



Case-based genomics education and training of neurologists: an Egyptian initiative

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Abstract

Many physicians lack confidence in providing genetic services to patients due to a lack of genetic/genomic knowledge. The study aimed to develop and implement a genomic education and training program for neurologists, a real initiative activity. The program consisted of three steps: (1) conducting an exploratory survey to identify knowledge gaps, attitudes, and concerns related to medical genetics/genomics in practical settings. (2) Designing the framework and implementing the national initiative program. (3) Preliminary evaluation of the program outcomes. The program was conducted as part of postgraduate education at a university hospital. Survey responses (42.5%) indicated that many neurologists expressed the importance of genomic medicine but lacked confidence in applying genomic tests in practice and addressing patient questions about genetic diseases. They expressed a preference for face-to-face learning, including case discussions and interpretation of genetic test results. The initiative comprised seven courses conducted over 24 months, with a total of 42 regular meetings. It involved three academic consultant neurologists as expert educators and 45 junior neurologists as trainees. Case discussions and interpretation of authentic genomic results were conducted for 46 patients. Evaluation of the initiative by trainees was promising. Neurologists reported increased genomic knowledge and felt more comfortable referring patients for genetic testing after receiving guidance from expert peers. Findings indicate that neurologists seek scalable and ongoing genomic education and training tailored to their field. Face-to-face, case-based learning led by expert educators in genomics, focusing on neurology, appears to be the most effective approach to address gaps in genomic education and training.

Introduction

Over the past two decades, significant progress has been made in genomics, with increased availability of genomic tests, advancements in genomic technologies, and the identification of genetic causes for many diseases. This has led to the integration of genetic knowledge into medicine, expanding the role of clinical genetics beyond diagnosis and counseling to include personalized treatment options based on individual genetic information. As technology becomes more affordable, genetic testing is expected to become more widespread globally, with genomic data from diverse

populations aiding in the understanding and treatment of genetic and rare diseases. Despite these advancements, studies from various regions of the world have shown that many practicing physicians have been slow to adopt genetic services and incorporate genomic data into clinical practice. In most countries, practicing clinicians are the primary healthcare providers responsible for ordering genetic testing and providing counseling to patients (Houwink et al. 2012; Salm et al. 2014; Dougherty et al. 2016; Chow-White et al. 2017; Jaitovich Groisman et al. 2017; Prokop et al. 2018; Kathrens-Gallardo et al. 2021; Mladenić et al. 2024). The shortage of genetic professionals, such as medical geneticists and genetic counselors, has necessitated offering genetics/genomics education and training to practicing physicians to ensure they have the necessary genomic skills (Maiese et al. 2019). Genetics/genomics illiteracy is repeatedly cited as a significant obstacle to the effective application of medical genetics. Genetic literacy involves having adequate knowledge in genetics and genomics, as well as the ability to understand and utilize genetic

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information for medical decisions related to genetic disorders and testing (Najafzadeh et al. 2013; Mikat-Stevens et al. 2015; Chou et al. 2021). Research indicates that clinicians worldwide, regardless of their country's development status, have received inadequate genetic education during medical school and throughout their medical careers. The genetic curriculum is typically covered in the preclinical years, comprising only a small percentage of the curriculum and lacking practical clinical relevance. Even in the clinical years, genetic education remains limited to descriptions of well-known genetic diseases (Hyland et al. 2019). In US and Canadian medical schools only 26% of schools reported formal genetics teaching in years 3 and 4 with the core curriculum generally aligns with the Association of Professors of Human and Medical Genetics guidelines. However, new topics like personalized medicine (21%) and direct-to-consumer testing (18%) have been incorporated, while eugenics (17%), linkage analysis (16%), and evolutionary genetics (15%) have been removed (Plunkett-Rondeau et al. 2015). Additionally, after completing medical school, the majority of M.D. graduates pursue specialties other than medical genetics (Maiese et al. 2019) and there are no strict requirements for ongoing genomic education in various medical specialties.

Studies indicated that professionals face barriers such as limited genetic knowledge, lack of risk assessment tools, high test costs, and time constraints (Mikat-Stevens et al. 2015; East et al. 2022). Concerns about insurance discrimination and skepticism towards genetic testing companies also exist (Hauser et al. 2018). Moreover, in many regions, genomics is not a priority in medical services for patients due to limited funding, resource constraints, healthcare infrastructure limitations, and resistance to adopting new genetic services and testing. Consequently, physicians may lack knowledge of genomics advancements and new genetic testing options (Korf et al. 2014; Mikat-Stevens et al. 2015; Chow-White et al. 2017; Talwar et al. 2017; Hauser et al. 2018; Carroll et al. 2019). Despite these challenges, there is a global push for enhanced genomics education particularly primary care physicians and specialists. There are international initiatives aiming to promote personalized medicine through genomic education (Paneque et al. 2016; Crellin et al. 2019; McClaren et al. 2020). Societies of human genetics in various countries (as Europe, the United States, Australia, and Canada, countries with abundant genetic resources) support genomic education initiatives and E-learning resources (Skirton et al. 2010; Korf et al. 2014; Talwar et al. 2017; Carroll et al. 2019; McClaren et al. 2020). It has also been indicated that collaboration among professionals from different countries has led to adaptable frameworks for genetics education. In this context, the European Society of Human Genetics has established competencies in genetics

for healthcare practitioners, providing a framework for genetics education tailored to national contexts and healthcare settings (Skirton et al. 2010).

In Egypt, there is no formal career path for medical students to specialize in medical genetics, similar to many other countries. The country lacks certification for clinical genetics specialties through its higher education institutions and does not prioritize the training of genetic specialists in counseling and laboratory work. Consequently, there is a shortage of medical geneticists, counselors, and other genetic specialists in Egypt, as well as a deficiency in postgraduate medical training in genetics and genomics. This situation places the responsibility of providing genetic services on clinicians in Egypt within clinical settings. Moreover, patients with hereditary diseases in Egypt face inadequate access to genetic services due to limited attention from stakeholders, high costs, limited funding, and a small genetic pool in the region. Healthcare services in Egypt, including genetic services, are predominantly paid for out-of-pocket by patients, as government insurance coverage for genetic testing is scarce and relies on sporadic international research funding (Hamed 2023).

The study aimed to develop a specialized genomics education and training program for Egyptian neurologists to enhance their ability to address genetic disorders in their patients effectively. The process involved three main stages: 1. Pre-implementation stage: This involved surveying neurologists to identify knowledge gaps, attitudes, and concerns in medical genetics/genomics. 2. Implementation stage: This included developing a course outline and structured program framework. 3. Feedback evaluation from trainees as an initial assessment of the program's impact.

To the best of our knowledge, this is the first completed genomic education and training program initiative in Africa for non-genetic health professionals specializing in Neurology.

Materials

Pre-implementation stage

Physicians' survey

During the pre-implementation stage, the authors, who also served as educators, conducted an exploratory survey in early 2022. The survey was distributed to national neurologists via email, with contact information obtained from various sources such as the Egyptian Society of Neurologists registries, workplaces, national conferences, and seminars. Personal outreach was also done through phone calls and WhatsApp. The survey, administered using the Google

online form tool. It consisted of 32 questions. Respondents were required to answer all questions except for the optional date of birth (<https://docs.google.com/forms/d/1DJjK3dmU-2Mw8RI1TGXbDWpW8Cjb1xyJP5OaehQcZ70/>). The survey recruitment spanned a 3-month period. The survey questions are provided in Appendix #1 (supplementary file). Data collection encompassed various aspects: (1) Demographics including email address, name, gender, age, role as a physician, medical career degree, current workplace, practice location, number of years in clinical practice, healthcare system type in workplace, and past genetic education/training; (2) Familiarity with genomics including relevance to specialty, knowledge of genomics and technologies, self-rating of genetic/genomic knowledge, prevalence of hereditary neurologic diseases, familiarity with genomic technologies, and availability of clinical guidelines for genetic service; (3) Clinical application of genomics including confidence in genomic knowledge, comfort in applying genomics, past genetic/genomic test orders, willingness to refer patients to genetic specialists if the option is available, availability of genomic resources, barriers to genomic practice, and existence of population genomic database; (4) Preferences and styles for genomic education and training including the need for additional education, updating genetic curricula for students and postgraduates, and desired educational styles and course contents.

The development and design of the genomic education and training program

This focused on selecting the appropriate audience, educators, course setting, and interventions. Educators included academic Neurology consultants and peer experts in genomics with relevant qualifications and experience. They possessed undergraduate and postgraduate education qualifications and demonstrated a strong interest in genomics, along with extensive experience in postgraduate genetic/genomics research. The target audiences/trainees/learners comprised doctors enrolled in the Neurology and Psychiatry residency program seeking a master's or MSc degree, doctors holding an MSc in Neurology and Psychiatry (specialists), and doctors enrolled in the M.D. degree program in Neurology (assistant lecturers). Individuals with and without prior knowledge or skills in genomics were welcomed to participate. The course was designed to take place at a University hospital and integrated into existing postgraduate educational programs and national genomic educational initiatives. No additional paperwork was required for the implementation of the courses, as it involved updating and expanding the existing postgraduate genetic curriculum. The courses were offered free of charge.

Implementation stage

This included the development of the structured program framework and the course outline. The design of the educational course was guided by survey responses and information from existing genetics/genomics education programs (Gaff et al. 2007; Korf et al. 2014; Nisselle et al. 2021). Our program shares common educational and clinical objectives with other existing and published programs. The intervention duration was 24 months (June 2022 to July 2024) for initial assessment. The interventions included: (a) Face-to-face learning through scheduled meetings (once a week for 6 consecutive weeks every 3–4 months, each lasting approximately 2.5–4 h), (b) Providing free online genetics/genomics courses as supplementary learning resources (<https://www.hugo-international.org/genetic-genomic-educational-resources/>), and (c) Case-based learning and group discussions during workshops. More meetings and patient outreach were conducted when free or funded genomic tests were available.

The course contents included: (a) Basic and updated genetics/genomics knowledge: This section provided a clear and concise introduction to fundamental principles in genetics/genomics, as well as the new testing methods. The information was presented in simple language, supported by text, images, and diagrams to enhance understanding. (b) Clinically-relevant case study: This component was highly valuable for clinicians, as it focused on common clinical learning objectives. The interpretation of test results was discussed within the context of real patient's presentation, without delving into the specifics of test methodology and variant interpretation processes.

Feedback evaluation from trainees about the effectiveness of the learning process

The trainee has to attend $\geq 80\%$ of the course activities at any point of time during the 24 months of the initiative to be considered in the evaluation assessment of the program. Evaluation involved analyzing trainees' responses through various methods: (1) Short quizzes and group discussions were conducted at the end of each educational meeting to assess understanding of the delivered knowledge; (2) Feedback was collected from a two-question survey completed after the course. Trainees were asked to rate their satisfaction with the learning materials, and rate their satisfaction with the case discussions on a scale of Poor, Good, Very good, or Excellent. (3) Comparing pre- and post (after 6 months)-course exam scores to measure knowledge change. (4) Feedback was also gathered through a one-question survey about the most recent genetic service activities applied in clinical practice within 6 months after course completion.

Data analysis

The analysis was conducted using the Statistical Package for the Social Sciences (SPSS), version 21.0 (International Business Machines Corporation, Armonk, New York, USA). Prior to the statistical analyses, all data were checked for outliers, skewness, and homogeneity of variance to select appropriate statistical tests. Categorical data were presented as numbers and percentages, while continuous data were presented as median (minimum to maximum). Comparative statistics were performed using Kruskal-Wallis H test and Chi-square (X^2) test. Linear modeling analysis was done for comparative analysis (pre- and post-course). A Bonferroni correction was applied to reduce the likelihood of errors and false positive results. Statistical significance was set at $\alpha < 0.05$.

Ethics approval and informed consent

The study protocol adhered to the ethical standards set by the national research committee of the Faculty of Medicine of Assiut (ID: AUFM_Neuro_00054/2022) and Al-Azhar University Hospitals, Assiut, Egypt, in line with the principles of the Helsinki Declaration and its subsequent revisions. Participants gave their written informed consent to take part in the study.

Result

Results of the pre-implementation stage (responses to the survey questions)

Demographics

The survey was sent to 200 neurologists, and 85 (42.5%) responded (55 male, 30 female). Their median age was 34 years (range: 27–62 years), with a median of 12 years in clinical practice (range: 3–35 years). About 53% had 3–10 years of clinical experience. 86% worked in university hospitals, while 14% worked in general (public) hospitals, all in urban areas. Approximately 60% saw patients from both urban and rural areas. 61% also worked in private clinics, and 50% were residents in Neurology and Psychiatry departments. All had some genetic education during medical school, but it was limited, accounting for less than 1% of the overall educational curriculum. The focus was on basic traditional concepts and common examples of genetic diseases. Around 53% of students were educated during a period of rapid advancements in genomics and related technologies, 35% received postgraduate genetic training,

and only 7% participated in genomic research projects (Table 1). Physicians reported that most patients (64.7%) paid for healthcare out-of-pocket (i.e., self-payment).

The familiarity of the field of genomics and genomic testing

Neurologists emphasized the importance of genomics in their field, especially for diagnosing hereditary neurologic diseases, providing counseling, determining disease susceptibilities, and conducting population surveillance. They acknowledge a high prevalence of hereditary neurologic diseases in clinics (around 20–30%) but expressed limited knowledge in genomics, with two-thirds sharing this concern. Their familiarity with new genomic tests varied, with most knowledgeable about karyotyping, single gene tests, gene panel tests, and single nucleotide polymorphism (SNP) arrays. Nearly half mention that genomic tests are not available in the country, and there is a lack of clinical guidelines for genetic services for patients with hereditary neurologic diseases. The majority (76%) are unaware of or unsure about the availability of genomic population databases and do not recognize their importance.

The application of genomics in practice

The majority (47%) of clinicians was aware of the limitations in their genomic education and training, and they rated themselves poorly in terms of competence and confidence in using genomic testing in clinical practice and interpreting test results. Only a few clinicians had previously ordered genomic tests for patients [single gene test=6; gene panel test=15; whole exome sequencing (WES)=12]. Most clinicians (76%) indicated that they would not accept referrals to other healthcare providers (clinical geneticists or counselors). Clinicians cited several barriers to the application of genetic services and ordering genetic tests. The top 5 barriers, in order of frequency, were: the complexity of genomics (100%), the cost of genomic testing (88%), genomic illiteracy (82%), work overload (70.6%), and lack of treatment options for genetic diseases (71%).

Preference styles for genomic education and training

There is a growing interest in implementing scalable genomic education. The preferred method for genomic education involves face-to-face introductory courses and case-based learning that focuses on interpreting test results in the context of a patient's presentation. A majority (70.6%) of respondents believe that updating genetic curricula in medical education and postgraduate courses is essential.

Table 1 Results of survey respondents

Survey items	Respondents (n = 85)	P-value
Demographics		
Gender		
Male	55 (64.7%)	0.01
Female	30 (35.3%)	
Age; years	27- 62	
Median	34	
27–35	35 (41.18%)	0.03
36–45	18 (21.18%)	
46–55	10 (11.76%)	
56 - 62	22 (25.88%)	
The number of years in clinical practice; years	3–35	
Median	12	
3–5	30 (35.29%)	0.01
6–10	15 (17.65%)	
11–15	12 (14.12%)	
16–20	18 (21.18%)	
>20	10 (11.76%)	
Work place		
Academic institute	73 (85.88%)	0.001
General Hospital (Neurology specialists)	12 (14.12%)	
Location of practice		
Urban	15 (17.65%)	0.01
Rural	20 (23.53%)	
Both	50 (58.82%)	
Job position		
Academic consultants (Professors and associate professors) (M.D. degree)	22 (25.88%)	0.003
Lecturers in the department of Neurology and psychiatry (M.D. degree)	10 (11.76%)	
Assistant lecturers in the department of Neurology and Psychiatry (MSc degree registered for MD degree)	10 (11.76%)	
Residents in the department of Neurology and Psychiatry) (registered for MSc degree)	43 (50.59%)	
General Hospital (Neurology specialists) (MSc degree)	12 (14.12%)	
Healthcare services		
National health insurance	12 (14.17%)	0.0001
A private health insurance (private centers)	8 (9.41%)	
Self-payment (out-of-pocket)	55 (64.71%)	
Combined	10 (11.76%)	
Past genetic education		
Medical school	85 (100%)	0.0001
Postgraduate educational activity	30 (35.29%)	
Research project	6 (7.06%)	
Familiarity with the field of genomics and its technologies		
Relevance of the field of genomics to specialty	85 (100%)	-
Self-rating for current knowledge of genetic/genomic field and its technology		
Poor	52 (61.18%)	0.001
Good	12 (14.17%)	
Very good	10 (11.76%)	
Excellent	11 (12.94%)	
Familiarity of the field of genomics		
Yes	65 (76.47%)	0.001
No	20 (23.53%)	
Familiarity with different types of genomic technologies		
Yes	70 (82.35%)	0.001
No	15 (17.65%)	
Familiarity with the following genomic tests		
Karyotyping	70 (82.35%)	0.01

Table 1 (continued)

Survey items	Respondents (<i>n</i> = 85)	<i>P</i> -value
Single gene test	45 (52.94%)	
Chromosomal microarray	6 (7.06%)	
SNPs array	30 (35.29%)	
Gene panel test	45 (52.94%)	
WES	30 (35.29%)	
WGS	0	
None	15 (17.65%)	
Availability of genomic resources in the country or workplace		
Yes	30 (35.29%)	0.01
No	40 (47.06%)	
Do not know	15 (17.65%)	
Existence of clinical guideline for genetic service		
Yes	15 (17.65%)	0.001
No	45 (52.94%)	
Do not know	25 (29.41%)	
Existence of genomic population database		
Yes	20 (23.53%)	0.01
No	30 (35.29%)	
Do not know	35 (41.18%)	
Application of genomic in clinical practice		
The perceived need for genomics in clinical practice		
Yes	60 (70.59%)	0.001
Do not know	25 (29.41%)	
Self-rating of confidence in the current genomic knowledge to answer patient's questions		
Poor	40 (47.06%)	0.01
Good	10 (11.76%)	
Very good	15 (17.65%)	
Excellent	20 (23.53%)	
Self-rating of comfort in application of genomics in practice		
Poor	42 (49.41%)	0.01
Good	18 (21.18%)	
Very good	15 (17.65%)	
Excellent	10 (11.76%)	
Ordering of genetic/genomic tests in the past		
Yes	32 (36.75%)	0.01
No	53 (62.35%)	
Accepting referral to clinical geneticist or genetic counseling if the option is available		
Yes	20 (23.53%)	0.01
No	45 (52.94%)	
Do not know	20 (23.53%)	
The concepts for the potential value of genomics in medicine		
- Investigating disease cause(s) or diagnosis	85 (100%)	-
- Facilitating family planning	85 (100%)	
- Determining disease susceptibilities	40 (47.06%)	
- Disease surveillance (screening practice)	60 (70.59%)	
- Pharmacogenomic testing	20 (23.53%)	
- All of the above	40 (47.06%)	
- None of the above	0	
Barriers for application of genomics in practice		
- The complexity and huge volume of genomic data	85 (100%)	-
- Genomic illiteracy	70 (82.35%)	
- Uncertain nature of the generated genomic data and lack of confidence in the reliability of next-generation sequencing technology for clinical application	20 (23.53%)	
- The cost of genomic test	75 (88.24%)	

Table 1 (continued)

Survey items	Respondents (<i>n</i> = 85)	<i>P</i> -value
- There is no treatment of genetic diseases	60 (70.59%)	
- Work overload	60 (70.59%)	
- The potential for genomic tests to cause psychological harm and the burden of having a potential responsibility to disclose additional findings outside the original cause of testing	30 (35.29%)	
- Ethical issues	50 (58.82%)	
Genomics educational and training needs and desires		
Preferences for future learning educational styles		
- Face-to-face learning	85 (100%)	0.001
- E-learning	35 (41.18%)	
- Case-based learning and workshops	85 (100%)	
- Others	-	
The perception of upgrading the medical student genetic/genomic education		
Yes	60 (70.59%)	0.001
No	0	
Do not know	25 (29.41%)	
The perception of the importance of the population genomic databases		
Yes	42 (49.41%)	0.01
No	35 (41.18%)	
Do not know	8 (9.41%)	

Approximately half of the respondents do not recognize the importance of a population genomic database (Fig. 1).

Results of the implementation stage

Framework of the course

The initiative spanned two years, from June 2022 to July 2024, with each course had three educators and an average of 45 learners, typically 20–30 participants per course. The educators were academic consultant neurologists ($n=3$). They were experts in genomics with extensive experience in research projects and educational events. They delivered key genomic knowledge, identified suitable patients for tests, selected appropriate genetic tests, interpreted results, and facilitated small-group discussions. Additionally, they provided learners with free online genetics and genomics resources to enhance their understanding (<https://www.hugo-international.org/genetic-genomic-educational-resources/>). The trainees included residents in the Department of Neurology and Psychiatry (enrolled in the MSc program) ($n=30$), assistant lecturers of Neurology (holding an MSc and enrolled in the M.D. program) ($n=10$), and neurology specialists (holding an MSc and/or enrolled in the M.D. program) ($n=5$) ($P<0.0001$). Residents and assistant lecturers worked in the University hospital, while neurology specialists worked in general or public hospitals ($P<0.0001$). Some learners attended multiple courses, with approximately 15–20 individuals participating in 3–4 courses due to their work in the same setting. About 33–44% of trainees attended 3–4 course sessions, particularly case discussions,

due to their work in the course setting. The courses were conducted face-to-face, totaling seven courses over 24 months. Each course comprised of six meetings, held weekly for six consecutive weeks every 3–4 months and lasting 2.5 to 4 h per session. The first three meetings focused on basic introductions, while the remaining three were dedicated to case-based learning, with 2–3 cases discussed per session.

Course outlines

Genomic education and training courses covered a variety of topics, including an introduction to human nuclear and mitochondrial genomes, inheritance patterns, family history importance, gene expression regulation, genomic variations, classification of genomic variants, and different types of genetic tests. Case-based learning focused on skills such as analyzing family pedigrees, selecting appropriate genetic tests [e.g. karyotyping, single gene test, chromosomal microarray, gene panel test, WES, whole genome sequencing (WGS), others], interpreting test results, determining the pathogenicity of variants, understanding genotype-phenotype correlations, developing clinical action plans, and managing genetic conditions (e.g. genomic approaches to treatment, counseling, risk assessment, segregation analysis, prenatal screening, etc.). The training emphasized ethical considerations and the importance of proper test ordering.

Clinicians were advised not to order genetic tests in certain situations, such as when guidelines were unclear, the clinical phenotype was ambiguous, the test was outside their specialty, lack of adequate training, inability to interpret the results effectively and unclear implications of test results for

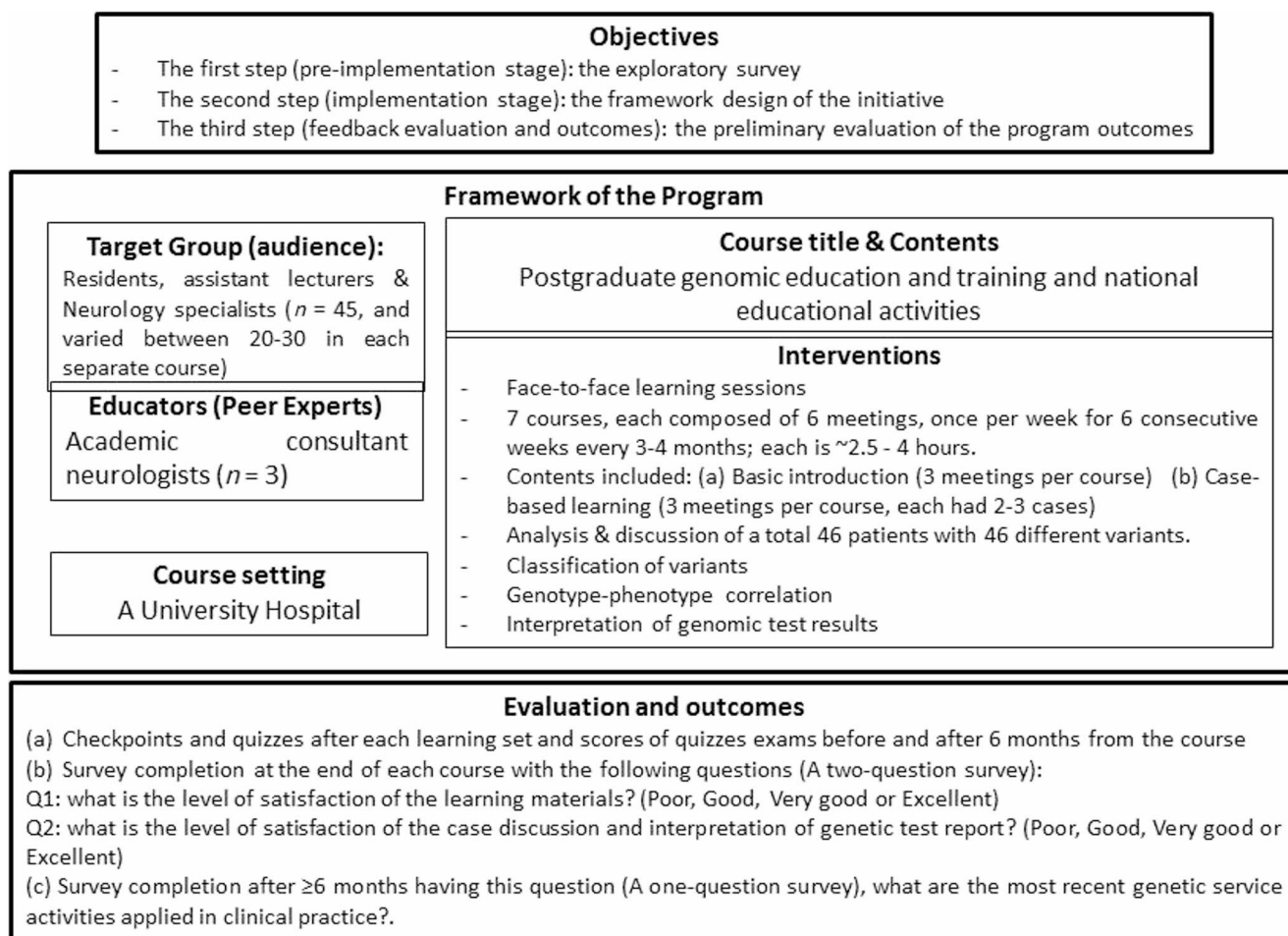


Fig. 1 Schematic representation of the objectives, framework of the initiative program and its evaluation

patients and their families. During the program, 46 patients with different genetic variants were analyzed and discussed. Cases included inherited muscular dystrophy such as dystrophinopathy ($n=18$), dysferlinopathy ($n=10$), calpainopathy ($n=4$), autosomal recessive hereditary peripheral neuropathy ($n=8$), and hereditary ataxia ($n=6$). Each case discussion involved a detailed history, phenotyping, inheritance patterns, variant classification, genotype-phenotype correlations, and interpretation of genomic test results.

Results of feedback evaluation and outcomes

The trainees' correct responses to checkpoints and quizzes improved significantly after each learning session, ranging from 60% to 100% compared to 20% to ~50% before the course, indicating a significant increase ($P < 0.001$) in knowledge scores in genomics in the post-course evaluation compared to pre-course. The average rating for the usefulness of the learning materials and satisfaction level with case discussions and interpretation of genetic test results was very good (20% or 9/45) and excellent (80% or

36/45), respectively. Their preferences for ordering genetic testing included single gene tests ($n=26$, 58%) or gene panel tests ($n=19$, ~42%) for non-complex neurologic phenotyping, such as hereditary muscular dystrophies, Huntington's disease, spinocerebellar ataxia, etc. Approximately 71% ($n=32$) of the trainees expressed intentions to integrate genetics/genomics into their practices in the future. Many were cautious about requesting WES or WGS without consulting peers due to the high cost and the vast amount of information produced, which could include secondary findings and ethical considerations. Neurologists reported engaging in new genomic activities in their real-world practice six months after completing the genomic course, such as ordering genomic tests ($n=12$, 26.67%) for patients with specific conditions like Duchene (DMD) and Becker (BMD) muscular dystrophies and spinal muscular atrophy (SMA), and educating junior physicians in their discipline about genomic services ($n=20$, 44.4%) (e.g., importance of detailed phenotyping, family history, mode of inheritance, and construction of pedigree charts) (Table 2).

Table 2 Results of evaluation, trainees' feedback evaluation and outcomes

Evaluation	Respondents Before course (<i>n</i> = 45)	Respondents After course (<i>n</i> = 45)
Correct responses to short quizzes and group discussions after the end of each meeting; <i>N</i> (%)		
Median	9 (20%)	27 (60%)
Minimum	16 (35.67%)	36 (80%)
Maximum	23 (51.1%)	45 (100%)
<i>P</i> -value		0.001
Response to a two-question survey		
Rating satisfaction with the learning materials	-	
Poor		-
Good		-
Very good		9 (20%)
Excellent		36 (80%)
Rating satisfaction with the case discussions	-	
Poor		-
Good		-
Very good		9 (20%)
Excellent		36 (80%)
Response to one-question survey		
The most recent genetic service activities (within 6 months after course completion)		
Ordering genetic testing		
Single gene test	-	26 (57.78%)
Gene panel test	-	19 (42.22%)
Intentions to integrate genetics/genomics into in the future	-	32 (71.1%)
Educating junior physicians about genomic services	-	20 (44.4%)

Discussion

This study outlines the creation, execution, and assessment of a case-based genomics education program for Neurology trainees in Egypt spanning from 2022 to 2024. The program consisted of a pre-implementation needs assessment, course implementation, and evaluation. We considered a response rate of 42.5% for a survey of physicians on the use of genomics in practice within an acceptable and favorable range. Studies have indicated that the typical response rate for surveys among physicians ranges from 20% to 50% (Cho et al. 2013). Physicians are often difficult to reach due to their busy schedules and time limitations. Factors such as the perceived complexity of the field of genomics or lack of familiarity with genomics can also contribute to physicians' reluctance to respond to surveys related to clinical genomics and genomic medicine. The initial survey conducted before the program revealed key insights: (1) Neurologists acknowledged the importance of genomics in their field, (2) Many neurologists admitted to having limited knowledge

of recent genomics advancements and new genetic tests, (3) Neurologists lacked confidence in addressing genetic inquiries from patients and felt uneasy applying genomics in clinical practice, and (4) Neurologists expressed a desire for specialized genomic education and training. They favored in-person learning and strongly endorsed enhancing genomic education. The study results indicated that approximately 53% of respondents were educated during a period of rapid advancements in genomics and related technologies. However, they did not show a significant difference in self-assessed knowledge of genomics and its technologies compared to older neurologists ($p > 0.05$), indicating that the formal education received during a period of substantial technological advancements did not result in significantly higher knowledge of the subject matter. This finding is consistent to many previous studies (Kaphingst et al. 2016).

In this study, neurologists acknowledged the importance of genomics in their specialty and the high prevalence rates of hereditary neurological diseases in their clinics (around 20–30%). They were familiar with various genetic tests such as karyotyping, single gene tests, gene panel tests, SNP arrays, WES, and WGS. However, they acknowledged that genomic advancements and access to genetic testing in clinical settings were limited to a small group of specialists, approximately 7%, who were involved in international research collaborations.

Similar studies have demonstrated high levels of familiarity with genomics and genetic testing in specialties dealing with a high prevalence of genetic diseases like Neurology, Pediatrics, Oncology, Cardiology, and Gynecology (Salm et al. 2014; Jaitovich Groisman et al. 2017; Mladeni'c et al. 2024). Salm et al. (2014) conducted a survey to investigate the knowledge, attitudes, and practices regarding genetic tests among neurologists ($n = 163$) and psychiatrists ($n = 372$) listed on the American Medical Association. The study found that a higher percentage of neurologists (74%) had ordered genetic tests in the past 6 months compared to psychiatrists (14%). However, both groups expressed concerns about ethical issues and potential psychological harm to patients, with 50% reporting such concerns. The study also revealed that 49% of neurologists and 75% of psychiatrists did not have access to genetic counselors for patient referrals. Jaitovich Groisman et al. (2017) conducted a global survey of 204 neurologists to explore their perspectives on the benefits and concerns of using WGS for diagnosing hereditary neurologic diseases. The findings showed that neurologists who would or would not offer WGS to patients agreed on the benefits outweighing the drawbacks, which included financial and legal issues. However, approximately 54% of respondents felt inadequately informed to use WGS in practice. Mladeni'c et al. (2024) investigated the knowledge, behavior, and attitudes toward medical

genetics among Obstetrics and Gynecology, Pediatrics, and Neurology residents and specialists in Croatia. The study involved 182 participants who completed an online questionnaire. Results showed that neurologists had the highest overall knowledge score in medical genetics (76.7%), followed by pediatricians (80.5%) and obstetrician-gynecologists (70.2%). Neurologists also demonstrated the highest level of knowledge in genomics (83.3%) and were more confident in ordering genetic tests (69.6%), interpreting results (62.5%), and discussing them with patients (89.3%) compared to pediatricians and gynecologists.

In this study, the majority of neurologists expressed a lack of confidence in addressing patients' inquiries about their genetic conditions. They also indicated a low level of comfort or lack of familiarity with integrating genomics into clinical practice. Previous research has shown that physicians often feel under-informed about medical genetics and genomics, citing their inadequate prior training and knowledge as barriers to incorporating medical genetics into healthcare services (Houwink et al. 2012; Talwar et al. 2017; Hauser et al. 2018; McClaren et al. 2020; Hajek et al. 2022; East et al. 2022).

In this study, most neurologists recognized the importance of genomics and genomic testing. The top 4 reasons for using genomic testing were identified as: diagnosing genetic diseases (100%), aiding in counseling and family planning (100%), population surveillance (~71%), and determining disease susceptibilities (47%). However, they cited their lack of genomic knowledge as a major obstacle (82%), leading to a limited integration of genomics in practice. Other barriers mentioned included high cost of genetic testing and inadequate funding (88%), limited availability of these tests (65%), heavy workload (70%), and lack of treatment options for genetic diseases (70%), psychological concerns (35%), and ethical issues (59%). Previous studies reported that physicians worldwide have shown a keen interest in incorporating genetic information into their practice, provided they have the required knowledge and tools (Najafzadeh et al. 2013). In some parts of the world, only a few institutions have received funding for projects involving gene panel sequencing tests (<https://www.genome.gov/GenomeEd>; <https://www.jax.org/education-and-learning>; www.ashg.org/education/careers.shtml; <http://gcp.wustl.edu>). For instance, in Australia, the government has made WES/WGS testing available and funded for certain conditions. This initiative has led to a significant increase in referrals of patients in need of genomic testing from various medical specialties to clinical genetics services (Nisselle et al. 2021).

In this study, it was widely acknowledged that additional genetic/genomic education is necessary to enhance competency in integrating genomics into practice. The findings of this study align closely with similar data reported in high-income countries with better healthcare resources.

Chow-White et al. (2017) surveyed medical oncologists in British Columbia, Canada, with a response rate of 52.5%. About 42% of respondents expressed the need for improvements in the medical training program to enhance knowledge in genomics and its new technologies. The majority anticipated a significant impact of genomics on drug discovery (67.7%) and treatment selection (58%) within the next 5 years. The main barriers identified for the integration of genomics into practice were cost (61.3%), patient genomic literacy (48.3%), and the clinical utility of genomics (42%). In a study by Diamonstein et al. (2018), an electronic survey was distributed to 157 physicians in Texas through state physician organizations. The responses varied among different specialties. Around 50% were aware of genetic testing and services in the area, 75% noted the increasing impact of genetics on medicine, 61% reported more discussions about genetics in daily practice over the past 5–10 years, and 20% considered genetics very important to their specialty. More than two-thirds rarely or never referred patients to genetic counselors or clinical geneticists. Over 75% expressed interest in genomic education and raising awareness of genetic services and referral processes. Kathrens-Gallardo et al. (2021) sent a link to an anonymous 49-question Red-Cap survey to coordinators in October 2017 to distribute to registered obstetrician-gynecologists in residency programs in the USA or Canada. Eighty two residents responded, stating that they obtained genetics information through discussions with physicians, lectures, courses, and publications. The majority (81–85%) emphasized the importance of attending educational courses to enhance their genetics knowledge, but 28% did not see the value of reinforcing concepts learned in course work. Most reported a low level of confidence in providing genetic services, particularly in hereditary cancer counseling. They highlighted the need for ongoing clinically relevant genomic education and training.

In this study, most physicians preferred face-to-face learning, such as case-based learning over online methods for genetics/genomics education. Previous studies indicated that despite the availability of online resources, around 90% of physicians are willing to attend in-person courses or workshops as online modules alone may not be as effective without clinical application models (Crellin et al. 2019), problem-based learning, competency-based training, and experiential learning modules (Houwink et al. 2012; McClaren et al. 2020; Nisselle et al. 2021; East et al. 2022). Workshops that involve result interpretation and application with experts are considered most beneficial (Bishop et al. 2019; Chen et al. 2019; Evans et al. 2020). McClaren et al. (2020) conducted interviews with 86 physicians from various medical specialties across Australia, excluding clinical geneticists, to investigate the continuing education needs in genomic medicine. The authors identified emerging concepts

related to educational needs through ongoing comparisons and discussions. Physicians acknowledged the importance of experiential learning in genomic medicine for building confidence and skills in clinical genomics implementation. The main themes identified included tailoring education to specialties, case reviews, individualized approaches, peer interactions, and the need for more experience to develop confidence and skills. On the other hand, some studies have shown that approximately 70% of primary care physicians favor E-learning for genomic education. This preference is driven by time constraints and the need to enhance access to genetic services in underserved regions. They specifically seek out concise, reliable online resources, evidence-based guidelines, and personalized educational materials (Carroll et al. 2019; Evans et al. 2020; Chou et al. 2021).

In June 2022, we conducted a genomic education and training program consisting of structured regular meetings within the Department of Neurology as part of postgraduate and national educational activities. The new genomic activities included gathering interested peer experts, holding 7 courses with a total of 42 meetings over 24 months to discuss and interpret real genetic test results of 46 cases with hereditary neurologic diseases, and providing guidelines for genetic services for hereditary neurologic disorders. The program courses were conducted face-to-face, with a total of 45 trainees (residents, assistant lecturers and neurology specialists) over 24 months. The educators were academic consultant neurologists who also were peer experts in genomics. In this study, the course evaluation focused on the learners' perspectives. The relevance of the course was assessed by analyzing learner comments throughout the course, as well as their responses to an end-of-course survey and exams. In general, it is widely recognized that assessing the effectiveness of the learning process is crucial for enhancing and refining educational models or programs. Conducting follow-up surveys is a common method used in many studies (Chow-White et al. 2017; Diamonstein et al. 2018; Kathrens-Gallardo et al. 2021). In this study, the neurologists trainee showed increased genomic knowledge, as evidenced by score differences between responses to quizzes before and after the course and exams after 6 months from the course completion ($P < 0.0001$). They felt more comfortable referring patients to order genetic tests (28%) after receiving advice from peer experts and they educated junior residents on skills for genetic services (44%) including detailed history, phenotyping and family pedigree construction. Also all expressed intentions to refer patients for genomic testing in the future, particularly when there are available fund resources. Previous research has shown that physicians who participated in live training interventions and experienced a boost in confidence levels regarding critical skills necessary for applying

genomics in medical practice. They showed a substantial increase in their confidence levels in reviewing, comprehending, and utilizing genomic test result reports to inform patient care outcomes (Chen et al. 2019; East et al. 2022). In the systematic literature review by Talwar et al. (2017), the authors conducted a search across 5 electronic databases from 1990 to 2016 to assess genetics/genomics education programs for non-genetic health professionals. They identified numerous programs tailored for this audience, with the majority being delivered through face-to-face instruction, while approximately 30% were theory-based. The curricula covered topics such as basic and applied genetics/genomics, ethical, legal, and social issues, genomic competencies, and recommendations for specific medical professional fields. The authors noted positive outcomes in areas like knowledge, attitudes, intention, self-efficacy, comfort level, and practice. However, they highlighted concerns about data validity and reliability. In 2019, Bishop and colleagues evaluated their Massive Open Online Courses by gathering feedback from 360 learners who completed the end-of-course survey and 14 mentors who guided the course. The participants indicated that the course effectively addressed learners' needs, offering a deeper insight into WGS and its healthcare applications. Learners particularly appreciated the interactive discussions with mentors online. In a recent study conducted by Nisselle et al. (2021), the authors examined the genomic medicine practices, perceptions, preferred models of practice, and continuing education preferences of medical specialists across Australia through a survey. The study included 409 physicians from 30 different specialties, with 20% being pediatricians. The majority (70%) worked in public hospitals in metropolitan areas. 53% reported access to local genetics services, and 54% had ordered or referred for gene panel or exome/genome sequencing tests in the past year. About 67% believed that genomics would have an impact on practice in the near future. Most participants reported conducting family history assessments and counseling, with intentions to order genomic tests in the future. Approximately 34% had recently completed genomic education, but only 25% of them felt comfortable applying genomics in practice.

In this study, preferences for the trainees regarding ordering genetic testing included single gene or gene panel tests for non-complex neurologic phenotyping, such as hereditary muscular dystrophies, Huntington's disease, spinocerebellar ataxia, etc. They did not agree to order WES or WGS by themselves because of the high cost of the tests, low availability, and additional findings provided by these tests which raise ethical concerns. Previous studies indicated that while the application of WGS is particularly relevant in neurology, as many conditions are difficult to diagnose, the use of WES or WGS in clinical settings has

raised a number of controversial scientific and ethical issues (Jaitovich Groisman et al. 2017; Prokop et al. 2018).

Data from Africa show that this study represents the first completed genomic education and training program initiative in Africa for non-genetic health professionals specializing in Neurology. Also data on genomic training in Africa show that the oldest program dates back to 1990. In 2017, the African Genomic Medicine Training (AGMT) Initiative was launched by the African Bioinformatics Education Committee (ABEC) in partnership with the Human Genetics Department at the University of Cape Town (African Genomic Medicine Training Initiative, https://training.h3abionet.org/AGMC_2016/outputs/). The AGMT aims to integrate Genomic Medicine into mainstream healthcare in Africa by enhancing training for healthcare workers. A survey form was created and distributed to the African Genomic Medicine Training Initiative Mailing list and other stakeholders. Responses were collected from 19 countries (Egypt was not included). The majority (77%) of responses came from academic institutions. The survey findings revealed that about 40% of departments offer regular training in Genetics and Genomics, mainly targeting medical students, while lab technologists lack professional development opportunities. Some institutions have discontinued training programs due to a shortage of trainers and resources. The reported challenges in establishing Genomics and Genetics training courses included a lack of expertise in curriculum development, limited interest from potential trainees, and insufficient funds, time, and resources. Recently in 2023, a professional development course was also launched in Africa to provide genomics and genetics education to medical doctors. The course is currently in progress and is designed to improve the understanding, attitudes, and skills in genetics and genomics for a diverse range of participants. This includes research medical doctors in the biomedical field, specialist medical doctors in genomics/genetics, as well as general medical doctors and recent graduates in their everyday practice. The course materials are delivered through a distributed virtual classroom approach, online lectures, and in-person attendance at various classrooms across Africa (Course Information: Intro to Genomic Medicine for Medical Doctors in Africa Training (2023); https://zivahub.uct.ac.za/articles/educational_resource/African_Genomic_Medicine_Training_Initiative_AGMT_/21256161). It is important to note that our initiative is distinct from other African genomics educational and training programs.

Limitations of the study

The potential long-term benefits of genomics training remain uncertain due to limited access to genomic tests in the workplace and inadequate resources and funding.

Conclusion

The study findings highlight the attitudes and knowledge gaps among Egyptian neurologists regarding the integration of genomics in hereditary neurologic diseases. While neurologists acknowledge the importance of genomics in their field, they also recognize their deficiencies in genomics knowledge and the application of genomic testing in clinical settings. The study has identified specific educational and training needs. The outcomes of our genomics educational and training program have been promising, with positive feedback from participants, supporting the ongoing education of neurologists. However, continuing education and training are essential. Educational initiatives should be evaluated based on real long-term changes in practice, confidence, and knowledge. Our future plans include: (1) Continuing genomic education and training activities for neurologists, including those who have completed previous courses, (2) Updating genomic education curricula for postgraduate doctors as academic medical specialists and addressing changing patterns of genetic healthcare services, (3) Engaging more peer experts, (4) Implementing long-term follow-up for proper evaluation, improvement, and redesign of the educational process, (5) Establishing objective measures to evaluate the impact of genomic education and training, such as hospital-based audits of clinical practice, (6) Enhancing international collaboration through scientific projects, symposia, and educational events to update genomic knowledge, share educational ideas and experiences, and mobilize resources, (7) Strengthening cross-specialty relationships and promoting the formation of multidisciplinary professional teams, and (8) Increasing cultural awareness of genomics and genomic services.

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Declarations

Competing interests The authors declare no competing interests.

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