AWARENESS OF A REPRODUCTIVE POPULATION ON GENETIC DISORDERS AND CONGENITAL BIRTH DEFECTS: BASIS FOR MATERNAL COUNSELLING ACTIVITIES

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ABSTRACT

Genetic disorders and congenital disabilities significantly affect health and living conditions. This study aimed to determine the awareness level of the reproductive population regarding genetic disorders and congenital disabilities. A descriptive, correlational quantitative research design was used to assess respondents' awareness and to identify relationships among variables. A sample of 200 respondents from the Barangays Quirino, Roxas, Aggub, Osmeña, and Quezon of the municipality of Solano, Nueva Vizcaya was conveniently selected for the study. Descriptive statistics were used to determine the levels of awareness. Pearson's correlation coefficient was used to examine the relationship between participants' awareness levels of genetic disorders and congenital disabilities. The findings indicate that the reproductive population respondents are generally slightly aware of genetic disorders and congenital disabilities, both in terms of the basic concepts and risk factors for the two themes. The participants' level of awareness of genetic disorders is associated with their awareness of the basic concepts and risk factors of congenital disabilities. Thus, a need to enhance and clarify their understanding of the concepts and risk factors on said themes. The provision of strategic activities, such as maternal counseling and other information-dissemination efforts, can increase awareness and understanding, and, eventually, foster among the reproductive population, wise, informed decision-making and encourage preventive actions.

Keywords: Genetic counseling, genetic concepts, risk factors, developmental anomalies, maternal counseling

INTRODUCTION

Genetic disorders and congenital birth defects (CBDs) are significant public health concerns that continue to impact populations globally and locally. These conditions often result in chronic disability, emotional burden, social stigma, and even mortality, especially among newborns and infants. According to the World Health Organization (WHO, 2023), approximately 240,000 newborns worldwide die within 28 days of birth every year due to congenital disorders. Additionally, 3.5% to 5.9% of live births are known to have single-gene disorders. In contrast, chromosomal disorders occur in approximately 1 in every 263 individuals. Despite this alarming prevalence, awareness and understanding of genetic and congenital conditions remain critically low, particularly among reproductive populations.

In the Philippines, congenital anomalies such as Down syndrome, cleft palate, and congenital heart disease have consistently ranked among the top causes of infant mortality over the past five decades (VYLH Philippines, 2020). Despite public health campaigns and outreach programs, cultural beliefs, misinformation, and limited access to accurate reproductive health education persist. The study by Abad et al. (2014) highlighted how cultural misconceptions in the Philippines, such as attributing congenital and genetic disorders to supernatural or moral causes, can hinder scientific understanding and early intervention. These misconceptions may lead to negative health-seeking behaviors and poor reproductive planning.The seminal works of

Hamamy and Alwan (2023) and Pokhrel and Adhikari (2019) call for integrated genetic education, especially in reproductive health services. Hamamy and Alwan focused on the burden of these disorders in industrialized settings. At the same time, Pokhrel and Adhikari highlighted the consequences in low-income regions. These studies underline the global need for awareness and preventive strategies. However, local contexts remain underrepresented in such research, underscoring the necessity for community-specific studies in regions like Nueva Vizcaya.

Several factors increase the risk of genetic disorders and CBDs. These include maternal and paternal age, exposure to teratogens (such as radiation, drugs, and infections), consanguinity, and inadequate maternal nutrition. For instance, Barresi and Gilbert (2018) and Wolpert and Tickle (2016) identified the critical window during embryogenesis when teratogenic exposure can lead to irreversible structural or functional anomalies. Similarly, Klug et al. (2019) emphasized the correlation between advanced maternal age and chromosomal abnormalities such as Down syndrome and Turner syndrome.

Despite the existing scientific consensus, studies have consistently shown that awareness levels remain low. Sidhu et al. (2017) found that only 4.5% of married women in Sarabha, India, had a good knowledge of congenital malformations. In Rwanda, Niyibizi et al. (2023) observed that most pregnant women lacked sufficient understanding of genetic conditions, impacting their attitudes towards screening and counseling. Ogamba et al. (2021) and Lawal et al. (2015) similarly reported knowledge gaps in Nigeria, particularly among nursing mothers who were unaware of the causes and prevention of common congenital disabilities.

These findings suggest a global pattern of inadequate awareness, and the Philippine context reflects this reality. The reproductive population, defined by the WHO (2011) as individuals aged 15 to 49, includes those biologically capable of reproduction. The significance of studying this population lies in their potential to pass on genetic conditions and their role in childbearing and rearing. As parental age increases due to changing socioeconomic trends, the risk of genetic and congenital conditions also increases. Advanced Parental Age (APA), defined as 40 years or older at conception, is associated with a higher risk of chromosomal anomalies (Neeser et al., 2023; American College of Genetics and Genomics, 2018).

Maternal counseling serves as a powerful intervention to bridge the awareness gap. Verhagen and van de Laar (2017) emphasized that counseling promotes autonomy and informed decision-making. Choi and Kim (2014) explored culturally sensitive approaches, while Aliouche (2022) and Anzilotti (2021) underscored the emotional and psychological dimensions of counseling. Johnson and Dickert (2019) and ACOG (2019) linked preconception counseling to improved reproductive outcomes, suggesting that maternal counseling should be holistic, informative, and adaptable to individual needs.

Given this context, this study was designed to assess the level of awareness among reproductive-aged individuals in selected barangays in Solano, Nueva Vizcaya, regarding genetic disorders and congenital birth defects. Furthermore, the study aimed to explore the correlation between awareness of these two domains and propose culturally appropriate maternal counseling interventions. Using the Health Belief Model (HBM) as a theoretical lens, this study focuses on how perceived susceptibility, severity, benefits, and barriers influence awareness and decision- making regarding reproductive health.

METHODOLOGY

This research utilized a quantitative descriptive-correlational design to assess the awareness levels of the reproductive population on genetic disorders and congenital birth defects. The study was conducted from August 2024 to March 2025 in five barangays in Solano, Nueva Vizcaya: Quirino, Roxas, Aggub, Osmeña, and Quezon. These barangays were chosen due to their diverse demographic composition and accessibility.

The main research instrument was a researcher-developed questionnaire validated by experts in nursing, genetics, and developmental biology. The questionnaire consisted of three parts: demographic profile, awareness of genetic disorders (14 items), and awareness of congenital birth defects (14 items). Items used a 4-point Likert scale ranging from "Not aware" to "Very much aware." The instrument underwent pilot testing in a different barangay, resulting in a Cronbach's alpha of 0.953, indicating excellent reliability.

Participant recruitment followed a proportional convenience sampling strategy. A total of 200 participants aged 19 to 49 were included. Mentally challenged and seriously ill individuals were excluded. Researchers collaborated with barangay officials to identify eligible participants. Informed consent was obtained before data collection. Participants completed the questionnaire under the supervision of trained researchers who ensured clarity and consistency. Data collection adhered to ethical standards. Ethical approval was obtained from the Saint Mary's University Research Ethics Board.

Data were encoded and analyzed using IBM SPSS Version 20. Descriptive statistics, including means, standard deviations, and frequency distributions, were used to describe sample characteristics and awareness levels. To determine relationships between variables, Pearson's correlation coefficient was applied. Independent sample t-tests were used to compare awareness across demographic subgroups. Data were screened for normality, and missing data were handled using pairwise deletion.

Score, range, and qualitative interpretation of scores obtained.

Response	Mean	Level of Awareness on Genetic Disorders and Congenital Birth Defects
1	1.00 – 1.49	Not aware
2	1.50 – 2.49	Slightly aware
3	2.50 – 3.49	Moderately aware
4	3.50 – 4.00	Very much aware

The respondents' profiles were grouped by age, sex, educational attainment, socioeconomic status, ethnicity, and family history.

Profile Variables		Number	%
Age	19-23	85	42.5
	24-28	45	22.5
	29-33	18	9
	34-38	17	8.5
	39-43	14	7
	44-48	14	7
	49 and above	4	2
Sex	Male	68	34
	Female	126	63
Educational Attainment	Elementary	5	2.5
	High School	46	23
	College undergraduate	60	30
	College graduate	69	34.5
	Post-Graduate	6	3
	Others	5	2.5
Monthly Income	Not sufficient	32	16
	Slightly sufficient	55	27.5
	Moderately sufficient	72	36
	Completely sufficient	14	7
Ethnicity	Ilocano	93	46.5
	Igorot	9	4.5
	Ifugao	13	6.5
	Isinay	0	0
	Gaddang	4	2
	Tagalog	30	15
	Others	4	2
	with more than 2	43	21.5
Family History	Genetic Disorders		
	with history	58	29
	no history	141	70.5
	Congenital Birth Defects		
	with history	21	10.5
	no history	178	89

RESULTS AND DISCUSSION

Section 1. The awareness level of the respondents on genetic disorders

The respondents' awareness levels regarding genetic disorders, categorized by risk factors and general concepts, are presented in Table 5.

Statement	Mean	SD	Qualitative Description
1. Pregnant mothers who are aged 35 are at risk of a genetic disorder.	2.05	.888	Slightly aware

2. Couples who are related by blood can have children with genetic disorders.	2.61	2.392	Moderately aware
3. Women with advanced age (old age) are at risk of having DNA or chromosomal defects, thus, may have children with genetic defects, disease/disorder.	2.26	.936	Slightly Aware
4. Men of advanced age (old age) are at risk of having DNA or chromosomal defects, thus, may have children with genetic disease/disorder.	2.18	.936	Slightly aware
5. Exposure of individuals to radiation and harmful chemicals and/or drugs can cause defects in the eggs and sperm of individuals and eventually result in genetic disorders or diseases.	2.43	1.022	Slightly aware
Mean for Genetic Risk Factors	2.30	.919	Slightly aware
6. Albinism, manifested by individuals as Albino/Albina, or the so- called "Änak-araw", is a genetic disorder and can be inherited.	2.27	1.010	Slightly aware
7. Hemophilia, the inability of blood to clot, is an inherited disease/disorder	2.07	.879	Slightly aware
8. Color blindness, an inherited disorder, is common among males.	2.05	.930	Slightly aware
9. A defective gene can affect the development of a baby in the womb.	2.44	1.088	Slightly aware
10. Genetic disorder(s) is/are inherited from the mother or the father.	2.54	1.056	Slightly aware
11. Down's Syndrome or the so-called "Mongoloid condition" is a genetic disorder.	2.48	.997	Slightly aware
12. Diabetes as a disease results from the inability to produce normal amounts of insulin due to a DNA defect and is said to be inherited	2.43	.947	Slightly aware
13. Genetic disorders/diseases are due to defects in the genes and chromosomes that can be inherited.	2.39	.997	Slightly aware
14. Genetic disorders can be detected through diagnostic clinical testing during pregnancy.	2.65	1.004	Moderately aware
Mean for Genetic Basic Concepts	2.36	.796	Slightly aware
OVERALL MEAN	2.33	.804	SLIGHTLY AWARE

Means: 1.0-1.49 = Not aware; 1.5-2.49 = Slightly aware; 2.5-3.49 =Moderately aware; 3.5-4.0 = Very much aware

The data reveal that respondents have a limited understanding of genetic disorders, with an overall mean awareness score of 2.33, indicating they are only *slightly aware*. Awareness was highest regarding prenatal diagnostic testing (M = 2.65), suggesting moderate familiarity due to exposure to maternal health services. In contrast, awareness was lowest concerning the risk of genetic disorders in mothers over 35 (M = 2.05), highlighting a significant knowledge gap about age-related genetic risks. This limited awareness underscores a critical need for targeted education on genetic risk factors, including parental age, inheritance patterns, and environmental influences. Cultural beliefs and low maternal education levels further contribute to poor understanding, as supported by previous studies. To address this, the study recommends implementing genetic counseling, community seminars, school-based programs, and public

health campaigns—particularly in rural areas—to enhance awareness and promote early detection and prevention of genetic disorders.

Section 2. Awareness level of the respondents on congenital birth defects

The awareness level of the respondents on congenital birth defects, sorted into risk factors and concepts, is presented in Table 6

Statement	Mean	SD	Qualitative Description
1. Congenital hearing loss or deafness results from exposure of pregnant mothers to infectious viruses or bacteria	1.94	.865	Slightly aware
2. Using un-prescribed drugs during pregnancy will increase the risk of having a child with birth defects	2.78	.983	Moderately aware
3. Synthetic food and/or ingredients can cause malformation in babies	2.45	1.004	Slightly Aware
4. Pregnant mothers exposed to radiation (i.e., X-ray) may have babies with birth defects	2.37	1.029	Slightly aware
5. Alcohol consumption and smoking during pregnancy increase the risk of having a child with birth defects.	3.06	.961	Moderately aware
6. Women who contracted measles during pregnancy are at increased risk of having a child with birth defects.	2.33	.998	Slightly Aware
7. Women who have a folic acid deficiency are at higher risk of having a child with birth defects	2.24	1.032	Slightly Aware
Mean for Congenital Birth Defects Risk Factors	2.45	.767	Slightly aware
8. Polydactyly (extra fingers) is not an inherited birth defect.	2.03	.937	Slightly aware
9. A neural tube defect is an abnormality in the development of a baby's brain or spinal cord	1.86	.927	Slightly aware
10. Parents with normal or healthy hearts can have a baby with congenital heart disease.	1.95	.919	Slightly aware
11. Congenital birth defects can be prevented.	2.62	.979	Moderately aware
12. Congenital birth defects can be medically treated or alleviated.	2.46	.931	Slightly aware
13. A cleft lip and/or cleft palate is a congenital birth defect.	2.52	1.043	Moderately aware
.14. Congenital birth defects can be detected through diagnostic clinical tests during pregnancy.	2.50	1.046	Moderately aware
Mean for Congenital Birth Defects Concepts	2.27	.744	Slightly aware
OVERALL MEAN	2.36	.71911	SLIGHTLY AWARE

Means: 1.0-1.49 = Not aware; 1.5-2.49 = Slightly aware; 2.5-3.49 =Moderately aware; 3.5-4.0 = Very much aware

The highest awareness was found in the item, "Alcohol consumption and smoking during pregnancy increases the risk of having a child with birth defects" (M = 3.06, Moderately Aware), reflecting recognition of well-publicized teratogens influenced by public health messages, consistent with ACOG (2020) guidelines on prenatal risks.

Conversely, the lowest awareness was recorded for the item, "A neural tube defect is an abnormality in the development of a baby's brain or spinal cord" (M = 1.86, Slightly Aware), revealing a significant knowledge gap about critical and preventable conditions such as spina bifida. This is concerning, given evidence from Taruscio et al. (2017) and VYLH Philippines (2020) linking such defects to folic acid deficiency and stressing the importance of supplementation.

The overall "slightly aware" rating across most items suggests a superficial understanding of congenital defects, their causes, and prevention. This limited awareness may hinder early detection and health-seeking behavior, especially in rural or underserved areas. Cultural beliefs and misinformation, as noted by Abad et al. (2014), may contribute to misconceptions about the causes of birth defects. Similarly, Lawal et al. (2015) found that a lack of maternal knowledge is associated with poor preventive practices.

Section 3. Correlation on the level of awareness of genetic disorders and congenital birth defects of the respondents?

Table 07.
Correlation of the Awareness Level on Genetic Disorders and Congenital Birth Defects

		meanGRF	meanGBC	meanG	CRFmean	CBDmean	Cmean
meanGRF	Pearson Correlation	1	.758**	.947**	.660**	.701**	.711**
	Sig. (2-tailed)		.000	.000	.000	.000	.000
	N	199	198	198	196	191	196
meanGBC	Pearson Correlation	.758**	1	.928**	.735**	.733**	.770**
	Sig. (2-tailed)	.000		.000	.000	.000	.000
	N	198	199	198	196	191	196
meanG	Pearson Correlation	.947**	.928**	1	.743**	.764**	.789**
	Sig. (2-tailed)	.000	.000		.000	.000	.000
	N	198	198	198	195	190	195
CRFmean	Pearson Correlation	.660**	.735**	.743**	1	.817**	.955**
	Sig. (2-tailed)	.000	.000	.000		.000	.000
	N	196	196	195	197	192	197
CBDmean	Pearson Correlation	.701**	.733**	.764**	.817**	1	.951**
	Sig. (2-tailed)	.000	.000	.000	.000		.000
	N	191	191	190	192	192	192
Cmean	Pearson Correlation	.711**	.770**	.789**	.955**	.951**	1
	Sig. (2-tailed)	.000	.000	.000	.000	.000	
	N	196	196	195	197	192	197

**. Correlation is significant at the 0.01 level (2-tailed).

The correlation analysis showed a strong and statistically significant positive relationship between respondents' overall awareness of genetic disorders and congenital disabilities (r = 0.789, p = 0.000). This means that low awareness in one area is closely associated with low awareness in

the other. Similar strong correlations were found between specific components, such as genetic and congenital risk factors and their basic concepts, indicating that these areas of knowledge are interrelated.

These findings reveal a shared gap in understanding, suggesting that improving awareness in one domain may enhance knowledge in the other. As such, there is a clear need for targeted educational interventions and community-based health education to address these deficiencies. Integrating these topics into maternal counseling programs is recommended to support early prevention, informed decision-making, and improved reproductive health outcomes.

Section 4. Proposed Maternal Counseling Activities on Genetic Disorders and Congenital Disabilities Based on Study Findings

Recent studies emphasize the importance of awareness and screening for genetic disorders in healthcare. In India, a cross-sectional study by Arumugam et al. (2024) showed that only 36.5% of pregnant women had adequate knowledge of prenatal genetic screening, with awareness influenced by age, education, and pregnancy trimester. There is a need to integrate genetic awareness into primary care, given limited access to medical geneticists.

In response, a maternal counseling plan was developed to raise awareness and understanding of genetic disorders and congenital disabilities among individuals of reproductive age. Nursing students will lead the program, which consists of three main activities:

1. Seminar – This aims to educate reproductive-age individuals, especially mothers, prospective parents, student nurses, and parents of SPED students. Topics include definitions, causes, risk factors, common genetic disorders (e.g., hemophilia, color blindness), congenital disabilities (e.g., neural tube defects, cleft palate), and prenatal screening. Nursing students will lead short group sessions using printed handouts and simple presentations in barangays and schools.

2. Seminar-Workshop – Focused on prevention and management strategies, it covers genetic counseling, folic acid intake, avoidance of teratogens, and early medical interventions. Lifestyle changes, family support systems, and hands-on demonstrations will be emphasized. Activities include interactive games, role-play, and visual aids, conducted in health centers or SPED schools.

3. Awareness Campaign – Led by nursing students, this includes focus group discussions, flyer and pamphlet distribution, and short talks in barangay health centers and SPED classrooms. The goal is to ensure key information reaches the community and encourages informed reproductive decisions and preventive practices.

A sample culminating activity is a seminar workshop titled "Healthy Beginnings: Understanding Genetic Disorders and Birth Defects," targeting parents, prospective parents, and community health workers. The rationale emphasizes the persistent lack of awareness and its consequences, such as delayed diagnoses and increased burdens on families. Its general objective is to educate individuals of reproductive age about genetic disorders and congenital birth defects, equipping them with the knowledge and support needed for prevention, early intervention, and

advocacy. The seminar content includes: definitions and differences between genetic disorders and congenital disabilities; risk factors and common examples; prevention strategies; and interventions and support for affected individuals. This plan aims to bridge knowledge gaps, promote preventive behaviors, and empower communities through simple, accessible, and resource-efficient educational strategies.

CONCLUSIONS AND RECOMMENDATIONS

Conclusion

This study assessed the level of awareness of respondents from selected barangays in the municipality of Solano, Nueva Vizcaya, regarding genetic disorders and congenital birth defects. A total of 200 respondents, mostly young adults and females, participated. A descriptive-correlational research method was used, with 24-item Likert-scaled researcher-made questionnaires as the research instruments, and convenience sampling was employed for data collection. Based on the significant findings, the following conclusions were drawn.

1. Respondents are slightly aware of genetic disorders, both the general concepts and risk factors; therefore, there is a need to bridge the gap in their knowledge on the given themes (general concepts and risk factors).

2. Similarly, respondents are only slightly aware of congenital defects, both in general concepts and risk factors; thus, there is a need to enhance their knowledge and understanding of the given themes on congenital disabilities.
3. Respondents' awareness levels of genetic disorders and congenital disabilities are correlated, indicating that their knowledge and understanding of the general concepts and risk factors of genetic disorders are associated with their knowledge and understanding of the general concepts and risk factors of congenital disabilities. Thus, there is a need to clarify their understanding further and enhance their knowledge of the two conditions.

Recommendations

Based on the conclusions drawn, the following recommendations are proposed to help bridge the identified knowledge gaps, strengthen awareness, and promote prevention strategies among the target population:

1. The conduct of strategic activities, including seminars or forums, on two important themes: genetic disorders and congenital disabilities, and other related topics, for the target audience, the reproductive population.

2. The development of information-education campaign (IEC) materials, such as brochures, infographics, and video clips, for use in proper venues, including community health centers and maternal clinics, as well as in the academic setting as enhancement or reference materials in appropriate nursing courses. Community health campaigns that emphasize the importance of prenatal care and nutrition, school-based modules on maternal and fetal health, and capacity building for health workers to improve counseling on congenital risk factors should also be included in educational interventions.30

3. To explore variables or factors that influence the awareness, understanding and

practices of individuals on genetic disorders and congenital birth defects.

4. To consider the need to conduct a study on the management and practices of families with a history of cases of genetic disorders and/or congenital disorders as a basis for medical and/or nursing care intervention.

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