the precise association with atrioventricular septal anomalies was clarified by Abbott in 1931.

Pamela V. Daniels (2011) used a logistic regression model for binary outcome to study the timely use of prenatal care and its effect on birth outcome in black women of low economic status in the south. Binary logistic regression and ordinary least square regression were used for the Analysis. She compared the models' fit using a secondary data that examines agency factors, structural factor and risk health behavior in predicting timing of prenatal care utilization and compliance and its effect on preterm birth and low birth weight. Her main objective was to investigate why low- income black women still experience loss

of prenatal care use and compliance and worse adverse birth outcomes than other racial groups in the south.

According to the literature, descriptive statistics, such as means, percentages, and frequency counts, chi-square tests, linear regression techniques, etc. are being used more frequently to analyze student satisfaction in relation to different explanatory variables. To examine the impact of many explanatory variables on dichotomous outcomes, these study methodologies are the most suited and useful approach. Although the use of binary logistic regression in the medical profession is well known, multiple studies on socio-economic data were found that employed binary regression to illustrate the theoretical underpinnings of the mathematical model. Congenital cardiac and gastrointestinal malformations, immunological issues, oncological conditions, increased susceptibility to infections, growth disruption, and other disorders are some of the conditions associated with Down syndrome. Early intervention and educational therapy, therapeutic modalities, medications and dietary supplements, assistive technology, and other measures are used to treat Down syndrome.

Research Methodology

This part focused on the research design, research settings, target sampling approach, datacollection instrument, data collection validation method, and ethical consideration.

Research Design

To ascertain the population proportion of sickle cell disease patients in Adeoyo State Hospital, Ibadan, this study used a cross-sectional design.

Study location

The State Hospital in Adeoyo, Ibadan, Oyo state, served as the site of this study. The sickle cell clinic, where all sickle cell patients are treated, is where the data was gathered. The finalWednesday of each month is reserved for their monthly check-ups and their weekly clinic day is Friday.

Study population

According to our analysis of their record units, an average of 70 people visit the weekly check-up,and 150 people attend the monthly check-up. The survey used a sample size of 110 individuals, which was calculated using the cross-sectional study formula developed by Kish and Leslie in 1965. The sample size calculation took into account a 50% estimated proportion, a 95% level of confidence, and a 5% level of precision.

Sample size

The sample size to be used for this study is the Lesly Kish formula:

$$N = \frac{Z^2pq}{d^2} \quad (1)$$

Where:

N = Sample size

Z = the standard normal deviates corresponding to 95% C.I set at

1.96P = proportion from previous study (prevalence)

d = level of precision (5%)

Instrument for Data collection

In the Adeoyo State Hospital in Ibadan, a self-administered questionnaire was used to gather information on the numerous factors impacting sickle cell disease. The questionnaire was brokendown into four sections:

- The ramifications of social and economic variables
- Currently accessible government and multinational programs
- Potential Solutions

Traditionally, either linear discriminant function analysis or ordinary least square (OLS) regression were used to answer these research problems. Due to their strict statistical presumptions — multivariate normally with equal variance and covariances for discriminant analysis, and linearity, normality, and continuous for OLS regression—both techniques were subsequently found to be less than ideal for handling dichotomous outcomes (Cabecca, 1994; Angel Efron, 1975). In the 1960s and early 1970s, logistic regression was put up as a substitute; it first became widely used in statistical software in the early 1980s. Since then, the social sciences have increasingly used logistic regression. Logistic regression usage is still rising as a result of statistical software's accessibility. When attempting to forecast the existence or absence of traits or an outcome based on the results of a predictor variable test, logistic regression might be helpful. Despite resembling a linear regression model, it is more appropriate for models where the dependent variable is dichotomous or polytomous. It can be used in a wider variety of research situations. This study's approach was based on logistic regression. Seven independent/ predictor variables (financial constraint, shortage of health care workers, stigmatization, lack of awareness, poor utilization of HR, knowledge about SCD, and) and four dependent/outcome variables (availability of intervention program in respondent's neighborhood, accessibility of people with SCD to intervention program, benefits of people living with SCD to intervention program, and participation of people with SCD in intervention program) were used.

Discussion of Results

According to the demographic data, there were 46.7% (n=71) female survey respondents and 53.3% (n=81) male survey respondents. In terms of age, 59.2% (n=90) of the respondents are under20 years old, 27.6% (n=42) are between 21 and 26 years old, 9.9% (n=15) are between 26 and 30years old, 1.3% (n=2) are between 31 and 35 years old, and 0.7% (n=1) of the respondents are each in the age brackets of 36 to 40, 41 to 45, and over 45. This demonstrates that the age range between 0 and 30 years old has the highest percentage of respondents, whereas people older than30 are infrequently found. The respondents' marital status was also taken into account; 91.4% (n=139) of them are single, 7.2% (n=11) are married, and 1.3% (n=2) are separated. According to the researcher's analysis of the survey respondents' educational backgrounds, 8.6% (n=13) of the participants have completed primary school, 64.5% (n=98) make up the bulk of those who have completed secondary school, and 27.0% (n=41) have completed university education.

The survey's results on the participants' occupations show that 14.5% (n=22) of them are employed, 48.0% (n=73) are jobless, 30.9% (n=47) are self-employed, and 6.6% (n=10) are unskilled. The majority of participants are unemployed, as evidenced by this. To adequately account for the participants' earning potential, the researcher calculated the average participant income and divided it into four scales, as shown in Table 1. The greatest income range in the pollis less than 36,000 naira, and around 80.9% (n=128) of respondents fell into this category. About17.8% (n = 27) of the participants had incomes

between $36, 000 and $50,000, and 0.7% (n = 1) of them have incomes between $50,000 and $75,000 or more. The researcher also took into account the participants' ethnic backgrounds. The bulk of respondents—84.2% (n=128)—are Yoruba, followed by 11.2% (n=17) Igbo, 2.6% (n=4) Hausa, and 2.0% (n=2) from other ethnic groups When the researcher asked individuals about their religious affiliation, 49.3% (n=75) reported practicing Islam whereas 50.7% (n=77) reported practicing Christianity.

Table 1. Descriptive statistics of the dependent variable factors

Dependent Variable	Yes (%)	No (%)	Mean	Standard deviation
Availability of Intervention programme	46 (30.3)	106 (69.7)	0.3026	0.4609
Access to Intervention programme	69 (45.4)	83 (54.6)	0.4539	0.4995
Benefit from Intervention programme	68 (44.7)	84 (55.3)	0.4473	0.4989
Engagement in Intervention programme	66 (43.3)	86 (56.6)	0.4342	0.4973

The descriptive statistics of the dependent variable parameters for the 152 respondents in the Oyostate survey on sickle cell illness are shown in Table 1 above. With a mean of 0.3026 and a standarddeviation of 0.4609, 46 respondents (30.3%) report that there are intervention programs availablein their community, while 106 respondents (69.7%) report that there are none. With a mean of 0.4539 and a standard deviation of 0.4995, the participants who have access to the intervention program are 69 (45.4%), while 83 (54.5%) do not. 84 (55.3%) of the respondents did not benefit from the SCD intervention program, while 68 (44.7%) of the respondents did. These results had amean of 0.4473 and a standard deviation of 0.4989. 86 people (or 56.6%) have participated in anyintervention program, with a mean of 0.4342 and a standard deviation of 0.4973. Of the participants, 66 (43.3%) had participated in an intervention program.

The lack of HCWs (Health Care Workers) and ignorance of the need for adequate equipment are statistically significant with beta coefficients of (0.4751, 0.5578), respectively, according to the logistic regression model on the effect of intervention program availability on poor implementationof intervention program on sickle cell disease (SCD). The model equation is given as;

$$Y_1 = 1.6780 - 0.5329X_1 + 0.4751X^2 + 0.3374X_3 + 0.5578X_4 - 0.1145X_5 + 0.4291X_6 + 0.3122X_7$$

The logistic regression model result on effect of access to intervention program on poor implementation of an intervention program on Sickle Cell Disease shows that shortage of HealthCare Worker, lack of awareness and knowledge about Sickle Cell Disease are statisticallysignificant with beta coefficient of (0.6937, -0.5904, 0.6361) respectively.

$$Y_2 = -1.3573 - 0.45154X_1 + 0.6937X_2 + 0.2121X_3 - 0.5904X_4 + 0.1361 + 0.6361X_6 + 0.1989X_7$$

On the beneficiary's effect on intervention program on poor implementation of an intervention program on Sickle Cell Disease, financial constraints, lack of awareness, and inadequate equipment are all significant with beta coefficient (0.7299, -0.6145, 0.5529) respectively.

$$Y_3 = 0.7782 + 0.7269X_1 + 0.1645X_2 - 0.2583X_3 - 0.6145X_4 - 0.1683X_5 + 0.2342X_6 + 0.5529X_7$$

On the effect of engagement to intervention program on poor implementation of an intervention program on Sickle Cell Disease, financial constraint, stigmatization, knowledge about Sickle Cell Disease, inadequate equipment are statistically significant with a beta coefficient (0.8846, -0.4184,0.7787, 0.4355) respectively.

$$Y_4 = 2.0632 + 0.8846X_1 + 0.1098X_2 - 0.4184X_3 + 0.2222X_4 + 0.2019X_5 + 0.7787X_6 + 0.4355X_7$$

Conclusion

The analysis's findings indicate that the government has little to no interaction with those who have genetic disorders. The main factors contributing to the poor implementation of an intervention program include the following: a lack of qualified healthcare professionals, a lack ofknowledge about SCD, financial constraints, stigmatization, and inadequate equipment. The statistical significance of these variables is high. Therefore, a solution and a suggestion are required. A quality program for families of a person with a genetic condition, such as Newborn screening, should be available. Other potential intervention programs include providing enough healthcare workers, enough medical equipment, and these programs.

Limitations

There were inadequate fund and resources to carry out the study in a large form (Southwest Nigeriaand the whole region)

Recommendations

Based on the findings, the followings were suggested;

- Programmes like newborn screening, free drugs and medication, creation of vocationaltraining centres for people with genetic disorders should be adopted.
- Provision of Instructional materials in a diverse form that can help genetically disorderedpersons learn for positive contributions to the economy.
- An all-inclusive awareness on several platform like; social media, radio, television, postersetc. to enlighten the society on better ways for us to coexist.
- Provision of quality and adequate equipments to enhance treatment for the geneticallydisordered persons.

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