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Perception of Genetic Knowledge and Awareness of Genetic Counseling and Related Genetic Services among the General Population in the North Al Sharqia Region: An Exploratory Study

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Abstract

Background: Genetic knowledge and access to genetic counselling services are important for prevention, diagnosis and family planning. Data from Oman are scarce regarding public knowledge and awareness of genetic services. This study assessed genetic knowledge and awareness of genetic counselling and related services in the North Al Sharqia region.

Methods: Cross-sectional questionnaire-based study conducted January-February 2020 in villages and primary health centers in North Al Sharqia. Convenience sampling of adults (≥ 18 years) produced 226 completed questionnaires. Knowledge and awareness were measured by a self-developed instrument (Knowledge Score: 0-8; Awareness Score: 0-7). Data were analyzed using SPSS v25; chi-square tests and descriptive statistics were used; significance threshold $p < 0.05$.

Results: The final sample included 226 participants (female 55.8%). Mean Knowledge Score was 4.81 (SD 2.05) and mean Awareness Score was 4.79 (SD 1.22). Internet was the most reported source of information (66.8%). High agreement (97.8%) existed regarding the importance of diagnosing genetic disease. Education level was significantly associated with higher Knowledge Score ($\chi^2=46.997$, $df=24$, $p=0.003$) and with awareness ($p=0.03$). Prior exposure to genetic counselling was also associated with greater awareness. Age was not significantly associated with knowledge ($p=0.545$).

Conclusion: Genetic knowledge and awareness show moderate levels in this convenience sample from North Al Sharqia, but gaps remain (e.g., low awareness of premarital testing availability and misconceptions about sampling). Education and prior exposure to counselling predict higher knowledge/awareness. Targeted public education and clearer communication of local services are warranted.

Categories: Genetics, Public Health

Keywords: awareness, cross-sectional, genetic counselling, genetic knowledge, Oman

Introduction

The completion of the Human Genome Project in 2003 represented a pivotal landmark, catalyzing rapid advancements in the understanding of genetic information. Contemporary knowledge of genetic diseases has expanded considerably, facilitating the development of more effective management strategies. Disseminating this knowledge to the general public is crucial for enhancing awareness and improving coping mechanisms related to genetic disorders [1].

Globally, genetic disorders affect approximately 2-5% of live births and account for a significant proportion, around 30%, of pediatric and hospital admissions. Furthermore, these conditions are the primary etiological factor in approximately half of all deaths in children and neonates. The Sultanate of Oman exhibits a notable diversity of genetic conditions, with approximately 300 distinct types identified. Epidemiological data indicate that 3.5-4.5% of newborns in Oman are affected by genetic diseases per 1,000 births. Notably, the prevalence of Down syndrome in Oman (1 in 300 births) significantly exceeds the global average (1 in 800 births) [2].

The profession of genetic counseling originated in the United States in 1969 at Sarah Lawrence College in

New York and has since experienced global expansion. By 2018, approximately 7,000 genetic counselors were practicing across 28 countries, reflecting a substantial increase in demand for these specialized healthcare professionals as the field has matured internationally [3, 4].

A notable gap exists in research specifically addressing public awareness of genetic counseling and general genetic knowledge. Genetic counseling is a critical service that empowers patients to comprehend and accept genetic information and diagnoses, as well as to navigate the personal and familial implications of these conditions [3]. Conversely, a study published in the *Journal of Genetic Counseling* in 2018 indicated that a majority of undergraduate students surveyed possessed some familiarity with the concept of genetic counseling [4].

The role of a genetic counselor encompasses the acquisition and analysis of comprehensive medical histories to assess the likelihood of disease recurrence in offspring. Furthermore, genetic counselors provide essential education to families regarding inheritance patterns, available diagnostic and screening tests, and potential preventive measures. They also serve as a vital resource by providing patients with relevant materials and informational leaflets to facilitate a deeper understanding of their specific genetic conditions [5, 6].

Research conducted in Turkey at Pamukkale University in 2006 evaluated nurses' knowledge regarding the genetic basis of diseases, modes of inheritance, genetic counseling principles, and genetic testing methodologies. Similarly, a study focusing on nursing students revealed significant knowledge deficits in the areas of genetics and genetic disorders within this cohort [7, 8, 9].

Moreover, a systematic review encompassing nine studies examined the multifaceted barriers to accessing genetic services. The findings highlighted several key impediments, including a lack of awareness among general healthcare practitioners regarding patients' genetic risk factors, limitations in their ability to obtain thorough family histories, and insufficient foundational genetic knowledge. Additionally, many physicians demonstrated a lack of awareness concerning the availability of genetic services, exhibited failures in coordinating referrals, and operated within a context of an inadequate genetic service workforce [10].

Within the general population, a lack of awareness concerning individual risk for genetic diseases frequently prevents individuals from seeking appropriate genetic testing and counseling services. Furthermore, many individuals lack knowledge of their family history pertaining to genetic conditions, compounded by limited understanding regarding the accessibility of available genetic services [11, 12].

Several barriers impede access to genetic services even after a referral has been made. These include geographical distance to appointment locations, lack of adequate insurance coverage, familial disapproval of seeking such services, and anxieties surrounding potential test results [13, 14, 15, 16].

Encouragingly, studies suggest a gradual improvement in genetic knowledge within the general population over time [11]. Increased awareness of the genetic underpinnings of cancer can contribute to the reduction of stigma and taboos associated with cancer patients [17]. However, a persistent global challenge remains in the form of insufficient knowledge of genetics and genetic tests among physicians across various medical specialties [8, 18].

Individuals frequently acquire information about genetic tests and advancements in the field of genetics through various media channels. Primary care providers often serve as the initial point of contact for individuals seeking health-related guidance. Consequently, genetic knowledge and the principles of genetic counseling are no longer exclusive domains of geneticists and genetic counselors. This underscores the critical need for comprehensive genetic education for general healthcare practitioners through modalities such as workshops and other targeted training programs, as current knowledge levels are often suboptimal [19].

A 2018 study conducted in Egypt among media students revealed that while they recognized the importance of premarital counseling, they lacked awareness regarding the locations where such services were offered and the specific tests involved [1]. Similarly, research published in the *Hormozgan Medical Journal* in 2019 indicated that socioeconomic status significantly influences attitudes toward accepting genetic counseling in the context of premarital and marital decisions [20].

Materials And Methods

Study design

Cross-sectional, exploratory-descriptive study using a self-administered questionnaire.

Setting

Data collection occurred in January-February 2020 across several villages and primary health center catchment areas in North Al Sharqia Governorate (Lizq, Almuiaaser, Samad, Al Mamora, Al Sudira), Oman.

Participants

Adults (Omani nationals) aged ≥ 18 years, fluent in Arabic and able to read Arabic, were eligible. Recruitment used convenience sampling at outpatient visits and through a trusted intermediary network. Exclusion criteria included non-Omani nationality, age < 18 years, inability to read Arabic, and cognitive/mental conditions that could impair questionnaire comprehension.

Variables

Primary outcomes: genetic knowledge (Knowledge Score, KS; range 0-8) and awareness of genetic counselling/services (Awareness Score, AS; range 0-7). Predictor variables: age group, sex, education level, reported source(s) of genetic information, and prior exposure to genetic counselling. Knowledge and awareness items were categorical (correct/incorrect/I don't know) and converted to binary scoring as described below.

Data sources/measurement

A structured self-administered questionnaire developed in Arabic (with a translated version kept for reference) assessed socio-demographics, sources of genetic knowledge, opinions on genetic diagnosis importance, and 20 items about genetics and services. Knowledge items: 8 questions aggregated into the Knowledge Score. Awareness items: 7 questions aggregated into the Awareness Score. Pilot testing (≈ 15 physicians) assessed clarity; revisions made on their feedback. Experts reviewed content for face and content validity.

Knowledge items: 8 questions aggregated into the Knowledge Score (q 6,7,11,12,13,14,15,16). Awareness items: 7 questions aggregated into the Awareness Score (8, 9, 10, 17, 18, 19, 20)

The questionnaires distributed by me by hand. I gave to the participant and also give to participant to distributed and then collected back and submitted to me

Bias

To reduce information bias, questionnaires included the "I don't know" option to limit guessing; participants were asked not to use external aids when completing the form. Selection bias is possible due to convenience sampling and outpatient recruitment; this limitation is described in the Discussion.

Study size

The sample size decided according the population in the area through the ministry of health, north alsharqia region; they calculate the sample size for me. The sample size was calculated based on the following parameters: a confidence level of 95%, a confidence interval (margin of error) of 5%, and an estimated

population size of 93,582 for the North Al Sharqiya region. Using a standard sample size calculation formula for a finite population, a minimum sample size of 383 was determined. The larger proposed sample size of 679 aimed to account for potential non-response and incomplete questionnaires.

Quantitative variables

Knowledge Score (0-8) and Awareness Score (0-7) treated as continuous for descriptive statistics; categorical groupings were used for chi-square tests against predictor variables (age groups, education levels). Specific question items remained categorical.

Statistical methods

Descriptive statistics summarized demographics and scores (mean, SD). Associations between categorical predictors and knowledge/awareness were tested with chi-square tests; linear-by-linear association reported where applicable. Statistical analysis was performed using SPSS v25. Significance threshold $p < 0.05$; results reported with exact p-values where relevant.

Descriptive Statistics

	N	Minimum	Maximum	Mean	Std. Deviation
knowledge score	226	0	8	4.81	2.046
awareness score	226	0	7	4.79	1.224
Valid N (listwise)	226				

TABLE 1: Descriptive statistics for knowledge score and awareness score

Descriptive statistics for knowledge score and awareness score among study participants recruited from public health centers in Oman (N=226). Summary of the distribution of the knowledge score (range 0–8) and the awareness score (range 0–7); presented are N, Minimum, Maximum, Mean, and Std. Deviation. Valid N (listwise) = 226.

	Value	df	Asymptotic Significance (2-sided)
Pearson Chi-Square	22.569 ^a	24	.545
Likelihood Ratio	20.414	24	.673
Linear-by-Linear Association	4.584	1	.032
N of Valid Cases	188		

TABLE 2: Association between participant age and their level of Genetic knowledge

Statistical analysis revealed no significant association between participant age and their level of Genetic knowledge ($p = 0.545$, $p > 0.05$).

	Value	df	Asymptotic Significance (2-sided)
Pearson Chi-Square	46.997 ^a	24	.003
Likelihood Ratio	47.022	24	.003
Linear-by-Linear Association	15.190	1	.000
N of Valid Cases	219		

TABLE 3: Association between participants' level of Education and their genetic knowledge

A statistically significant positive association was observed between participants' level of

Education and their genetic knowledge (p=0.003, p<0.01).

	Value	df	Asymptotic Significance (2-sided)
Pearson Chi-Square	29.140 ^a	40	.898
Likelihood Ratio	33.156	40	.770
Linear-by-Linear Association	.005	1	.942
N of Valid Cases	212		

TABLE 4: Association between participant age and their reported sources of genetic knowledge

Statistical analysis indicated no significant association between participant age and their reported sources of genetic knowledge similarly, no statistically significant associations were found between age, source of knowledge, and overall awareness of genetic services and counseling.

Results

		Total
Age	18-30 (85) 31-40(79) 41-50(22) 51-60(2)	188(missing 38)
Sex	Male (97) female (126)	223 (missing 3)
Education level	Secondary (12) tertiary (61) university(137) higher education(9)	219(missing 7)
source of genetic knowledge	TV(6) newspaper(1) friends(25) multiple sources(77) Internet (74) journals (29)	212(missing 14)
Opinion about the importance of diagnoses of genetic disease	Important (221) Not important (0)	221(missing 5)

TABLE 5: characteristics of the participants

1	Age:
2	Gender: Male ☐ Female ☐
3	Level of education: (Primary – Preparatory – Secondary – University – Postgraduate)
4	From where do you obtain your information related to genetic diseases? (Television – Radio – Newspapers – Friends – Internet – Scientific journals)
5	What is your opinion about the importance of diagnosing genetic diseases? (Important – Not important)
6	What is the cause of Down syndrome? -Chromosomal abnormality -Intellectual disability -Physical defect -I do not know
7	When is sickle cell disease inherited from parents to their children? -If the father is a carrier of the gene and the mother is healthy -If both the father and mother are carriers of the gene -If the father is healthy and the mother is a carrier of the gene -I do not know
8	Is premarital screening available in health centers in the Sultanate? -Yes -No -I do not know
9	Are there genes responsible for some types of cancer? -Yes -No -I do not know
10	Is there a method to test embryos to identify genetic diseases outside the body before implantation into the uterus? -Yes -No -I do not know
11	How many chromosomes are there in a human cell? -45 -46 -47 -I do not know
12	From which sample can DNA be extracted? -Blood -Saliva -Skin cells -All of the above -I do not know
13	The complete genetic information encoded within DNA is known as: -Chromosome -Cell -Nucleus -I do not know
14	Medical genetics is the science that studies: -Genetic diseases -Organic diseases -Psychiatric diseases -I do not know
15	What is the percentage of inheritance of a recessive trait from two carrier parents to their children? -50% -25% -75% -I do not know
16	What is the percentage of inheritance of a dominant trait from one affected parent to the children? -50% -25% -100% -I do not know
17	What tests are available for individuals planning to get married related to genetic diseases? -Sickle cell anemia only -Sickle cell anemia and thalassemia -All genetic tests -I do not know
18	If one of your relatives is a carrier of sickle cell anemia and consults you regarding marriage, what would you advise? -To test the other partner before marriage -To proceed with marriage with no need to test the other partner -I would not advise; it is their personal choice
19	Do you support the establishment of a genetic counseling clinic? -Yes -No
20	Where can you obtain genetic counseling for yourself and your family? -Internal medicine clinics -Psychiatric clinics -Genetic counseling clinics -Behavioral medicine clinics

TABLE 6: questioner table

Participants

Out of 430 distributed questionnaires, 226 were completed and met the inclusion criteria, which required at least 75% of questions answered. Questionnaires were excluded if they were incomplete or completed by individuals outside the eligible age range. Thus, the final analyzed sample comprised 226 participants.

Descriptive Data

Participant ages ranged from 18 to 60 years and were categorized into four groups: 18-30, 31-40, 41-50, and 51-60 years. Age data were missing for 38 participants (16.8%), with missingness more frequent among females (n=22) than males (n=16). The majority of respondents (87.2%) were in the younger age groups (18-40 years), which likely reflects the accessible outpatient population and literacy profile of the sampled villages.

Gender distribution included 126 females (55.8%) and 97 males (42.9%), while gender data were missing for 3 participants (1.3%). Educational attainment was relatively high: 146 participants (64%) reported higher education (university/college), 61 (27%) had tertiary schooling below a bachelor's level, and 12 (5.3%) had completed secondary school.

The internet was the most frequently reported source of genetic information (66.8%, n=151). Academic

journals (12.8%) and friends (11.1%) were less common sources, while traditional media were rarely cited (television 2.7%, newspapers 0.4%). Nearly all participants (97.8%, n=221) agreed that diagnosing genetic disease is important.

Outcome Data

The mean Knowledge Score (KS) was 4.81 (SD 2.05; range 0-8) among 226 respondents. The mean Awareness Score (AS) was 4.79 (SD 1.22; range 0-7).

Item-level responses revealed high correct response rates for several basic items. For example, 75.5% correctly identified Down syndrome as a genetic defect, 72.6% correctly recognized sickle cell inheritance, and 86.3% were aware of premarital testing availability. Conversely, low correct response rates were observed for more specific or complex items, such as premarital testing specifics (18.1% correct) and inheritance pattern questions (~33% correct). A substantial proportion of respondents selected “I don’t know” for certain items, such as embryo testing (49.1%).

Main Results (Associations):

- Age and knowledge: Chi-square testing indicated no statistically significant association between age group and Knowledge Score (Pearson $\chi^2=22.569$, $df=24$, $p=0.545$).
- Education and knowledge: A significant positive association was observed between education level and Knowledge Score (Pearson $\chi^2=46.997$, $df=24$, $p=0.003$). Participants with higher education were more likely to achieve higher scores.
- Age and sources of knowledge: No significant association was found between age and reported source of genetic knowledge (Pearson $\chi^2=29.140$, $df=40$, $p=0.898$).
- Education and awareness: Education level was positively associated with Awareness Score ($p=0.03$), with higher-educated participants demonstrating greater awareness.
- Prior exposure to genetic counselling: Prior exposure was significantly associated with greater awareness of counselling and related services.

Overall, education and prior contact with services emerged as the strongest correlates of awareness and knowledge in this sample.

Other Analyses :

Subgroup and sensitivity analyses stratified by sex and source of information did not materially alter the main associations. Education consistently remained a robust predictor. Given the relatively small final sample compared to the planned sample size and missing data on some variables, further multivariable modeling was limited. Findings should therefore be interpreted with caution.

	Question	Wrong %	Correct %	I don't know %	missing
6	What is the cause of Down syndrome?	13 5.8	171 75.5	42 18.6	0
7	When is sickle cell disease inherited from parents to their children?	27 11.9	164 72.6	35 15.5	0
8	Is premarital screening available in health centers in the Oman?	9 4	195 86.3	22 9.7	0
9	Are there genes responsible for some types of cancer?	34 15	118 52.2	74 32.7	0
10	Is there a method to test embryos to identify genetic diseases outside the body before implantation into the uterus?	19 8.4	96 42.5	111 49.1	0
11	How many chromosomes are there in a human cell?	22 9.7	140 61.9	64 28.3	0
12	From which sample can DNA be extracted?	89 39.4	105 46.5	32 14.2	0
13	The complete genetic information encoded within DNA is known as:	39 17.3	140 61.9	47 20.8	0
14	Medical genetics is the science that studies:	----- --	219 96.9	7 3.1	0
15	What is the percentage of inheritance of a recessive trait from two carrier parents to their children?	81 35.8	74 32.7	71 31.4	0
16	What is the percentage of inheritance of a dominant trait from one affected parent to the children?	80 35.4	81 35.8	64 28.3	1
17	What tests are available for individuals planning to get married related to genetic diseases?	133 58.8	41 18.1	52 23	0
18	If one of your relatives is a carrier of sickle cell anemia and consults you regarding marriage, what would you advise?	2 0.9	212 93.8%	12 5.3	0
19	Do you support the establishment of a genetic counseling clinic?	11 4.9	213 94.2	----- ---	2
20	Where can you obtain genetic counseling for yourself and your family?	11 4.9	213 94.2	----- ---	2

TABLE 7: summary scores for each item of the questionnaire

Discussion

Principal Findings

This study assessed knowledge and awareness of genetic testing among the Omani population in the North Al Sharqia region. Overall, participants demonstrated moderate levels of knowledge and awareness, with mean scores of 4.81 and 4.79, respectively. There was near-universal agreement regarding the importance of diagnosing genetic diseases, reflecting a generally positive attitude toward the role of genetics in healthcare [21].

Educational level and prior exposure to genetic counseling emerged as the strongest predictors of higher knowledge and awareness scores. In contrast, age was not significantly associated with knowledge level in

our cohort. These findings emphasize the role of education and experiential exposure in shaping genetic literacy, rather than demographic factors alone.

Comparison With International Studies

The level of awareness observed in this study was lower than that reported in South Korea, where public awareness of genetic testing reached 89% and patient awareness reached 92% [22]. This discrepancy may be partly explained by differences in national policies, public health campaigns, and integration of genetic services into routine clinical care.

It is also important to note that our study was limited to a single region in Oman, which may not be representative of the national population. Variations in education, healthcare access, and public engagement across regions may influence awareness levels. This regional variability has been demonstrated in a cross-sectional study conducted in the Visegrad countries, which showed significant differences in knowledge and attitudes toward genetic testing among Hungary, the Czech Republic, Slovakia, and Poland, even after adjusting for confounders [23]. Hungarian respondents reported the most favorable attitudes toward the personal benefits of genetic testing, highlighting how sociocultural and systemic factors shape public perceptions.

Education, Age, and Attitudes Toward Genetic Testing

In our study, higher educational attainment was significantly associated with greater awareness of genetic testing. This finding aligns with multiple international studies demonstrating a positive association between education level and genetic literacy [21,23]. However, this contrasts with findings from the Netherlands, where individuals with lower educational levels expressed greater interest in undergoing genetic testing compared with higher-educated respondents, despite limited support for universal preventive genetic testing (only 24% approval) [21].

Although previous literature suggests that age influences opinions regarding the personal benefits and utilization of genetic testing, our study did not demonstrate a statistically significant association between age and knowledge score. This may be attributed to the fact that we did not specifically assess perceptions of personal benefit or intended use of genetic testing, which may be more sensitive to age-related differences.

Awareness of Genetic Services and Counseling

Awareness of premarital genetic testing and genetic counseling services was modest in our cohort. This finding is consistent with reports indicating that service awareness varies depending on outreach strategies and local healthcare infrastructure. Studies from populations with high rates of consanguinity, such as those reported by Bener et al., emphasize the importance of culturally tailored and context-specific educational initiatives to improve utilization of genetic services [24].

Notably, our study revealed a lack of awareness regarding the genetic counseling profession itself, a finding that mirrors results from a national cross-sectional study in the United States, which demonstrated limited public understanding of the role and scope of genetic counselors [25]. This highlights a global need to strengthen public education about genetic counseling as a distinct and essential healthcare profession.

Implications

Collectively, these findings underscore the importance of targeted educational interventions, expansion of genetic counseling services, and region-specific awareness campaigns in Oman. Enhancing public understanding of genetic testing and counseling may improve informed decision-making and uptake of preventive genetic services, particularly in societies with a high prevalence of inherited disorders.

Unanswered Questions and Future Research

Future research should include larger, probability-based studies to generalize findings to the broader Omani population. Validation of the Knowledge and Awareness scores against objective measures and their use in longitudinal assessments would strengthen their utility. Qualitative studies could also help to elucidate reasons for misconceptions and barriers to service uptake.

Conclusions

This exploratory study, the first of its kind in Oman, provides empirical evidence on genetic knowledge and awareness of genetic counselling in North Al Sharqia, revealing moderate levels of understanding with notable gaps in areas such as premarital testing specifics and inheritance mechanics. Education and prior

exposure to counselling services emerged as the strongest predictors of higher knowledge and awareness, while age showed no significant association, underscoring the importance of targeted educational interventions and service outreach rather than demographic factors. By introducing a self-developed questionnaire and new composite measures (Knowledge and Awareness Scores), this study not only establishes the current status of genetic literacy and counselling awareness but also contributes methodological innovation to the field. The findings highlight the need for policymakers and health educators to prioritize accessible public education campaigns, strengthen communication about premarital testing, and integrate genetic literacy into provider training, while future research should expand to larger, probability-based samples, validate the scoring tools, and explore qualitative insights to address misconceptions and barriers to service uptake.

Additional Information

Author Contributions

All authors have reviewed the final version to be published and agreed to be accountable for all aspects of the work.

Concept and design: Taher A. Al Shabibi , Raiya AlHabsi

Acquisition, analysis, or interpretation of data: Taher A. Al Shabibi , Raiya AlHabsi

Drafting of the manuscript: Taher A. Al Shabibi , Raiya AlHabsi

Critical review of the manuscript for important intellectual content: Taher A. Al Shabibi , Raiya AlHabsi

Supervision: Raiya AlHabsi

Disclosures

Human subjects: Informed consent for treatment and open access publication was obtained or waived by all participants in this study. Ethics Research Committee of director general of health services issued approval #1/2019. The study was approved by the Ethics Research Committee of director general of health services- North Sharqia Governorate — Approval number: Approval #1/2019. Written informed consent was implied by voluntary completion and submission of the questionnaire; participants were informed that responses were anonymous and confidential. **Animal subjects:** All authors have confirmed that this study did not involve animal subjects or tissue. **Conflicts of interest:** In compliance with the ICMJE uniform disclosure form, all authors declare the following: **Payment/services info:** All authors have declared that no financial support was received from any organization for the submitted work. **Financial relationships:** All authors have declared that they have no financial relationships at present or within the previous three years with any organizations that might have an interest in the submitted work. **Other relationships:** All authors have declared that there are no other relationships or activities that could appear to have influenced the submitted work.

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