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Assessment of Physicians' Knowledge of Molecular and Genetic Testing in Sana'a City, Yemen: A Cross-Sectional Study

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ABSTRACT

Background: Genetic and molecular testing is a cornerstone of modern personalized medicine, offering significant opportunities for early diagnosis and disease prevention. In Yemen, where consanguinity rates are high, the prevalence of genetic disorders necessitates a high level of physicians' knowledge. This study aimed to evaluate the level of knowledge regarding genetic and molecular testing among physicians in Sana'a City, Yemen, and to identify the demographic factors influencing their understanding.

Methods: A cross-sectional study was conducted between January and March 2025 among 384 physicians from four public and two private hospitals in Sana'a City using a self-administered questionnaire. Data were summarized using descriptive statistics.

Results: Of the 384 physicians conveniently included in this study, the highest knowledge level was observed for routinely used tests, where ≥80% of physicians were aware of polymerase chain reaction (PCR) for hepatitis B virus (HBV) and hepatitis C virus (HCV), and ≥65% were aware of thalassemia screening. However, knowledge of advanced genomic techniques, such as whole-genome sequencing (25.8%) and whole-exome sequencing (24%), were substantially lower.

Conclusion: There is a substantial knowledge gap among physicians in Sana'a concerning modern genetic and molecular testing applications. These results underscore the urgent need for integrating medical genetics into continuing medical education and clinical training programs to improve the utilization of genomic medicine in Yemen's healthcare system.

Keywords: Knowledge ▪ Physicians ▪ Genetic testing ▪ Molecular testing ▪ Yemen

1. Introduction

Genetic testing is a rapidly emerging area of healthcare with the potential to revolutionize disease prevention, diagnosis, and individualized treatment.[1,2] In addition, molecular diagnostics, such as polymerase chain reaction (PCR)-based methods, offer sensitive and specific approaches to the diagnosis of hereditary and non-hereditary diseases.[2] The rapid development of bioinformatics has played a crucial role in several medical specialties, including pharmacogenomics, and oncology, cardiology.[1–3] Moreover, advancements in next-generation

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sequencing technologies have made these tools more accessible to many laboratories in the world.[1]

Personalized medicine, where medical care is customized to suit each person's unique genetic composition, primarily relies on the thorough integration of genetic testing into routine clinical practice.[2,3] For example, chromosomal microarray (CMA) has been promoted as a first-tier diagnostic technique for individuals with developmental disability and/or congenital anomalies, illustrating the move towards genome-wide strategies to diagnosis in the clinic.[4]

The successful implementation of genetic testing faces a number of challenges. A key component is the readiness of healthcare providers, especially physicians, who are involved in referring, interpreting and communicating the results of genetic tests to patients.[5] Numerous global studies indicate a remarkable paucity of genetic testing knowledge among physicians due to inadequacies in their formal medical education.[6,7] This lack of knowledge may preclude the optimal ordering of important tests and the appropriate interpretation and communication of their results, resulting in compromised patient care.[8]

There are also complex ethical, legal, and social implications of genetic testing that go beyond the technical knowledge. Concerns about patient privacy, the risk of genetic discrimination, and accompanying psychological impact from knowing certain genetic information have made physicians more cautious.[9] This problem has been further complicated by the introduction of guidelines, such as the American College of Medical Genetics and Genomics (ACMG) recommendations for reporting incidental findings in clinical sequencing, which require physicians to make complex decisions regarding the disclosure of unanticipated yet potentially actionable results.[10]

Overcoming these complex challenges requires a cohesive approach of specific educational programs, definitive clinical guidelines and multidisciplinary collaboration that values the important input of genetic counselors and specialists. A majority of physicians in Yemen lack adequate exposure to genetic testing, and this may lead to lack of clarity while ordering or interpreting tests. Therefore, this study aimed to assess their knowledge of molecular and genetic testing.

2. Methods

2.1. Study design

A cross-sectional study was conducted among physicians from various specialties in Sana'a city. The study included physicians working in four public hospitals—Al-Thawra Modern General Hospital, Republican Teaching Hospital, Kuwait University Hospital, and Al-Sabeen Maternity and Child Hospital—as well as two private hospitals: University of Science and Technology Hospital and Modern European Hospital.

2.2. Sample size and sampling strategy

Using the online Raosoft® sample size calculator (www.raosoft.com), the minimum required sample size was estimated at 377 physicians, assuming a 50% response distribution, a 95% confidence level, and a 5% margin of error. However, 384 physicians were included in the study. Physicians from the six hospitals were selected via convenience sampling until the target sample size was reached.

2.3. Data collection

A self-administered online questionnaire was used to collect data on the demographic and professional characteristics of physicians, as well as well as their knowledge of basic genetic concepts, molecular tests for infectious diseases, cytogenetic and chromosomal analyses, genetic tests for single-gene disorders and complex diseases, and other genomic and genetic investigations. The questionnaire was

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developed based on a review of pertinent published literature. To ensure content validity of this revised instrument, it was evaluated by an expert panel consisting of a microbiology instructor, a physician, and a statistician. The panel assessed the questionnaire's appropriateness, relevance, clarity, and its effectiveness in capturing the required information.

2.4. Data analysis

The data were imported from an Excel sheet into IBM SPSS Statistics version 25.0 for Windows® (IBM Corp., Armonk, NY, USA). Categorical variables were presented as frequencies and percentages.

3. Results

3.1. Study population characteristics

Of the 384 physicians, Table 1 shows that more than half (58.3%) were males. The mean age was 35.5±11.7 years, with 54% aged 30 years or older. Most physicians were married (71.9%) and had children (68.2%). Regarding professional rank, most physicians (45.8%) were general practitioners, followed by specialists (32.3%) and consultants (21.9%).

Table 1: Characteristics of physicians included in the study

Characteristics	*N* (%)
Gender	
Male	224 (**58.3**)
Female	160 (**41.7**)
Age (years)	
Mean ± SD: 35.5±11.7	
<30	178 (**46**)
≥30	206 (**54**)
Marital status	
Single	108 (**28.1**)
Married	276 (**71.9**)
Parental status	
Yes	262 (**68.2**)
No	122 (**31.8**)
Professional rank	
General practitioner	176 (**45.8**)
Specialist	124 (**32.3**)
Consultant	84 (**21.9**)

* The total number of participants was 384. SD, standard deviation.

3.2. Knowledge of basic genetic concepts

Table 2 shows that 74.7% of physicians identified the possibility of early detection of certain disorders using DNA testing, 73.7% correctly recognized the potential to use genetic knowledge to prevent or treat a disorder, 66.1% were aware that healthy parents can have a child with a hereditary disease, 64.3% acknowledged the importance of DNA testing for relatives, and 63.5% knew that the genotype is not susceptible to human interventions. However, only 10.2% recognized a gene as a molecule that controls hereditary characteristics.

Table 2: Knowledge of basic genetic concepts among physicians in Sana'a City, Yemen (2025)*

Basic genetic concept	Knowledge *n* (%)
A gene is a molecule that controls hereditary characteristics.	39 (**10.2**)
The genotype is not susceptible to human interventions.	244 (**63.5**)
Healthy parents can have a child with a hereditary disease.	254 (**66.1**)
It is possible to early detect certain disorders using DNA testing.	287 (**74.7**)
DNA testing is important for relatives.	247 (**64.3**)
It is possible to use genetic knowledge to prevent or treat a disorder.	283 (**73.7**)

* The total number of participants was 384. DNA, deoxyribonucleic acid.

3.3. Knowledge of infectious disease molecular testing

Table 3 shows that the highest proportion of physicians were aware of real-time PCR for HBV (88%), HCV (83.1%) and HIV (82.3%). Fewer physicians were knowledgeable about real-time PCR for TB (72.9%) and CMV (70.1%), and *Toxoplasma gondii* PCR (68.8%). Approximately two-thirds (65.4%) recognized HPV real-time PCR and 60.9% were aware of EBV PCR, while just over half of the physicians (56.5%) were aware of HCV genotyping. However, less than half recognized H1N1 real-time PCR and viral genetic testing in general (31%).

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Print ISSN: 2959-4138

Table 3: Knowledge of infectious disease genetic testing among physicians in Sana'a City, Yemen (2025)*

Infectious disease genetic testing	Knowledge *n* (%)
HBV quantitative real-time PCR	338 (**88.0**)
HCV quantitative real-time PCR	319 (**83.1**)
HIV quantitative real-time PCR	316 (**82.3**)
TB quantitative real-time PCR	280 (**72.9**)
CMV quantitative real-time PCR	269 (**70.1**)
Toxoplasma gondii PCR	264 (**68.8**)
HPV quantitative real-time PCR	251 (**65.4**)
EBV PCR	234 (**60.9**)
HCV genotyping	217 (**56.5**)
H1N1 quantitative real-time PCR	186 (**48.4**)
Viral genetic testing	119 (**31.0**)

* The total number of participants was 384. PCR, polymerase chain reaction; HBV, hepatitis B virus; HCV, hepatitis C virus; TB, tuberculosis; HIV, human immunodeficiency virus; CMV, cytomegalovirus; HPV, human papillomavirus; EBV, Epstein–Barr virus; H1N1, influenza A virus subtype H1N1.

3.4. Knowledge of cytogenetic and chromosomal testing

Table 4 shows that 66.1% correctly identified Down syndrome testing. However, just over half had knowledge regarding Edwards syndrome testing (52.3%), X-chromosome analysis (55.5%), and Y-chromosome analysis (50.3%). Less than half were aware of karyotype analysis (47.9%), Patau syndrome testing (42.4%), and prenatal genetic testing (40.6%). The lowest proportion (24.7%) were aware of Y-chromosome azoospermia factor region analysis.

Table 4: Knowledge of cytogenetic and chromosomal testing among physicians in Sana'a City, Yemen (2025)*

Cytogenetic and chromosomal testing	Knowledge *n* (%)
Down syndrome testing	254 (**66.1**)
X-chromosome analysis	213 (**55.5**)
Edwards syndrome testing	201 (**52.3**)
Y-chromosome analysis	193 (**50.3**)
Karyotype analysis	184 (**47.9**)
Patau syndrome testing	163 (**42.4**)
Prenatal genetic testing	156 (**40.6**)
Y-chromosome AZF region analysis	95 (**24.7**)

* The total number of participants was 384. AZF, azoospermia factor.

3.5. Knowledge of crossmatch and transplantation testing

Table 5 shows that most physicians recognized human leukocyte antigen (HLA) typing for the transplantation of kidneys (72.4%), liver (65.6%) and bone marrow (57.6%). In contrast, less than half (44.8%) were aware of crossmatch procedures for kidney transplantation. Overall, more than half of physicians (56%) were aware of the HLA typing test in general.

Table 5: Knowledge of crossmatch for transplantation testing among physicians in Sana'a City, Yemen (2025)*

Crossmatch for transplantation testing	Knowledge *n* (%)
HLA typing for kidney transplantation	278 (**72.4**)
HLA typing for liver transplantation	252 (**65.6**)
HLA typing for BM transplantation	221 (**57.6**)
HLA typing test	215 (**56.0**)
Crossmatch for kidney transplantation	172 (**44.8**)

* The total number of participants was 384. HLA, human leukocyte antigen; BM, bone marrow.

3.6. Knowledge of genetic testing for single-gene disorders

Table 6 shows that most physicians were aware of the genetic testing of β- (70.3%) and α-thalassemia (65.4%). More than half recognized DNA fragmentation test (54.7%) and recurrent abortion genetic testing (47.9%). However, less than half recognized genetic testing for cystic fibrosis (48.7%), HLA-B27 (41.9%), familial Mediterranean fever and inherited metabolic disorders (40.1% each), fertility-related disorders (40.6%), Gaucher disease (39.3%), arthritis and asthma (38.8% each), epilepsy (37.8%), and prothrombin G20210A mutation (37%). Knowledge of other genetic testing ranged from 18.8% for promyelocytic leukemia-retinoic acid receptor alpha fusion gene to 33.6% for inherited neurological disorders.

3.7. Knowledge of genetic testing for complex diseases

Regarding genetic testing for complex diseases, Table 7 shows that the highest proportion of physicians were aware of DNA testing (64.6%), followed by the genetic testing for diabetes (49.7%) and food allergy (49.2%), and both allergy and hereditary genetic testing (46.1% each). However, less than 45% of physicians were aware of genetic testing for other diseases, ranging from 25% for rare disease genetic screening to 44.8% for general genetic mutation analysis.

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Table 6: Knowledge of genetic testing for single-gene disorders among physicians in Sana'a City, Yemen (2025)*

Molecular genetic test/disorder	Knowledge n (%)
β-Thalassemia	270 (**70.3**)
α-Thalassemia	251 (**65.4**)
DNA fragmentation test	210 (**54.7**)
Cystic fibrosis	187 (**48.7**)
Recurrent abortion genetic profile	184 (**47.9**)
Hemochromatosis	179 (**46.6**)
HLA-B27	161 (**41.9**)
Fertility genetic testing	156 (**40.6**)
FMF	154 (**40.1**)
Inherited metabolic disorders	154 (**40.1**)
Gaucher disease	151 (**39.3**)
Arthritis genetic testing	149 (**38.8**)
Asthma genetic testing	149 (**38.8**)
Epilepsy genetic testing	145 (**37.8**)
Prothrombin G20210A mutation	142 (**37.0**)
Inherited neurological disorders	129 (**33.6**)
JAK2 V617F mutation testing	125 (**32.6**)
Huntington's disease testing	117 (**30.5**)
Genes associated with sleep disorders	109 (**28.4**)
Factor V Leiden (G1691A) mutation	107 (**27.9**)
Rare disease genetic testing	104 (**27.1**)
MTHFR gene mutation	88 (**22.9**)
CALR gene mutation testing	80 (**20.8**)
KRAS mutation testing	78 (**20.3**)
BRAF mutation testing	76 (**19.8**)
PML-RARA fusion gene	72 (**18.8**)

*The total number of participants was 384. FMF, familial Mediterranean fever;. HLA, human leukocyte antigen; KRAS, Kirsten rat sarcoma viral oncogene homolog; BRAF, B-Raf proto-oncogene; JAK2, Janus kinase 2; MTHFR, methylenetetrahydrofolate reductase; PML-RARA, promyelocytic leukemia-retinoic acid receptor alpha; CALR, calreticulin; DNA, deoxyribonucleic acid; HLA-B27, human leukocyte antigen-B27 allele.

Table 7: Knowledge of genetic testing for complex diseases among physicians in Sana'a City, Yemen (2025)*

Molecular genetic test/condition	Knowledge n (%)
DNA testing	248 (**64.6**)
Diabetes genetic testing	191 (**49.7**)
Food allergy genetic testing	189 (**49.2**)
Hereditary genetic testing	177 (**46.1**)
Allergy genetic testing	177 (**46.1**)
General genetic mutation analysis	172 (**44.8**)
Marfan syndrome gene screening	167 (**43.5**)
Wilson disease gene screening	162 (**42.2**)
Hereditary disease gene mutation screening	156 (**40.6**)
Hereditary cancer gene screening	153 (**39.8**)
Cardiovascular genetic testing	146 (**38.0**)
Obesity genetic testing	141 (**36.7**)
BRCA1/BRCA2 mutation testing	138 (**35.9**)
Noonan syndrome gene screening	136 (**35.4**)
Recurrent pregnancy loss screening (**28** genes)	134 (**34.9**)
BCR-ABL fusion gene testing	122 (**31.8**)
Neurological disorder gene mutation screening	119 (**31.0**)
Targeted genetic testing	113 (**29.4**)
BCR-ABL T315I mutation analysis	104 (**27.1**)
Rare disease genetic screening	96 (**25.0**)

*The total number of participants was 384. DNA, deoxyribonucleic acid; BRCA, breast cancer susceptibility gene; BCR-ABL, breakpoint cluster region-Abelson; PCR, polymerase chain reaction.

3.8. Knowledge of miscellaneous genomic and genetic testing

Table 8 shows that 66.9% of physicians were aware of genetic testing for celiac disease, and 56% were aware of metabolic genetic testing. However, low knowledge levels were observed for genetic lineage examination (27.9%) and screening for genetic changes associated with aging (27.6%). Likewise, lower proportions of physicians were aware of whole genome sequencing (WGS) and whole exome sequencing (WES), being identified by 25.8% and 24% of physicians, respectively. The lowest proportion of physicians had knowledge of microbiome genetic testing (23.4%).

Table 8: Knowledge of miscellaneous genomic and genetic testing among physicians in Sana'a City, Yemen (2025)*

Genomic/genetic testing	Knowledge N (%)
Celiac disease genetic testing	257 (**66.9**)
Metabolic genetic screening	215 (**56.0**)
Paternity testing	107 (**27.9**)
Screening for genetic aging-associated changes	106 (**27.6**)
WGS	99 (**25.8**)
Screening for genes related to general health	97 (**25.3**)
WES	92 (**24.0**)
Microbiome genetic testing	90 (**23.4**)

* The total number of participants was 384. WGS, whole genome sequencing; WES, whole exome sequencing.

4. Discussion

To the best of our knowledge, this is the first study to assess the knowledge of physicians in Yemen regarding molecular and genetic testing. The study revealed substantial gaps in basic genetic knowledge, with only 10.2% of participants recognizing a gene as a molecule controlling hereditary characteristics, whereas 74.7% recognized the potential of genetic testing for early detection of disorders. This indicates that physicians were more familiar with the practical applications of genetic testing than with basic genetic concepts. These findings align with a finding from Lebanon, where only 6% of primary care physicians scored highly on actual genetic knowledge assessments, despite 38% perceiving their knowledge

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as adequate.[11] In the United States, less than half of primary care physicians had formal training in genetics, although they understood its clinical applications.[12]

As regards cytogenetic and chromosomal tests, approximately two-thirds of the physicians reported knowledge of Down syndrome. However, lower proportions demonstrated knowledge of testing for Edwards syndrome (52.3%), Patau syndrome (42.4%), standard karyotype analysis (47.9%), and Y-chromosome azoospermia factor (AZF) region analysis (24.7%). Similar patterns have been documented in studies from Saudi Arabia and the United States, where clinicians tend to show greater knowledge of common chromosomal abnormalities than of rarer syndromes. [13,14]

Knowledge of single-gene disorder testing also showed variability. Nearly two-thirds of physicians reported knowledge of genetic tests for β-thalassemia and α-thalassemia, whereas familiarity with assays such as BRAF mutation testing, PML-RARA fusion gene analysis, KRAS mutation testing, as well as tests for inherited neurological disorders, sleep-related genetic conditions, and rare diseases was markedly lower. Comparable patterns have been reported in other settings,[15,16] underscoring the influence of disease prevalence and the content of medical training on physicians' familiarity with specific genetic tests.

Understanding of genetic testing for complex diseases was generally moderate. Physicians were most familiar with DNA testing (64.6%), diabetes genetic testing (49.7%), and food allergy genetic testing (49.2%). In contrast, more specialized applications—such as targeted genetic testing (29.4%), BCR-ABL T315I mutation analysis (27.1%), and rare disease genetic screening (25.0%)—were much less recognized. Similar patterns have been reported in Brazil and the United States, indicating that more advanced genetic testing modalities remain underutilized or insufficiently appreciated in routine clinical practice.[11,17]

Knowledge of advanced genomic technologies was notably limited, with approximately one quarter of physicians being familiar with WGS and WES, while roughly 23–28% recognized tests such as screening for aging-related genetic changes, genetic health profiling, and microbiome genetic testing. By contrast, substantially higher proportions were aware of celiac disease genetic testing (66.9%) and metabolic genetic screening (56.0%). Similar patterns have been described in studies from the United Kingdom and Canada,[18,19] highlighting widespread international gaps in clinicians' understanding of emerging genomic technologies. Improving early diagnosis, prevention, and patient management of hereditary and complicated diseases depends on increasing physicians' fundamental knowledge as well as hands-on instruction in genetic testing.

This study unveils the gaps in Yemeni physicians' knowledge of molecular and genetic testing. Nevertheless, some limitations should be considered. Data collection was restricted to physicians practicing in Sana'a City using convenience sampling, limiting generalizability to other regions of Yemen. Reliance on self-reported data may have introduced social desirability bias. Additionally, although the questionnaire was adapted from validated tools, formal pilot testing or calculation of validity indices was not conducted, although prior usage supports its validity.

5. Conclusion

Physicians in Sana'a generally lack sufficient knowledge of molecular and genetic testing, particularly in basic concepts and advanced genomic tools. Improving their understanding is critical to support accurate diagnosis, prevention, and effective patient care. Targeted training programs should address essential topics such as common genetic disorders, modern testing methods, and practical case ap-

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plications, with emphasis on integrating genetics into medical education and continuous professional development.

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Ethical approval

Ethical approval for this study was obtained from the Research Ethics Committee of the Faculty of Medicine and Health Sciences at the University of Science and Technology, Sana'a, Yemen (Ethical Approval No.: 1447/0065/UREC/UST). In addition, informed consent was obtained electronically from all participating physicians through an online form, following a clear explanation of the study's objectives. All participant information was treated with strict confidentiality and was securely managed by the research team.

Conflict of Interest

The authors declare no conflict of interest associated with this article.

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