Perspectives of Clinical Genetics in Wisconsin: A Qualitative Analysis to Enhance Awareness About the Specialty and Guide Improvement

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ABSTRACT

Introduction: Genomic advances have launched medicine into a new frontier of preventive and personalized care, driving increased demand for services amid a recognized provider shortage. We aimed to highlight perspectives of clinical geneticists to support growth and sustain genetics care for patients in Wisconsin in the face of rising demand.

Methods: A literature review informed the development of interview questions for appreciative inquiry-guided interviews. University of Wisconsin-affiliated geneticists were interviewed in the summer of 2024 about their experiences and insights. Transcripts were analyzed using an open inductive process, and hierarchical qualitative analyses were conducted to identify themes and opportunities to increase interest in genetics as a career.

Results: Regarding their profession, 92% of respondents identified "problem-solving" as a favorite aspect, highlighted the benefits of interprofessional collaboration, and identified diagnostic improvements as the most notable change over their careers. Regarding the future of genetics, 75% expressed excitement about improving treatments, anticipated a shift in their roles from diagnostics to management, and encouraged trainees across health professions to engage in genomic medicine learning opportunities.

Conclusions: Clinical geneticists expressed strong enthusiasm for their careers, noting problem-solving, continuity of care, and patient diversity as key highlights. Their roles provide opportunities to lead interdisciplinary teams, collaborate across specialties, and engage in lifelong learning. Their experiences offer insight into future diagnostic and treatment advances, which will require innovative care delivery approaches, including greater delegation and expanded training for the broader health care workforce. These perspectives may help foster interest in clinical genetics among health care trainees.

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INTRODUCTION

Over the last decade, significant advances in the field of genetics have had a positive impact on patient care. With personalized approaches to preventive medicine, providers are making more accurate diagnoses, providing more effective treatments, and identifying risk factors to help mitigate health complications.[1] These advances range from pharmacogenetics—using genomic data to predict therapeutic responses and avoid adverse reactions[2]—to advances in prenatal care that can significantly influence health outcomes for future generations.[3]

As genomic testing becomes increasingly accessible, demand for these services has increased, although payer-related barriers remain.[4] Genetic testing has rapidly evolved from sequencing single genes to genome- and exome-wide sequencing, placing increasing burdens on the already limited pool of qualified clinical geneticists.[5] These test results yield large volumes of data that require substantial time and effort to interpret. Often, there are many variants of uncertain significance, necessitating additional testing, thoughtful follow-up, and investigation in the literature and the Clinical Genome Resource, a standardized international organization with expert panels that delineate genetic variants.[6]

The multisystem involvement and complexity of many genetic conditions—including incomplete penetrance, variable expressivity, etiologic heterogeneity, and diverse inheritance patterns—require that providers understand a broad spectrum of

symptoms presenting in patients and their families. Exome-wide sequencing is becoming a standard of practice to help diagnose some conditions, while genome-wide sequencing is becoming more readily available, resulting in increased demand for clinical genetics services.[7]

With this growing need, there is concern regarding multiple factors, including the limited number of trainees entering the field, with residency and fellowship positions going unfilled, and a workforce in which many clinical geneticists are approaching retirement.[8] Despite newer delivery models such as telehealth to improve access, limitations persist due to the overall shortage of providers.[9]

To address the demand for genetic services, we aimed to gain a deeper understanding of the field in Wisconsin by gathering insights directly from clinicians affiliated with the University of Wisconsin School of Medicine and Public Health (UWSMPH). Our goal was to identify strengths and successes that could inform quality improvement interventions to expand and sustain clinical genetics care for patients in Wisconsin and beyond.

METHODS

Project Design

A literature review spanning 6 years (2019-2025) was performed to outline the current state of clinical genetics in the United States. The following search terms were used: ("medical genetics workforce"[tiab] OR "genetics, medical"[tiab] OR "medical genetics"[tiab] OR "medical geneticist"[tiab] OR "clinical geneticist"[tiab] OR "clinical genetics"[tiab] OR "genetics, clinical"[tiab] OR "genetic services"[tiab]) AND (workforce[tiab] OR workforces[tiab] OR "work force"[tiab] OR staffing[tiab]).

Insights from the review guided development of 6 interview questions and probes to explore the perspectives of clinical geneticists currently or recently affiliated with UWSMPH, including changes they have observed in their practice over time, and perceptions of the field's future. The interview questionnaire was developed by research team members (IAS and EMP) using an appreciative inquiry approach[10] based on the "Discover and Dream" structure. This strengths-based approach emphasizes interviewees' positive experiences to identify opportunities for sustainable and transformative change.[11]

A preamble was used before each interview to outline study goals and obtain consent for recording and transcription. The University of Wisconsin-Madison Institutional Review Board (IRB) Quality Improvement/Program Evaluation Self-Certification Tool (irb.wisc.edu/is-it-research) was used to determine that this project met criteria for quality improvement, met the federal definition of research pursuant to 45 CFR 46, and did not require IRB review.

Participant Recruitment

To recruit interviewees, 16 emails were sent to all current physicians practicing genomic medicine, 2 former faculty members, and 1 former trainee affiliated with UWSMPH. Of the 15 who responded, 1 declined participation and 2 were unavailable within the 4-week study period. All 12 physicians who were available and willing were chosen as participants.

Table 1. Respondent Demographics

	Percent (n)
Primary certification	
Pediatrics	83% (10)
Secondary genetics certification	
Biochemical	33% (4)
Clinical	92% (11)
Molecular/Cytogenetics	8% (1)
Years of experience practicing genomic medicine	
0–9 years	25% (3)
10–19 years	33% (4)
20–29 years	8% (1)
30+ years	33% (4)
University of Wisconsin School of Medicine and Public Health Affiliation	
Current faculty member	75% (9)
Training (medical school, residency, or fellowship)	75% (9)
Former faculty member	17% (2)
Respondent race	
American Indian or Alaska Native	0%
Asian	17% (2)
Black or African American	0%
Hispanic or Latino	0%
Native Hawaiian or Other Pacific Islander	0%
White	83% (10)
Respondent gender	
Female	50% (6)
Male	50% (6)
Nonbinary/other	0%

Following each interview, demographic data were collected, including institutional affiliation, board certification, years of experience, and self-reported race and gender identity.

Data Analysis

Transcripts were generated from interview recordings and evaluated and corrected as needed to ensure accuracy. Using an open inductive process,[12] a codebook was developed from concepts identified in the transcripts. An independent coder (NJS) was recruited, and the codebook was used as a guide for the 2 coders (IAS and NJS) to independently code transcripts.

Initial intercoder reliability was approximately 90% (211/235 agreements). Following independent coding, the coders met to reach consensus on 24 discrepancies among the 235 codes. Using the finalized data, hierarchical qualitative analyses were performed to identify key themes and potential areas for intervention to address the clinical geneticist shortage.

RESULTS

Physician Interviews

Of the 12 current and former UW faculty members and trainees who participated, 6 identified as female and 6 as male; most identi-

Table 2. Discover Medical Genetics: Themes Identified Highlighting the Field

Themes,	% of Respondents (n)	Quote
Highlights of being a clinical geneticist		
Problem-solving	92% (11)	I like the problem-solving aspect; you're presented with unusual signs, symptoms, examination, features, laboratory results, trying to put that all together. It's like putting a puzzle together or being a detective, I imagine.
Continuity of care	67% (8)	We see them throughout their lifespan – for several of these disorders, we can have a child that would otherwise have significant developmental delay or may have passed away from that disorder – they can be happy toddlers coming into your office and then normal sullen teens and now… we get to help care for them as they have their own families too.
Patient diversity	50% (6)	In genetics, you deal with all populations, so I have patients from basically the newborn period through geriatric age patients… there's also diversity in terms of the kinds of issues they're dealing with, from intellectual and cognitive issues to physical issues… and there's a lot of long-term chronic issues but there's also acute issues… I really like that diversity.
Lifelong learning	33% (4)	We learn new things and get to apply new kinds of cutting-edge technology to patient questions that maybe could not have been answered a year or 2 years ago, that now we can answer… I find that sort of constant learning and interpretation of literature to real world, practical patient care very exciting and stimulating.
Good community	33% (4)	I think the people who are in genetics are awesome.
Public health/ advocacy	25% (3)	Newborn screening is a great public health program, and as clinical geneticists we are an important component to that public health program that really can provide great preventive care and help with morbidity and mortality for rare metabolic disorders.
Practice variety	8% (1)	It's also been rewarding in terms of related nonclinical issues like being able to participate in research and teaching and mentoring.
Roles of a clinical geneticist		
Multidisciplinary teamwork within clinic	92% (11)	In our clinic, I work with genetic counselors, nurses, medical assistants, a genetic counseling assistant, and also dietitians.
Multidisciplinary teamwork across specialties	83% (10)	Most of the multidisciplinary clinics or teams that we're involved with as geneticists deal with relatively rare conditions. When they involve other specialists, I mean the clinics that I think about are involving surgeons, sometimes neurosurgeons, sometimes other surgical specialists, dermatology is a very common accompanying service, neurology…
Leadership role	50% (6)	I had more of a leadership role in genetics, really making sure we had really strong interdisciplinary teams and conferences.
Multidisciplinary teamwork across institutions	33% (4)	I find that when you reach out for help, people are really willing to give you advice on what their experience has been and also let you know either who are the right people to ask for if there's clinical trials going or not, but there's ways to get in contact with them… there's a lot of collaboration within the community and I shouldn't just say in the US because it is abroad as well.
Educator role	25% (3)	One of the other things I love about [being a clinical geneticist] is that there's learners from different backgrounds and fields.
Learner role	17% (2)	If you have a patient where you really haven't seen this before, there's always someone who is happy to have you run it by them and you can learn from their experience to help better care for the patients.
Changes in the field of clinical genetics		
Diagnostic improvement	92% (11)	We're able to diagnose so much more now that we have higher yield in the testing… that has limited the diagnostic Odyssey. For years…we'd do all kinds of blood work and MRIs and muscle biopsies… A lot of the things we find now we never would have anticipated… that has just revolutionized the field, and it's just made it more productive.
Increased need for lifelong learning	33% (4)	My role is really lifelong learning and trying to keep up to date. Every patient I see in clinic – literally there are hours of preparation in the sense of reading like what's all the current research, what's new because things change so rapidly. I guess I really like… the intellectual curiosity.
Expanding diversity of patients/conditions	25% (3)	With the increase in testing, we can help people with other conditions that we might not have seen as much before.
Increased utilization of genetics services	25% (3)	Thinking about utilization management around genetic testing, especially if a genetic specialist is not shepherding every single genetic test through the process. How do we put up guardrails to help people successfully order testing, order the right test at the right time?
Increasing negative financial pressure	17% (2)	Trying to make a financial case for our services, even a simple financial case for our services is very challenging.
Newborn screening improvement	8% (1)	Now, in the NICU when we suspect something, within days, we have… [a] diagnosis back. So, it's very reassuring that we can do diagnoses very fast, and it can change management."

Abbreviations: MRI, magnetic resonance imaging; NICU, neonatal intensive care unit.

fied as White (10/12), with the remaining respondents identifying as Asian (2/12). Most respondents held a primary board certification in pediatrics (10/12) and a secondary certification in clinical genetics and genomics (11/12). Although their primary certification was in pediatrics, these clinicians treat patients of all ages, including those with adult-onset genetic conditions. Additionally, one-third of the respondents had been practicing for 30 years or more, and 9 were current UWSMPH faculty members (Table 1).

Discover Themes: Responses to Questions About the Profession

Highlights of Being a Clinical Geneticist: Participants most frequently cited problem-solving (11/12) when asked to identify highlights of being a clinical geneticist. Continuity of care was

Table 3. Future of Medical Genetics: Themes Identified in Discussing the Future

Themes	% of Respondents (n)	Quote
Future benefits of clinical genetics		
Treatment improvement	75% (9)	I'm much more optimistic in my counseling of families than I was earlier in my career, because I honestly believe that many of these disorders, most of them are going to have treatments in the near future.
Diagnostic improvement	58% (7)	The more we learn about the human genome and the more tests we do on our patients the better we'll be able to make diagnoses.
Greater understanding of variants and polygenic disease	33% (4)	The collation and understanding of variants – which variants are pathogenic. I think we will get there, because ultimately there is a finite number of variants. But, maybe that's through the use of AI (artificial intelligence) where we start to determine which variants we need to worry about.
Prognostic improvement	33% (4)	The broader, more accessible genomic testing means in general people are getting diagnosed earlier and that gives us a longer runway to kind of understand their trajectories and to lean on research to optimize their care.
Improvement of genomic medicine delivery	25% (3)	Working with genetic counselors to figure out what is the most effective way to utilize clinical geneticists that are broadly trained in genetics versus in specific specialty areas. I don't think the old model where we saw every patient with genetic conditions is a viable model in the future and so we [need to] harness our expertise as well as share that expertise.
Public health and access improvement	17% (2)	We're on the cusp of sort of a new generation of clinical genomic testing that we'll have access to that insurance will pay for.
Newborn screening improvement	8% (1)	We've increased the number of disorders we can screen for so we can start our treatments really early, and I think we're going to see genomic newborn screening.
Future of the clinical geneticist role		
Increased delegation/ oversight role	75% (9)	There's a mismatch between demand and my ability to have a personal touch on every case… I think there might have to be an evolution where certainly we have to teach people to better understand the nuances of genetic testing and… interpretation. I wonder if there needs to be a sort of more supervisory role of geneticists or expansion of other people with expertise.
Increase access to genomic medicine training for other providers	67% (8)	I think we need to do more with collaborating with other boards… internal medicine or family practice. There should be opportunities to get certifications in genetics to show that these individuals have taken courses, passed knowledge tests, and are capable of doing more advanced genetic practice than others who haven't had that training. They can be sort of the ones in their specialties focused on genetic aspects that their colleagues can go to for help if there's not a geneticist immediately available.
Diagnostician to management	25% (3)	I'm hoping that we will be able to be not only diagnosticians but have an increased role in management and treatment than what we've previously had.
More involved in common disorders	25% (3)	I hope that we have testing for multifactorial common disorders. I hope that we are involved in that in some way and that can be kind of rolled out to the general medical field, so things like polygenic risk scores. Even better than that, to be able to predict how multiple small effects result in common disorders.
Increased advocacy for health equity	17% (2)	Especially thinking about health equality – we need to make sure that this type of care is available regardless of what your insurance is and regardless of where you live… we need to be very mindful of how we go about training people to make sure that everybody has the opportunity to take advantage of genomic medicine and have their care improved from [it].
More specialized	17% (2)	I think we're going to be more specialized, where a geneticist might focus in a particular area, somebody who is trained in cancer genetics or neurogenetics or some other discipline and they focus on patients in that with that population and/or general geneticists that really focus on the multisystemic conditions that are more complex and that are rare enough or unique enough.
Advice for current trainees in health care		
Embrace lifelong learning	75% (9)	[I hope] that everybody in every specialty would be interested in the genomic basis to the kinds of conditions they're seeing in their own clinical practices and that they would have opportunities for additional lifelong learning and training to gain that expertise so that they're comfortable and understand the utility [and validity] of genomic tests [and know] how to provide information.
Increase collaboration with genetics	50% (6)	People in genetics are happy to talk with other subspecialties about how it could be integrated into their field… so that the right patients are getting to us and the right type of genomic medicine is being used within their practice. Those are conversations we're going to need to have to make sure everybody can benefit from this expansion of testing and knowledge.
Know your own limitations	25% (3)	If you think you're able to provide genetics care, even limited in your specialty, you need to do all the aspects that that care provision requires, including the counseling, navigating the insurance parts, and all of that.
Understand the limitations of genomic	17% (2)	Sometimes we get consults for very nonspecific sorts of things because they don't know what's going on. Well, that's not a use for genomic technology in most cases. You need to have objective clinical findings that are suggestive of a testing Mendelian disorder if you want to use single gene or genomic testing to try to solve that problem.
Advocate for health equity	8% (1)	I want to make sure that if we have new technology, be it new gene therapies or new genetic tests, that those are widely available by the insurance we have, that we apply those opportunities equally, and everybody has the opportunity to benefit. I think working towards those equity issues is really important.

the second most common theme identified (8/12), followed by patient diversity (6/12).

Role of a Clinical Geneticist: When asked to "tell me about your role as a clinical geneticist on a multidisciplinary team," 11 respondents emphasized the diversity of their teams within genetics clinics, including medical assistants, nurses, advanced practitioners, and genetic counselors, while 10 respondents discussed the specialists and subspecialists whom they work alongside. Leadership within and across clinics was the third most commonly highlighted role (6/12).

Changes in the Field of Clinical Genetics: Nearly all respondents (11/12) commented on significant improvements in diagnostic testing, with the increased need for lifelong learning mentioned next most often (4/12). "Expanding diversity of patients and conditions" and '"increased utilization of genetic services" were each mentioned by 3 respondents (Table 2).

Dream Themes: Responses to Questions About the Future of Clinical Genetics

Future Benefits of Clinical Genetics: When asked about some of the most promising ways medical genetics may benefit patients in the coming years, respondents most frequently identified improved treatments for genomic disorders (9/12), followed by diagnostic improvements (7/12) and greater understanding of variants and polygenic diseases, as well as improved prognostication (4/12 each).

Future of the Clinical Geneticist Role: The majority of participants anticipated that the role of the clinical geneticist will shift toward greater oversight, with increased delegation of responsibilities to genetic counselors, advanced practitioners, or other physicians (9/12). Similarly, 8 respondents noted increased access to genomic medicine training materials for nongenetics physicians and other providers, and 3 predicted a shift from predominantly diagnostic roles to greater involvement in patient and condition management (3/12).

Advice for Current Health Care Trainees: In response to, "What are your hopes or advice for trainees in various health care roles as genomic medicine becomes more clinically applicable, accessible, and in demand?" participants most commonly encouraged embracing lifelong learning—particularly regarding genetic disorders and treatments (9/12—and recommended that nongenetics providers increase collaboration with clinical geneticists (6/12). Additionally, 3 respondents emphasized awareness of providers' own limitations when delivering genomic medicine (Table 3).

DISCUSSION

Discover Themes

A "Discover and Dream" approach was used in the interviews to identify highlights that could help guide future recruitment efforts, given workforce shortages. Overall, participants expressed strong enthusiasm for their careers. The majority reported enjoying the problem-solving aspects of their roles, citing the ability to spend substantial time with patients, answer questions, and achieve a deeper understanding of conditions.

Participants also valued continuity of care, noting they care for entire families, see patients grow up, and support them building families of their own. Half of the interviewees cited patient diversity as a major contributor to their job satisfaction, noting that patients span all ages, backgrounds, and abilities across both acute and chronic conditions.

Regarding their roles, participants cited examples of multidisciplinary teamwork within their clinics and across specialties, institutions, and even internationally. Some attributed this to the field's collaborative and close-knit nature, particularly in the context of diagnosing and managing rare disorders. Participants also valued their roles as leaders, educators, and lifelong learners, as many work with students and find opportunities to learn through teaching and collaborating with patients and colleagues.

When asked about changes in the field, participants identified rapid advances in diagnostic techniques and treatment options. One respondent stated, "Gene therapy, gene editing, and gene modification are technologies that are already in place today. There are FDA-approved products now—gene therapies, gene modifications - that are essentially curing our patients. That was inconceivable when I started."

Given this rapidly evolving landscape, respondents also emphasized the need for lifelong learning, including the effort required to interpret emerging research and stay current with developments in the field (Table 2).

Dream Themes

Overall, respondents expressed excitement regarding the future of clinical genetics. Most anticipated that the treatment of genetic disorders will evolve, with more gene therapy and precision medicine options on the horizon. Some also noted anticipated advances in prognostic disease monitoring, enhanced newborn screening, and improved access to genomic services.

Many participants mentioned the value of expanding diagnostic capabilities and improving understanding of variants and polygenic disease in the future to optimize patient care. It was through this lens that some discussed the role of the clinical geneticist expanding into care teams of more common, chronic diseases that have genetic components, such as hypertension and diabetes.

In response to these changes and increasing demands, most respondents anticipated that they will delegate more to advanced practice providers and genetic counselors to help address the shortage of clinical geneticists. Many also emphasized the need for access to comprehensive genomic medicine training for providers and trainees at all levels as precision medicine continues to grow in applicability and relevance.

Some participants noted challenges affecting the field's desirability, including limited positive exposure to clinical genetics during training, increasing training costs relative to lower compensation compared with other specialties, and complexity of insurance coverage and reimbursement for emerging genetic tests and technology. These factors, combined with rising patient demand, were viewed as further compounding the provider shortage.

Most participants recommended that current health care trainees embrace lifelong learning and maintain curiosity about the field. Specifically, they encouraged trainees to understand the genetic and genomic basis of conditions affecting their patient populations to provide the most accurate and effective diagnoses, management, and treatments. Some also conveyed the importance of recognizing the limitations of genomic testing, as well as one's own expertise.

Participants expressed hope that more providers reach out to genetics colleagues when questions arise regarding testing and treatment for patients with genetic conditions. Finally, one respondent emphasized the importance of advocating for health equity, especially as new diagnostic and therapeutic options—many of which are not yet covered by insurance and can be costly—become available. They highlighted the need to engage historically marginalized and underrepresented communities in policy decisions and to work together to best serve all populations, particularly in the field of genetics (Table 3).

Study Limitations

Qualitative research has inherent limitations, including subjectivity of data and potential researcher influence during interviews. The generalizability of this quality improvement study is limited by the small sample size and focus on a single institution. Additionally, the appreciative inquiry "Discover and Dream" approach may have biased findings toward positive aspects of the field. The study did not comprehensively address in-depth the challenges or threats to the profession.

Future Directions

To more completely investigate the perspectives of clinical geneticists in Wisconsin, future studies should include all practicing clinical geneticists in the state and expand to other professionals providing genetic services, such as advanced practice providers, genetic counselors, and dietitians.

Research examining care models that fully integrate genetic counselors and advanced practice providers into genetics specialty clinics may identify ways to expand access and improve equity, particularly in underserved rural and urban populations. Exploring training opportunities for advanced practice providers and subspecialists to obtain certifications in genomic medicine may also ensure there are enough clinicians adequately trained to provide genomic services within their respective fields.

Similarly, genetics service workflows should be analyzed and optimized to meet increasing patient demand. These areas warrant further investigation, including more comprehensive literature review and potential meta-analysis to better define current trends and inform recruitment and genomic medicine delivery efforts.

Finally, further exploration of health care trainees' perspectives may help identify opportunities to enhance education and address the plateau in interest in advanced genetics training. Future studies should examine factors that may discourage learners from pursuing careers in genetics—including compensation insurance-related challenges—while identifying strategies to attract and retain physicians in this field.

CONCLUSIONS

Clinical geneticists in this study expressed strong enthusiasm for their profession, noting problem-solving, continuity of care, and patient diversity as key highlights. Their roles afforded opportunities to lead and participate in interdisciplinary teams, collaborate with a variety of specialists, and engage in lifelong learning. Their experiences provided insight into the future of the field, highlighting diagnostic and treatment innovations that will require new care delivery approaches, including greater delegation to other providers.

Participants also recognized the importance of ensuring adequate training for all providers to better address the genetic and genomic contributions to health, morbidity, and mortality. Overall, their reflections illustrate both the current rewards of the profession and its future promise, which may help encourage health care trainees to consider clinical genetics when exploring careers.

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