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Human Genomics

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The HUGO Clinical Genomics & Genomic Medicine Education Survey: clinicians globally need and want genomic medicine training

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Abstract

Background The Clinical Genomics & Genomic Medicine Education subcommittee of the Human Genome Organization (HUGO) reports on the results of an exploratory genomic medicine survey of healthcare providers (clinicians) in low- and middle-income countries.

Results 281 clinicians from 17 low- and middle-income countries responded to the survey. Representing 20 medical specialties, only 13.6% of clinician respondents are comfortable when discussing genomics in clinical settings. Over 90% of clinicians recognize that genetics and genomics are relevant to their practice and that there needs to be genetics/genomic assessment of patients. Respondents report a wide range of learning style preferences including self-paced online (asynchronous) and live-online educational programming covering introductory content, clinical guidelines for monogenic disorders and when to utilize genetic or genomic testing.

Conclusions Clinician respondents recognize the importance of genomics and are eager for continuing medical education. Reponses from the survey suggest that a larger scale, second phase survey should be distributed globally.

Introduction

To better understand the educational needs of practicing clinicians, the HUGO Clinical Genomics & Genomic Medicine Education Subcommittee developed a survey to assess both clinician perceptions and concerns regarding the application of genomic medicine as well as their educational needs and learning style preferences for continuing genomic education. While preliminary, the survey identifies key concerns and perceived knowledge gaps of practitioners, highlights interest in genomics education and outlines preferred educational formats that may accelerate the application of genomics to medicine across the globe.

Over the last quarter century, promises of transformational change in the practice of medicine have been predicted regularly during the development and

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advancement of the field of human genomics [1, 2]. Progress in applying genomics to patient care has been made across a number of fields and sub-specialties, particularly in precision oncology, rare disease diagnosis, pharmacogenomics, and most recently, in the application of polygenic risk scores to common multifactorial diseases [3]. The application of genomic medicine to clinical practice has been most extensive across the higher income countries around the world. Broad and global application of genomic medicine in patient care has lagged behind the rapid pace of research and discovery enabled by low cost and advanced genomic technologies. Education empowers healthcare professionals to integrate genomics into their clinical practice, and practitioner training and education are important to realizing the full promise of human genomics. This, however, may require parallel improvements in the provision of local infrastructure, such as a stable electricity supply, and in equipment and facilities, including large-scale data analysis and storage capacities.

Previous surveys in 2016 and 2022 gathered broadly comparable data across a range of African countries [4, 5]. The African Genomic Medicine Training Initiative [4], with 33 complete responses from 19 countries, reported that only 41% of the sampled institutes provided training in genetics and genomics and that several training programs were terminated due to a dearth of lecturers. Existing training focused on medical students and laboratory technologists. The African Academy of Sciences (AAS) [5], reporting on 55 complete responses from 12 countries, provided policy recommendations across a range of topics. With regards to awareness, education, and training the AAS survey found that only 14% of respondents thought scientists in their countries were ready to implement precision or genomic medicine and 67% of respondents felt that a lack of genetics-focused education of physicians and caregivers was a major barrier to the implementation of genomic medicine.

Table 1 Self-reported countries of respondents optional response, "In which country do you practice?"

Countries reported	# Responses
Argentina	4
Australia	1
Brazil	2
Colombia	1
DR Congo	1
Ecuador	1
Egypt	81
India	5
Indonesia	16
Malaysia	5
Mexico	3
Nepal	1
Scotland	1
South Africa	2
Tanzania	5
US	1
UK	2

Promoting genetic and genomic literacy of patients and the public is an associated challenge that also needs to be addressed [6–7]. Achieving the full promise of genomic medicine will require ongoing public education and information exchange between clinical care staff and patients, allowing for informed decision making and appropriate utilization of genetic and genomic information. Genetic literacy remains difficult to measure [6]; however, a number of national population genomic projects [7] and large-scale efforts such as the *All of Us* Research project in the United States are working to increase genomic literacy across a range of audiences.

Methods—The survey

The HUGO *Clinical Genomics* survey was developed in late 2022 and distributed from April 2023 to May 2024. The survey aimed to measure clinicians' understanding of and concerns regarding the application of genomic medicine to patient care and to inform future educational content development and educational delivery modalities.

The survey targeted healthcare professionals from low- and middle-income countries, and provided World Bank guidance so that respondents could characterize the countries where they practice. The anonymous Qualtrics™ online survey covered the following four general topics using 23 content questions and two personal questions (age and option to re-contact):

1. Demographics: medical specialty, location of practice, type of healthcare system, and past genetics education;
2. Familiarity with Genomics: current knowledge, comfort using or discussing genomic topics, use of clinical guidelines, and availability of local genomic resources;
3. Application of Genomics: relevance to their practice, and need for genetics in their specialty;
4. Education Need and Desire: clinical genomics topics of interest, learning style preferences, and need for medical education curricular updates or additions.

Survey participation was voluntary and it was permissible to answer some but not all questions. While 300 responses from 17 countries were collected (Table 1), no specific questions required an answer for survey completion and the number of responses provided for each question showed a wide range (120–220); 175 respondents reached the final content question of the survey.

The survey questions and number of responses per questions can be found in the supplementary materials.

The survey was distributed via HUGO newsletters, genomic education meetings, and broadcast emails sent by committee members to their professional networks. Respondents included medical doctors, nurses, and other direct care providers. Survey responses from genetic counselors and clinical geneticists were removed from the data because practitioners in these fields may have advanced training and knowledge in genetics and genomics.

Results—Multi-country, multi-specialty responses

A total of 300 responses were collected, and the results described here do not include 19 responses from genetic counselors and clinical geneticists. For each of the following, the denominator represents the number of individuals who responded to the question. The respondents were primarily from Africa 90/133 (67%) and Asia 27/133 (20.3%), with fewer responses from South and Central America 9/133 (6.8%) (Table 1). There was a relatively large response from Egypt ($n = 81$, of 133 who provided the country where they practice). Clinician respondents came from 20 specialties (Table 2A), with the most common being: Neurology 40/132 (30%); General Practice/Internal Medicine 18/132 (14%); Psychiatry 18/132 (14%); and Pediatrics 17/132 (13%). Table 2B reports others clinical roles as self-reported in an open text field. Most respondents were from middle-income countries (newly emerging economies or emerging economies), 101/142 (71%), with slightly more than a quarter, 37/142 (26%), from low-income (developing) countries. Respondents had varied durations of clinical practice. One-third of respondents 46/141 (32.6%) reported practicing for more than 20 years. Over 90% of respondents, 148 of 160 (92.5%) reported having received basic training in genetics. Most of the clinician respondents (81%; 195/241 with multiple responses allowed) reported some familiarity with the standard or traditional genomic technologies, including microarrays, SNP chips, gene panel sequencing, and whole exome and whole genome sequencing. However, considering the number of years of their clinical practice and the recent emergence of human genomics and genomic medicine, it is likely that many respondents received genetics training prior to or soon after the completion of the first draft human genome.

Table 2 A and B—Column A: Self-reported clinical specialty from survey pick list; Column B: open text clinical specialty, self-reported if specialty was not found on pick list

A: Role as practicing physician	#	B: Additional roles (self-reported clinical role)	#
Neurology	40	Allergy and immunology/allergist	2
General medicine/Internal medicine	18	Audiology	1
Psychiatry	18	Biochemistry	2
Pediatrics	17	Cancer clinical and molecular cytogeneticist	1
Otolaryngology	6	Clinical Geneticist	2
Orthopedic surgery	4	Dentist	2
Ophthalmology	4	General Practitioner	3
Urology	3	Geneticist	3
Hematology	3	Germinal variant analyst	1
Anaesthesiology	2	Molecular pathologists with clinical DX	1
Endocrinology	2	Investigator	1
Immunology	2	Laboratory Genetics	1
Obstetrics/Gynecology	2	Laboratory Medicine	1
Rheumatology	2	Laboratory Scientist/Genomic technologies	1
Cardiology	1	Laboratory Technician	1
Epidemiology	1	Medical Director	1
Geriatric medicine	1	Medical Scientist	1
Nephrology	1	Pediatrician	1
Pulmonology	1	Pharmacist	1
Radiology	1	Audiology Consultant	1
Total	129	Urology Surgery	2
		Total	30

Total replies 132, 3 participants gave 2 responses

The survey revealed the extent of gaps in clinicians' knowledge and comfort levels regarding genomic medicine. Only 32/147 (21.8%) felt sufficiently knowledgeable to answer patient questions about genomics, and only 18/147 (13.6%) reported very good or excellent levels of comfort when discussing genomics in clinical settings.

Clinicians expressed several concerns regarding the integration of genomic medicine into their practices. The top concerns clinicians identified (Fig. 1) with multiple responses allowed) included insufficient knowledge about currently available genomic technologies, their capabilities and interpretation of their results 109/150 (22%), a lack of treatments for genetic disorders 104/150 (21%), and the complexity and volume of genomic data 77/150 (15.5%). Despite these concerns, 153/159 (96%) clinicians recognized the need for genomic assessment in clinical practice (Fig. 2, bottom). When asked about the potential value that genomics would bring to their practices, clinicians selected investigating disease cause(s), facilitating diagnosis and family planning 134/150 (89.3%), guiding screening practices (i.e., disease surveillance) 91/150 (60.7%), and determining disease susceptibilities and motivating healthier lifestyles 88/150 (58.7%) as the top three likely benefits. Amongst respondents, there was agreement regarding the relevance of genomics to clinical practice, with 148/159 (93.1%) acknowledging the importance of genomic medicine. Clinicians were aware of the importance of having specific genomic information for patient populations, as 126/142 (88.7%) respondents

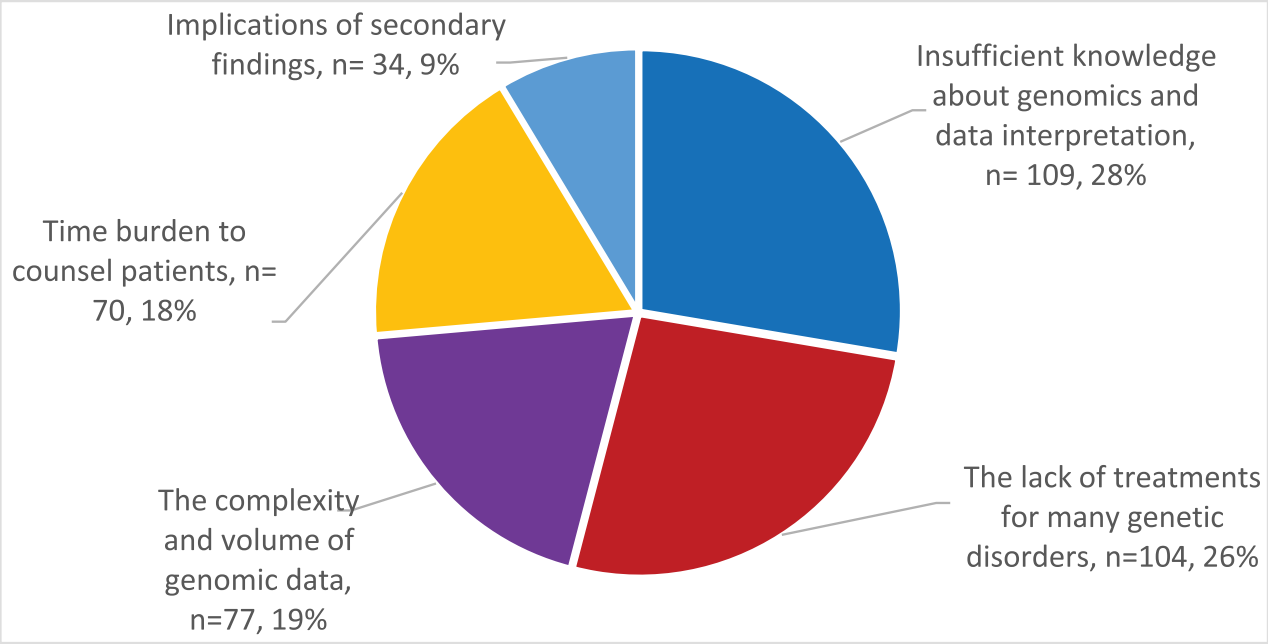

Fig. 1 Survey respondents' concerns about the clinical utility of genomics. The survey asked "Do you have concerns about the current clinical utility of genomic medicine?" One-hundred and fifty respondents selected from a list of possible concerns that ranged from insufficient knowledge to implications of secondary findings

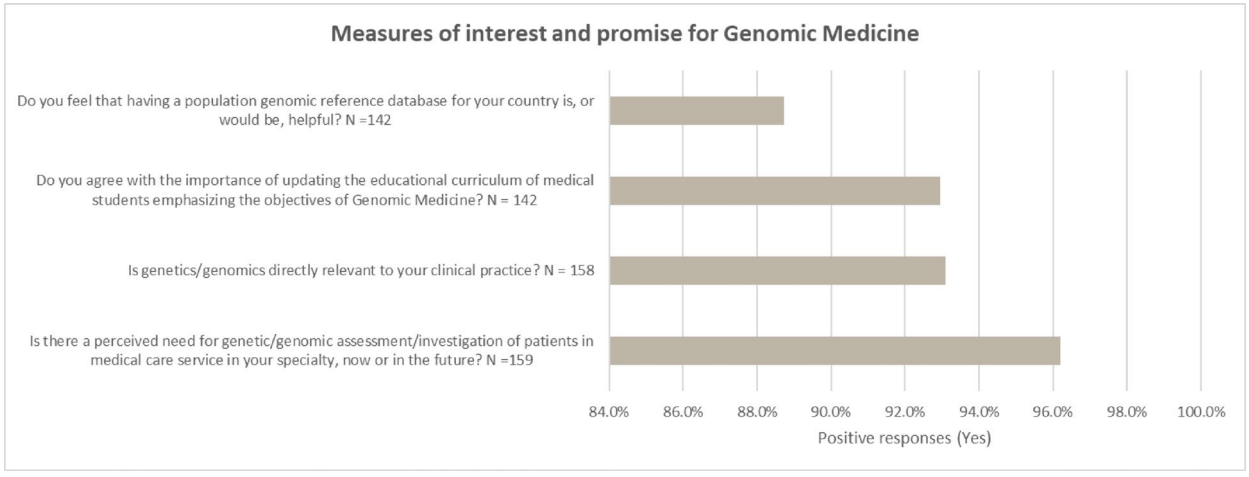

Fig. 2 Healthcare provider interest in genomic medicine. Four questions in the survey assessed clinician interest in, or relevance of, genomic medicine to their practice. Approximately 96% of respondents view genomic medicine, access to education in the field, and regional human genomic resources as important

acknowledged the importance of population-specific genomic reference data for enabling genomic medicine.

The HUGO Education Committee was especially interested in learning style preferences that could be deployed most effectively to serve busy clinicians. The survey identified particular interest in self-paced 106/145 (73.1%) and live-online courses 68/145 (46.8%) covering introductory content 104/154 (71.7%), delivered as modules 91/145 (62.7%), as favored over accredited in-person workshops 51/145 (35.1%) or accredited webinars 47/145 (32.4%) (multiple responses allowed). The top three topics selected for future educational programs included: (1) Concise and easy-to-understand introduction on the available genomic technologies, covering their capabilities, laboratory and bioinformatics testing processes and personalized medicine, targeted treatments, or pharmacogenomics; (2) Clinical guidelines for monogenic disorders; and (3) When testing could be useful.

Discussion—Signals for additional education

The survey highlights the need for comprehensive educational efforts to bridge the gap between research-based genomic advancements and clinical practice. While the sample size is small, the responses provide four important signals with regards to realizing the promise of genomic medicine.

- Clinicians recognize the importance of genomic medicine across a range of practice areas;
- Clinicians have a range of concerns about their knowledge and the application of genomics to their practices;
- There is willingness from clinicians across lower-, and middle-income countries for additional genomic education;
- Self-paced or live-online modular education may be a viable and preferred option for clinicians, whether they have practiced for less than 5 years or more than 20 years.

The survey provides valuable, but only preliminary, data necessary to plan and execute genomic medicine education programming around the globe. It is acknowledged that the data may be skewed by selection bias. The HUGO Education Committee will translate the survey into Arabic, Bengali, Chinese Mandarin, French, Indonesian, Portuguese, Russian, Spanish and Urdu. An aggressive second phase of the survey will need to use targeted distribution strategies to enhance global reach and participation, and the HUGO team welcomes communication from medical societies around the world such that survey distribution collaborations can be established.

HUGO proposes to undertake global genomic medicine education via its continued development of educational web-pages, open-access peer-reviewed publications, development of microcredentials in genomics and personalized medicine, as well as through open-access virtual and in-person courses workshops. Finally, the organization plans to continue to provide genomics educational workshops on relevant topics at HUGO international conferences and to work with other active global genomic educational organizations and programs.

Conclusion

Understanding clinicians' knowledge and educational preferences is important for integrating genomic medicine into clinical practice. The findings from this survey effort provide beneficial insights into the steps necessary to expand clinical applications of genomics to ensure that all of humanity benefits from the advances in genomic technologies. By addressing the identified knowledge gaps and concerns, through targeted educational efforts and funding, the HUGO Education Committee stands ready to support clinicians in effectively integrating genomics into their practice to improve patient care.

Supplementary Information

The online version contains supplementary material available at https://doi.org/10.1186/s40246-025-00841-7.

Supplementary Material 1

Author contributions

The original survey that formed the basis of the report was written by ET, QA, DK, IS, LS, and SH. AH and CW edited the survey. CW administered the survey and wrote the main manuscript text with edits from all authors. ET and AH provided CW with additional review and final edits of the manuscript.

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Data availability

Data from the exploratory survey can be shared upon request of the authors. The survey will be re-used as a larger global survey of a larger group of clinicians from low and middle income countries.

Declarations

Competing interests

The authors declare no competing interests.

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