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Contents

Utilisation of Genomic Services Among Physicians in Kenya

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UNIVERSITY OF NAIROBI

Background

With advancement of genetic technology and mainstreaming of genetics into all specialties of medicine, physicians will be required to play a pivotal role in the coordination and provision of primary genetic services.
Objective: To describe the delivery of basic genetic services among physicians and factors affecting integration of genomic medicine into their practice.

Materials and Methods

An online descriptive cross-sectional study was conducted among registered physicians practicing within Kenya and physicians in training at University of Nairobi. A self-administered online close-ended questionnaire that assessed the following domains: delivery of genetic services, attitude and perceptions towards genetics, barriers to delivery of genomic services, knowledge of genetics and physician demographics.

Results

The response rate was 41% with 190 of the eligible 464 physicians completing the survey. Eighty-seven percent of respondents had not received formal training in genetics, with 80% reporting involvement in evaluation of genetic patients. Physician involvement in genetic testing and pharmacogenomics was low at 31% and 29% respectively. Sixty-four percent of the respondents graded their graded their knowledge of genetics as moderate. Participants identified limited access to medical geneticist (80%), lack of referral guidelines (86%), high cost of genetic services (93%), inadequate knowledge of genetics as barriers to genetic service delivery.

Demographic characteristics of the participants

Variable	N = 190
Sex	
Female	109 (57%)
Male	81 (43%)
Years in clinical practice	
0-9	120(63%)
10-19	56 (29%)
20-29	8 (4.2%)
30-39	6 (3.2%)
Specialty	
Cardiology	9 (4.7%)
Dermatology	8 (4.2%)
Endocrinology	9 (4.7%)
Gastroenterology.	1 (0.5%)
Infectious disease	2 (1.1%)
Internal medicine (no sub-specialization)	79 (42%)
Nephrology	6 (3.2%)
Pulmonology	2 (1.1%)
Registrar in internal medicine	73 (38%)
Rheumatology	1 (0.5%)
Has training in genetics (apart from embryology)	25 (13%)
Has a special interest in genetics	60 (32%)

Clinical Practice of Genetics

Variable	N = 190
Proportion of patients you complete a family history	
0	2 (1.1%)
25%	33 (17%)
50%	45 (24%)
75%	69 (36%)
100%	41 (22%)
How often family history is updated	
At every visit	20 (11%)
Every 2-4 years	13 (6.8%)
Every 5-10 years	17 (8.9%)
Never	99 (52%)
Yearly or at periodic exam	41 (22%)
A two or three generation family history	104 (55%)
The family's ethnic background	116 (61%)

Physicians attitudes towards genomic medicine in categories

Genomic medicine is going to make important contributions in diagnosis and management of: -Prenatal conditions	
Moderate	13 (6.8%)
Negative	12 (6.3%)
Positive	165(87%)
-Pediatric conditions	
Moderate	18 (9.5%)
Negative	11 (5.8%)
Positive	161(85%)
-Adult-onset conditions.	
Moderate	20 (11%)
Negative	16 (8.4%)
Positive	154(81%)
Parents should be able to have their children (under 18) tested for susceptibility to adult-onset genetic diseases.	
Moderate	45 (24%)
Negative	22 (12%)
Positive	123(65%)
I can identify useful sources of information regarding genetics in my practice	
Moderate	62 (33%)
Negative	61 (32%)
Positive	67 (35%)
I can find information about the genetic tests available within our health system	
Moderate	46 (24%)
Negative	115(61%)
Positive	29 (15%)
I know how to contact my local genetics center.	
Moderate	32 (17%)
Negative	145(76%)
Positive	13 (6.8%)
Variable	N = 190
I find genetics and genomics an exciting part of my practice.	
Moderate	81(43%)
Negative	38(20%)
Positive	71(37%)
There is need to incorporate genomic medicine into my practice.	
Moderate	30(16%)
Negative	22(12%)
Positive	138(73%)
I need to keep up to date with advances in genomic medicine.	
Moderate	29 (15%)
Negative	17 (8.9%)
Positive	144(76%)
There are sufficient benefits to warrant testing for inherited adult-onset diseases	
Moderate	26 (14%)
Negative	17 (8.9%)
Positive	147(77%)
Advances in genomic medicine will improve my patients' outcome	
Moderate	26 (14%)
Negative	11 (5.8%)
Positive	153(81%)
It is important for me to learn about personalized patient care based on targeted or whole genome sequencing	
Moderate	33 (17%)
Negative	17 (8.9%)
Positive	140(74%)

Barriers to the provision of genetic services

Variable	N = 190[1]
Limited access to a medical geneticist for referrals for testing and/or consultation	
Usually	152 (80%)
Lack of referral guidelines	
Usually	163 (86%)
High cost of genetic services such as testing.	
Usually	177 (93%)
Inadequate knowledge of genetics, genetic testing and genetic counselling.	
Usually	117 (62%)
Lack of patient interest in genetic evaluation.	
Usually	78 (41%)
Limited access to genetic testing services	
Usually	155 (82%)
Lack of detailed or updated family history	
Usually	88 (46%)

Conclusion

Uptake of genetic service provision to patients by physician remains low due to inadequate genetics training, limited genetic specialist and prohibitive costs of genetic testing. Mitigation of these factors is required to improve genetic access to care.

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Training Teams to Decipher the Genomics of Developmental Disorders in Africa

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Background

- Rare developmental disorders (DD) affect 1 in 20 individuals with significant lifelong challenges[1].
- Genetics play a role in up to 50% of cases, with whole exome sequencing (WES) now the standard diagnostic approach employed [2].
- **Most African countries have limited access to genetic services, and few formalised genomic medicine training initiatives exist on the Continent.**

DDD-Africa Training Program

- The Deciphering Developmental Disorders in Africa (DDD-Africa) project aim to address these challenges.
- **South Africa** and the **Democratic Republic of Congo** lead the program, with support from KU Leuven, Belgium.
- Clinician and Scientist duos from **10 African countries** are participating.

1. Improving knowledge in Genetics and Genomics for Medical professionals (Online coursework)

2. Practical training of Clinicians and Scientists - Workshop hosted in South Africa

3. In-country Implementation - Recruitment, WES & Data interpretation.

Predicted Outcomes & Impact

References:

1. World Health Organization & United Nations Children's Fund, 2023; ISBN (WHO) 978-92-4-008023-2.
2. Manickam K, *et al*. Genet Med, 2021;23(11):2029-2037.

Funding:

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IMPLEMENTATION OF A GENE THERAPY EDUCATION INITIATIVE BY ASGCT AND MUHAS

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INTRODUCTION

The rapid advancement of gene therapy has led to an expanding list of approved clinical products, many of which are highly relevant to low- and middle-income countries (LMICs). However, access to these therapies is often hindered by high costs and limited local expertise. To address this gap, the American Society of Gene and Cell Therapy (ASGCT) partnered with the Muhimbili University of Health and Allied Sciences (MUHAS) to develop an educational initiative aimed at training faculty in gene therapy.

COURSE CONCEPTUALIZATION AND DEVELOPMENT

- The Introduction to Gene Therapy for Educators (IGTE) course was designed as an 8-session certificate program, delivered via Zoom over four weeks
- Aimed at university faculty in LMICs to integrate gene therapy into curricula.
- Developed collaboratively by ASGCT Global Outreach Committee and MUHAS.
- Featured expert-led lectures, interactive discussions, and supplemental reading materials.
- Targeted diseases included sickle cell anemia, hemophilia, HIV, and cancer.

COURSE IMPLEMENTATION

- Held in October 2022.
- Participants: 45 individuals from 9 African countries successfully completed the course *(figure 1)*

- **Participant Demographics:**
  Diverse mix of scientific and medical professionals (Bachelor's, Master's, PhD, and MD holders).

- **Engagement Metrics:**
  - Average module length: 89 minutes
  - Average attendance: 30.5 participants per live session
  - 100% speaker participation across North/South America, Europe, Africa, and India.

- **Evaluation & Feedback:**
  - 67% found the content entirely new.
  - 33% indicated it enhanced their existing knowledge.
  - Ethics, regulatory, and advocacy sessions were rated lower for relevance but highest in confidence to teach.
  - 97% appreciated the multi-presenter format.
  - 60% preferred a hybrid learning model.

LESSONS LEARNED

- Positive Reception: The course was well-received and demonstrated a significant need for gene therapy education in LMICs.
- Improvements for Future Courses:
  - Stronger emphasis on gene therapy basics.
  - Increased clarity on attendance and certification requirements.
  - Inclusion of quizzes to assess participant comprehension.
  - Expansion to other under-resourced regions.
  - Future plans for in-person training initiatives.

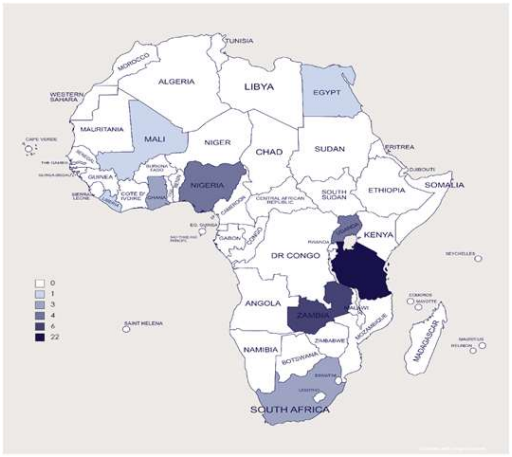

Figure 1: Participants by country

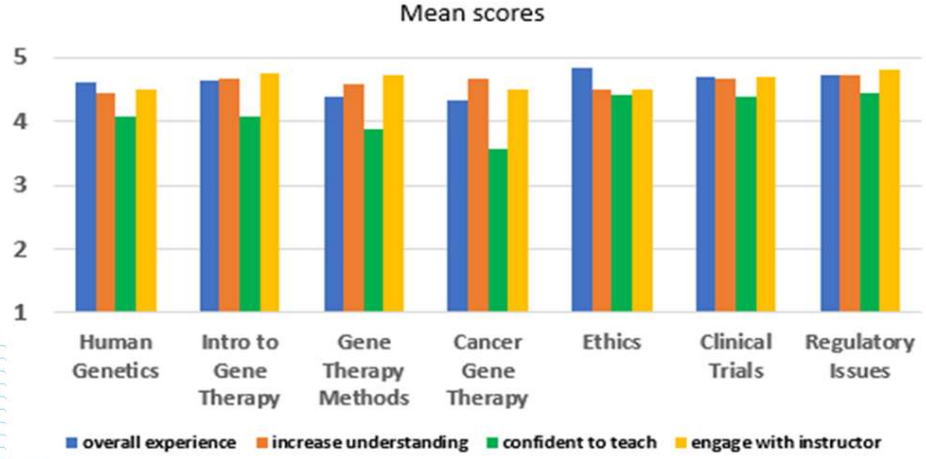

Figure 2: Course assessment and feedback ratings

ACKNOWLEDGEMENTS

Special thanks to ASGCT, MUHAS faculty, and all expert contributors who developed and delivered course content.

Scan for references and to read more

CariGenetics

Genomic Education, Training and Execution in the Caribbean through the Caribbean Genome Programme

Dr Carika Weldon, Professor Miten Jain and Dr Jacqueline Hall

Transforming Healthcare Through Genomics

The Caribbean Genome Programme (CGP), led by **CariGenetics,** in collaboration with local and regional authorities, is a landmark initiative designed to establish **local genomic** and predictive medicine **capabilities**. Traditionally, Caribbean DNA samples are sent abroad for testing, leading to:

- High costs
- Long wait times
- Brain-drain
- Limited research access to local genetic data

The CGP is building **regional sequencing and data analysis capacity** to provide **faster, cost-effective, and data-secure genomic services** for clinical and research use.

Breast Cancer Pilot	*Prostate Cancer CGP POC*	*CGP Pilot*	*CGP Full Launch*	*Global Expansion*
Bermuda | n=102	*10 Islands | n=1000*	*10 Islands | n=10,000*	*All 33 Islands | n=100,000*	*n=1,000,000*
WGS Sequenced Locally | Link Medical Records | Establish Biobank	Site Activations | Decentralised DNA Extraction	Scaling Infrastructure	Full Scale	Diaspora, LATAM, Africa
Q4 2023	Q3/Q4 2024	Q2 2025	Q1 2026	Q1 2028

The Value of Caribbean Genomic Data

The Caribbean is a **genetic goldmine**, shaped by centuries of diverse migration, and **indigenous, African, European, South Asian, and East Asian ancestry combined with** founder effects, island genetics.

- Underrepresented
- An ethnic melting pot
- Island population genomics

The Impact of CGP

- Local expertise in genomics & precision medicine
- Data sovereignty
- Development of regional scientific research
- Faster, localized clinical diagnostics for better patient outcomes
- A global leader in diverse genomics research
- Sustainable healthcare infrastructure

Building a Skilled Caribbean Genomics Workforce

The CGP is creating jobs and training experts across the Caribbean, reducing reliance on foreign labs by training:

- Clinical research
- Genetic counsellors
- Laboratory technicians
- Data analysts
- Operational staff

Each island undergoes a Precision Medicine Maturity Assessment to develop customized training programs, including:

- Identify key drivers needed to integrate genomics into local systems.
- Understand the skills, roles, and training pathways required for effective adoption.
- Develop rapid, scalable training programs **to equip healthcare professionals with genomic expertise.**
- Develop strategies to mitigate barriers to accelerate adoption.
- Facilitate cross-border access to genomic education tools.

Partners

Oxford Nanopore Technologies · CG Insurance · Bermuda Hospitals Board · Bermuda College · UWI · ECS · GENOMICS genomes.io · AminoChain · Future Barbados

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Continuous Education Program in Medical Genetics for Primary Healthcare Professionals in Brazil

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Introduction and aim

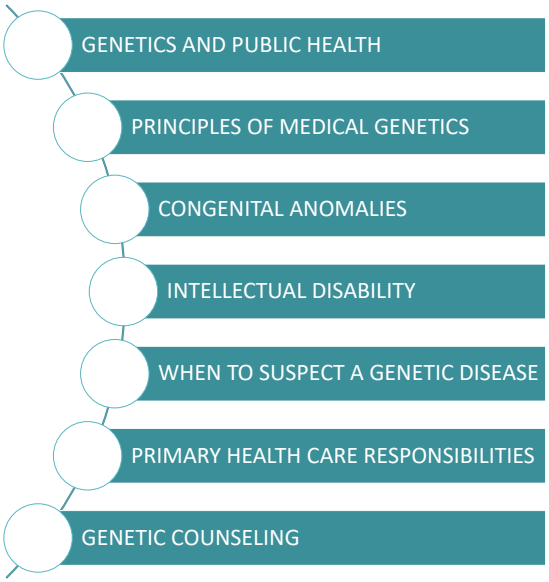
- In Brazil, primary healthcare (PHC) is the main entry point to the Unified Health System (Sistema Único de Saúde, SUS).
- To incorporate genetics into healthcare, the National Policy for Comprehensive Care for People with Rare Diseases within SUS assigns specific responsibilities to PHC, in alignment with global guidelines on the subject.
- This educational project aimed to integrate medical genetics into PHC in Brazil, in accordance with the National Policy for Comprehensive Care for People with Rare Diseases.

Methods

- The project focused on developing a genetics education guide for PHC professionals.
- Developed over nine months (November 2022–July 2023), it involved 16 expert medical geneticists affiliated with the Brazilian Society of Medical Genetics and Genomics (SBGM).
- The guide's content was defined through a comprehensive review of national and international competencies and evidence-based practices.

Results

- The resulting guide is structured into seven chapters, covering key topics in medical and clinical genetics:



- It provides practical instructions on identifying genetic conditions, utilizing diagnostic tools, and implementing patient management strategies within PHC.
- Special emphasis is placed on linking theoretical knowledge with actionable steps in clinical practice.

Conclusions

The guide seeks to address the critical shortage of genetic specialists in Brazil by equipping PHC professionals with foundational genetics knowledge and practical skills, thereby promoting early diagnosis and coordinated care.

It is freely available online and serves as a valuable resource for integrating genetics into PHC practice. It has been used in several training initiatives promoted by SBGM.

Scan here to read the guide

Catalyzing Clinical Genomic Services in British Columbia, Canada: A Collaborative Approach to Education and Implementation

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25YEARS GenomeBritishColumbia
Genomics positively impacts life, every day.

Abstract

Genomics holds significant potential to enhance health outcomes, provided that patients and healthcare providers can effectively leverage this technology. This multi-step project aims to catalyze the appropriate uptake and access to clinical genomic services across British Columbia (BC), Canada.

We employed a program logic model[1] as a best practice exemplar in genomics education and evaluation. We began with a situation and opportunity analysis of local resources and conducted interviews with a broad range of stakeholders, including researchers, industry representatives, provincial health authorities, genomics education specialists, and genomics service providers. After synthesizing the recurrent themes and opportunities, we developed a framework with key tactics for engaging our partners in genomic education strategies. Tailored to the BC healthcare ecosystem, this framework guided three exemplar projects aimed at educating and enabling non-genetic specialists to improve the uptake and access of clinical genomic services in BC.

We detail our successes, challenges, and emerging outcomes for these exemplar projects

1. Building capacity for genetic services guidelines using Pathways, a digital tool that supports access to referral processes, and point-of-care tools.
2. Supporting cardiologists and primary care providers with resources to disseminate genetic results for familial hypercholesterolemia (FH).
3. Providing patients and clinicians with handouts to support informed decision-making for private-pay pharmacogenomic tests.

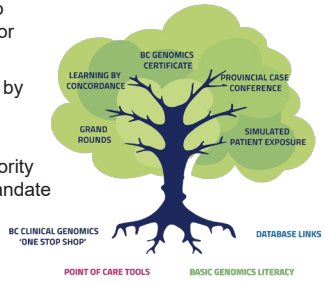
Educational interventions for healthcare professionals are most successful when tailored to the local context and designed through a collaborative approach, leveraging existing relationships, projects, and tools.

Education Framework: Vision & Principles

From our consultations we developed the vision for the genomics education implementation framework.[2] Represented by a tree, the vision is rooted by an essential foundation of knowledge. This includes basic genomics literacy and point of care tools supported by clinical guidelines. This would lead the learner to seek out and sustain their genomic competencies in a variety of ways and provide new clinical privileges.

These framework principles are key to directing our efforts to build capacity for genomic education in BC including:

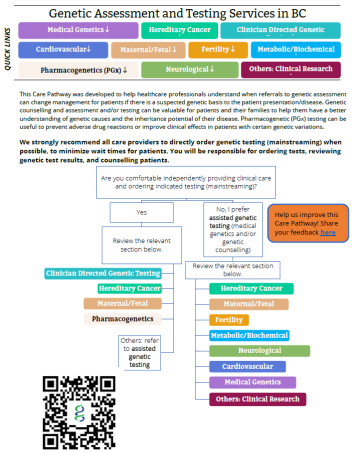
- Leverage existing opportunities led by others
- Identify innovative approaches
- Co-create new initiatives that fill priority gaps with education and clinical mandate groups
- Explore new funding models for sustainability
- Encourage the development BC clinical guidelines and Canadian genetic competencies for health professionals



Improving access to genetic services in partnership with local clinical support tool

Pathways
www.pathwaysbc.ca

Pathways is an online clinical support tool used by >95% of health professionals in BC. This project built out the Genetics specialty sections available on Pathways with the goal to enhance awareness about the existing genetic and genomics services and resources.



Successes:

- Supported updates to 13 clinic profiles
- Added 18 new resources for clinicians and patients
- 250% increase in views for the genetics pathway over 18 months

Filling the gap between pharmacogenetic research and practice

In Canada, while research continues to explore the implementation of pharmacogenetic (PGx) testing, patients and clinicians need help now to support responsible use of direct-to-consumer testing PGx testing.

Genome BC in partnership with PGx researchers & genetic counsellors has released info sheets for patients and clinicians to help answer the question "Should I get PGx testing?" including benefits and drawbacks, cost, and common medications. There is also a list of companies that provide testing. Evaluation is ongoing to see if they fit the need.

Coming soon: A video simulating a conversation between a primary care provider and a patient about how to use their PGx results to support depression medication prescribing.

Hit and a Miss on in-time FH Education

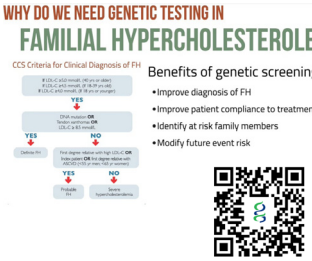
Genome BC took the opportunity to provide in-time education for cardiologists and family doctors that were receiving FH genetic test results as part of a research study after their patient had an early onset heart attack. The material was distributed as a QR code in the report and as a clinician tool on the FH Canada website.

Success:

- Co-created 6min video and online resource portal that met the identified needs
- Well received when viewed via partner website & at rounds

Challenges:

- No uptake by physicians from the report sent by fax
- Limited options for distribution outside of the study



Footnotes

1. Nisselle et al. Front. Genet., 07 Nov 2019 https://doi.org/10.3389/fgene.2019.01057
2. Genome BC. 2021 https://www.genomebc.ca/wp-content/uploads/2021/12/Genome-BC-HCP-Education Phase-II-Implementation-Framework_FINAL.pdf

A preliminary Canadian Genomics Education Asset Map reveals many activities confined to individual jurisdictions

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5 Department of Genetics, Children's Hospital of Eastern Ontario, Ottawa, ON, Canada and Children's Hospital of Eastern Ontario Research Institute, University of Ottawa, Ottawa, ON, Canada

Background

Within Canada, healthcare is publicly funded and delivered through provincial health systems that serve the 10 provinces and 3 territories, in 2 official languages (English and French). Healthcare providers (HCPs) are trained at academic medical centres throughout the country (17 medical schools; 7 medical schools that train medical geneticists; 10 clinical lab geneticist training sites; 4 genetic counselling training programs), resulting in significant variability in local context and specific training opportunities. Last revised in 2017 for genetic content, updated genetics and genomics objectives will be added to the Medical Council of Canada (MCC) Examination Objectives for undergraduate medical education in 2026.

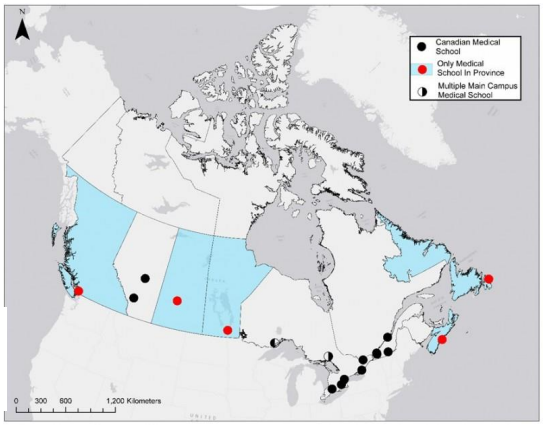

Figure 1. Map of medical schools across Canadian provinces. From Rourke J *et al,* Rural and Remote Health 2018; 18: 4426. https://doi.org/10.22605/RRH4426

Methods

- Workshop conducted by the Canadian College of Medical Geneticists (CCMG) with its members in 2024 to identify educational needs of several end users: medical students, residents, non-genetic physicians, and allied health professionals
- Horizon scan of Canadian genomics education resources to develop a preliminary asset map

Results

- Training for HCPs in Canada is fragmented and variable, with educational activities sequestered within academic centres and hospitals. Many centres have developed mainstreaming approaches, mainly for cancer genetics, developing documents and videos to facilitate provider upskilling and patient education and consenting.
- Some organizations including Genome BC and Genetics Education Canada: Knowledge Organization (GECKO) have developed and made widely available genomics education content (see Figures 2-4 for examples). A minority of available content is Canadian-made, with many sites linking out to international resources.

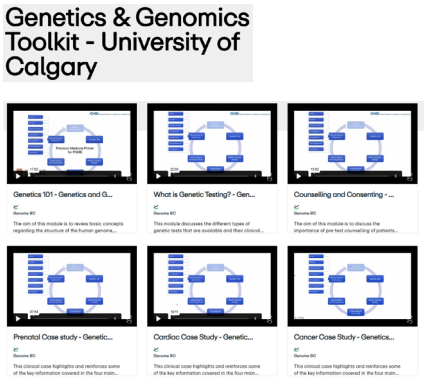

Figure 2. Genetics & Genomics Toolkit, a series of videos developed by medical genetics residents at the University of Calgary for non-genetics residents and practicing clinicians. Videos cover basic competencies such as Genetics 101, Counselling & Consenting, and Interpreting and Delivering Genetic results.

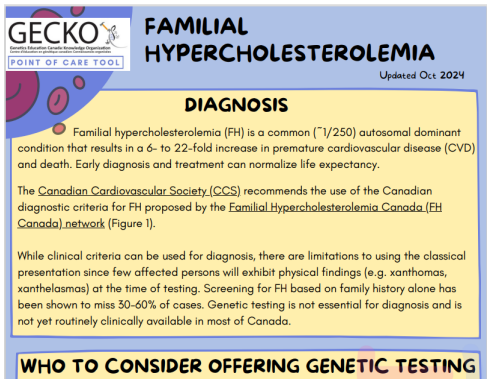

Figure 3. GECKO point of care tool for familial hypercholesterolemia. GECKO develops educational content in the form of point of care tools, "GECKO on the run" (concise summaries) and "GECKO deep dive" (comprehensive reviews). Content is grouped by specialty (e.g. preconception and prenatal genomics, cardiogenomics, technologies) and is reviewed and updated by content experts every 2 years.

Figure 4. UBC graduate certificate in Genomic Counselling and Variant Interpretation, a part-time, 12-credit, post-graduate certificate designed to upskill genetic counsellors in bioinformatics and genomic counselling. The certificate is composed of 4 courses designed for GCs and genetic nurses (Genomic Testing & Clinical Bioinformatics; Applied Variant Interpretation; Evidence-based Genomic Counselling; Emerging Genomic Topics).

Conclusions

HCPs have diverse learning priorities and genomic knowledge and no single education strategy will improve care for patients with rare genetic disorders. The lack of a comprehensive centralized genomics education resource has resulted in fragmented educational resources. A comprehensive asset map of all activities is required and a national strategy to make Canadian and global resources available is urgently needed.

Needs Assessment for Curricular Innovation in Medical Genetics Residency in Colombia

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Introduction

The medical genetics (MG) residency program at PUJ in Colombian is pioneer in the country. However, the program faces the need for curricular innovation to address emerging challenges in medical genetics as well as in health and educational systems. This is the result from needs assessment for this purpose.

Methods

A study was conducted involving surveys, interviews, focus groups and documental review. All the answers were categorized into: experience, pertinence, and MG competences core (2) .

Results

1. Participation

Survey:
19/34 graduates (56%)

Focus groups/interviews:
13 faculty members.
4 medical residents.
4 graduates.

Documental review:
Syllabus, pedagogic strategies, assesment methods.
Global recommendations.

2. Perception of compliance with core competencies (ACMG)

>= 4,2/5

- Medical histories, physical exam. (4,8)
- Ethical issues (4,7)
- Effective communication (4,4)
- Management plan(4,2)

3,9-4,1/5

- Order and interpret genetic tests (4,0)
- Integrate sources of information to quantify risk (3,9)

<3,9/5

- Core public health functions (3,6)
- Clinical or translational research (3,3)
- Counseling whole genome /exome seq. (3,2)

These are the results of the Likert scale ratings that graduates give to their perception of compliance with general competencies and milestones in medical genetics defined by the ACMG and ACGME (1,2)

3. Experience and pertinence

- **Skills:** integral clinical approach, quality of care
- **Teachers:** professionalism, ethical integrity, interdisciplinarity.
- **Processes**: Variety of patients, infrastructure, technology, library access.

- **Skills:** Clinical bioinformatics, genome-exome, oncogenetics, adult diseases.
- **Teachers:** teaching skills and Learning environment.
- **Processes:** Assesment and feedback.

These are the results of the subcategories (skills, teachers, processes) analyzed in focus groups and interviews with graduates, professors, and active residents where there were agreements in the responses

Conclusions

- Overall, a very good balance in the qualitative and quantitative perceptions of the medical genetics program curriculum. The areas of professionalism, patient care, ethical and technical integrity were the best evaluated.
- Among the key improvement opportunities identified was to integrate Clinical Bioinformatics into the curriculum, Expand the public health axis to genomics and complex conditions and enhance translational research.
- The evaluation did not reveal significant discrepancies among the perceptions of the different stakeholders involved.

References
1. Competencies for the Physician Medical Geneticist in the 21st Century. Report of a Working Group of the American College of Medical genetics (ACMG), 2013
2. Medical Genetics and Genomics Milestones. The Accreditation Council for Graduate Medical Education. 2nd revision, 2019

COMMUNITY GENETICS IN CUBA:
HUMAN RESOURCE CAPACITY BUILDING AS A KEYSTONE

BEATRIZ MARCHECO-TERUEL National Medical Genetics Centre, Havana, Cuba

INTRODUCTION

Cuba initiated a national programme for the diagnosis, management and prevention of genetic disorders and birth defects in the early 1980s. The programme is an integral part of the country's universal health services and operates through the National Medical Genetics Network focused on community-based care and linked to secondary and tertiary care. Each Cuban municipality has at least one professional genetic counsellor. Training personnel has been a top priority for setting up genetics services

The training of professionals in the field of medical genetics has been the cornerstone for establishing services across all three levels of the national health system and expanding them to ensure comprehensive coverage for the entire population.

TIMELINE FOR THE DEVELOPMENT AND IMPLEMENTATION OF MEDICAL GENETICS SERVICES IN CUBA

1977 — Clinical Genetics was officially recognized as a medical specialty

1981 — The national program was launched by the Ministry of Public Health

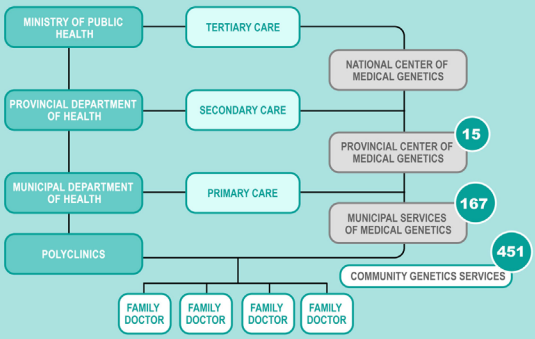
1984-1999 — A national network of Medical Genetics Centers and Services was gradually implemented in two periods:

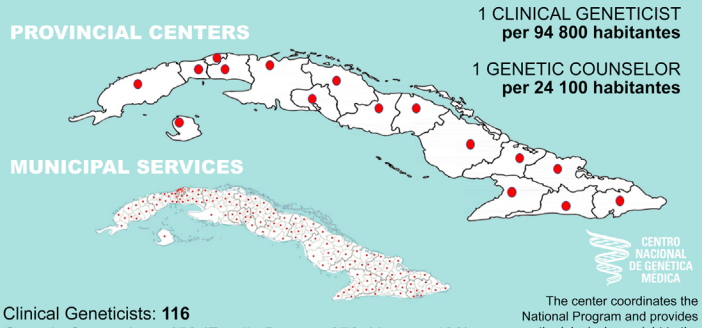
- Provincial centers

2000-2006

- Municipal and community genetics services were integrated into all three levels of the National Health System

MEDICAL GENETICS SERVICES WITHIN THE CUBAN HEALTH SYSTEM AND THEIR NATIONWIDE DISTRIBUTION





1 CLINICAL GENETICIST **per 94 800 habitantes**

1 GENETIC COUNSELOR **per 24 100 habitantes**

Clinical Geneticists: **116**
Genetic Counselors: **456** (Family Doctors 276, Nurses: 180)
Specialists in 38 labs: **152**

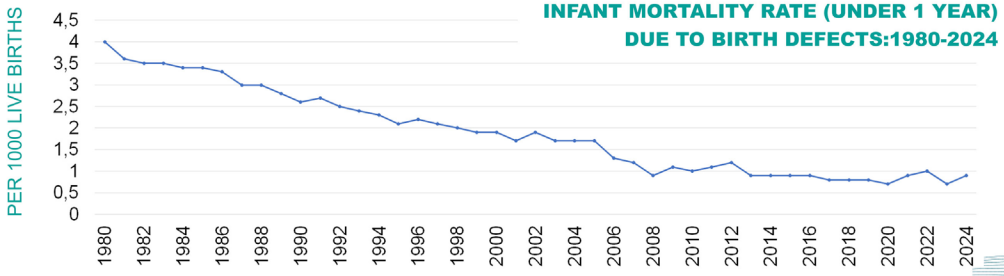
CENTRO NACIONAL DE GENETICA MEDICA

The center coordinates the National Program and provides methodological oversight to the national network of Medical Genetics centers and services

TRAINING HEALTH PROFESSIONALS WITH KNOWLEDGE/EXPERTISE IN MEDICAL GENETICS

- In 2004, a Medical Genetics course was introduced into the medical school curriculum, spanning 54 hours during the fourth semester
- Clinical Genetics specialists are trained in six of the country's 17 medical universities, following a standardized curriculum coordinated by the National Center of Medical Genetics. 239 clinical geneticists have graduated.
- Most medical specialties have included Medical Genetics courses into their Residence curriculum.
- Additionally, there are two Master's degree programs –Medical Genetics and Genetic Counseling– each lasting two years. Since 1998, 1013 genetic counselors have been trained in 17 programme cohorts , with 60% being family doctors and 40% higher-level nurses

IMPACT ON HEALTH INDICATORS



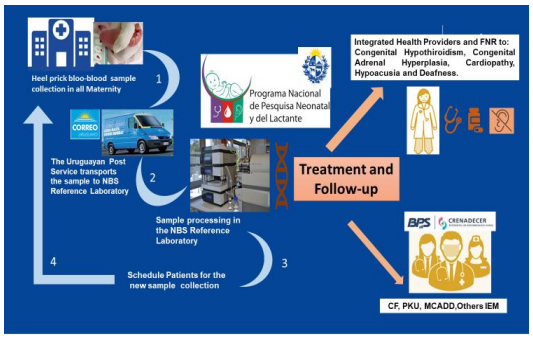
Neonatal Screening in Uruguay: Innovation and Commitment to Public Health

Mariela Larrandaburu,[1] Montevideo, Uruguay

Neonatal screening is a fundamental public health strategy to prevent disability and congenital conditions by enabling the early detection of diseases through simple tests.

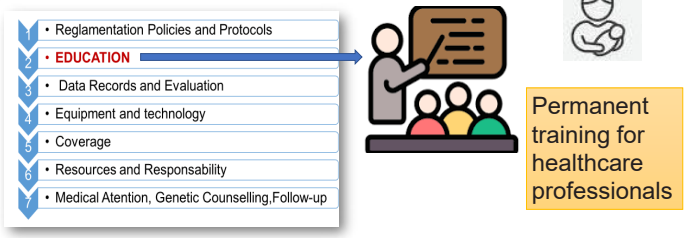
This intervention significantly improves the prognosis of affected children, offering them the opportunity to lead full and productive lives, benefiting both themselves and society. Uruguay's Neonatal and Infant Screening **Programme began in 1994 with the detection of congenital hypothyroidism**. Since 2013, it has evolved to include three components: dried blood spot testing, otoacoustic emissions, and physical examination. In 2018, congenital heart disease screening was added using pulse oximetry. This **programme is mandatory, universal, free of charge, and has achieved high nationwide coverage.** The Ministry of Public Health (www.msp.gub.uy) has prioritised the dissemination, awareness, and training on neonatal screening. This work highlights two key initiatives:

1. **The creation of the NBS Stamp in 2024**, issued by the national postal service, a key institution in transporting heel prick blood samples from all maternity hospitals across the country to the sole National Neonatal Screening Reference Laboratory. This stamp positions Uruguay in the international philatelic community, showcasing its leadership and innovation in Latin America. It is essential to continue developing educational resources and awareness campaigns to empower the public and maintain an ongoing training agenda for healthcare professionals.

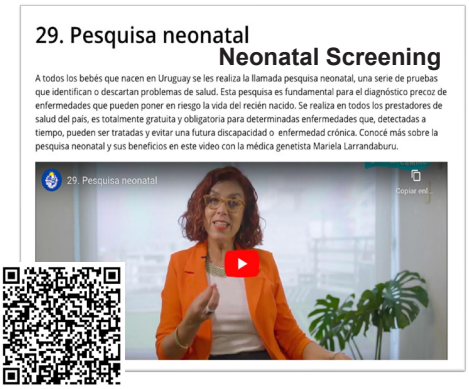


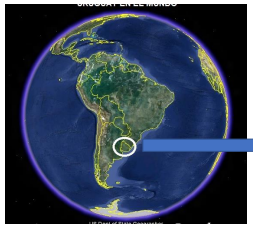
Components of a Neonatal Screening Programme

NBS GENOMIC TESTING IN URUGUAY
PKU, CF, Propionic Acidemia, MCADD Hemoglobinopathies, Congenital Adrenal Hyperplasia.



2. An **informational video** aimed at the general population, clearly and accessibly explaining the purpose and importance of this preventive public health measure.





Annual Case Estimate [3]
•123 Congenital Cardiopathy
•30 Congenital Deafness
•12 HTC
•6 CF
•4 CAH
•4 PKU
•2 Hemoglobinopathie
•1 MCADD
•2 EIM other

Population:
3.500.000 hab.

Annual Birth:
32.000 newborn

Scan to hear more

Footnotes

1 Rare Disease and Congenital Anomaly Programme, Ministry of Public Health
2 https://www.gub.uy/ministerio-salud-publica/bebe-en-camino
3 Larrandaburu et al, 2019. https://doi.org/10.1590/2326-4594-JIEMS-2019-0002

Establishing La Sociedad Profesional Latinoamericana de Asesoramiento Genético (SPLAGen) to advance the field of genetic counseling in Latin America

Daniela Diaz Caro,[1] Laurie Simone,[2] La Sociedad Profesional Latinoamericana de Asesoramiento Genético board of directors
GH Sergievsky Center Columbia University Medical Center,[1] Center for Genetic and Genomic Medicine, Hackensack University Medical Center[2]

Background

- Genetic counseling integrates risk assessment, patient education, and psychosocial support to help patients and families understand genetic conditions, make informed healthcare decisions, and navigate the emotional and medical implications of genetic information.[1]
- Genetic counseling has developed as an independent profession around the world. Outside of Cuba, genetic counseling is not a recognized profession in Latin America (LATAM).[2]
- In LATAM, genetic counseling is provided predominantly by medical geneticists, nurses, or other healthcare professionals with varying degrees of training in genetics and counseling.[2-3]
- The growing demand for genetic services, driven by advancements in genomic medicine and the rise of personalized healthcare, is increasing the need for professionals trained in genetics and genetic counselling. However, unlike Brazil and Cuba, most LATAM countries lack the necessary training programs and institutional support to develop and sustain a genetic counseling workforce.[2-4]
- **SPLAGen was established to advance the genetic counseling profession and practice in LATAM.**

Institutional Goals and Structure

Mission	To promote awareness of and access to genetic counseling services in Latin America through the promotion of education, professional support, research and public policy.
Vision	Empower members to integrate genetic counseling into all aspects of healthcare, education and research in Latin America.

- SPLAGen's leadership structure includes executive roles (e.g., president, secretary, and treasurer), regional representatives, and committee chairs.
- SPLAGen has **four membership categories***
  - Full members who are genetic counselors
  - Full members from other professions practicing genetic counseling (e.g., medical geneticists)
  - Allied professionals interested in our mission
  - Students interested in our mission

* All members receive educational resources, networking, and professional development, but only full members can hold leadership roles, including board and committee chair positions.

Current Member Led Initiatives

Educational Committee is focused on advancing our educational content to expand knowledge of genetic counseling practices.
- Quarterly webinars focus on genetic counseling topics, including newborn screening, cancer genetics, and student research in LATAM.
- The Curriculum Development Working Group focused on the initial steps of developing a standardized framework for genetic counseling education in Latin America.

Research Committee is focused on advancing our research goals to promote the profession, recognition, and utility of genetic counseling practices.
- Quarterly meetings focused on discussing current research initiatives, resource sharing, and networking.

Marketing and Membership Committee works to expand SPLAGen's reach, engage the community, and promote genetic counseling in Latin America.
- Internship program for LATAM undergraduate students.
- Manage social media outreach and newsletter to inform members and the public of genetics-related content.

Public Policy and Access Committee is focused on advocating and understanding how to promote professional recognition of genetic counselors and access to genetic counseling services.

Our History

2020
- Discussions within Minority Genetics Professional Network and between Laurie Simone, MS, CGC and Sonia Margarit, MS, CGC about the need for a unified organization.
- Needs assessment conducted to evaluate the state of genetic counseling in Latin America.

2021
- SPLAGen directory established to track genetic counseling professionals.
- Ongoing discussions on who is providing genetic counseling and how to support them.

2022
- Bylaws drafted to formalize the structure of the organization.
- First board established, marking the official leadership formation of SPLAGen.

2023
- Launch of educational webinars, expanding SPLAGen's role in professional development.
- Obtained US IRS non-profit status (501c3), solidifying SPLAGen as a recognized entity.

2024
- New round of board members are onboarded, ensuring continued leadership and growth.
- Continuing to further develop our organizational initiatives.

Conclusion

SPLAGen plays a vital role in the expanding access and knowledge of genetic counseling in Latin America. Through education, advocacy, public policy, research and collaboration, the society continues to build a strong foundation for the integration of genetic counseling into the healthcare landscape in Latin America.

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4. Diaz Caro D., & Simone L. The role of the Latin American Professional Society of Genetic Counseling (SPLAGen): Advancing genetic counseling in Latin America. *Genet Med Open*. 2024;2(Suppl 2):101870. Published 2024 Jul 16. doi:10.1016/j.gimo.2024.101870

Scan QR Code to watch a short presentation

The Genetics & Genomics Nursing Practice Survey: The Value of Open Access Instruments

Kathleen Calzone, PhD, RN, AGN-BC, FAAN[1], Emma Tonkin, PhD[2], Andrew Dwyer, PhD, FNP-BC, FNAP, FAAN[3], Sarah Dewell, PhD, MSc., RN[4], Laurie Badzek, LLM, JD, MS, RN, FNAP, FAAN[5]

[1] Genetics Branch, Center for Cancer Research, National Cancer Institute, National Institutes of Health, United States. [2] Genomics Policy Unit, Faculty of Life Sciences and Education, University of South Wales, United Kingdom. [3] Connell School of Nursing, Boston College, United States. [4] School of Nursing, Thompson Rivers University, Canada. [5] Ross & Carole Nese College of Nursing, Penn State University, United States

GGNPS OVERVIEW

- Variable response instrument aligned to Rogers Diffusion of Innovation domains.
- Assesses knowledge, skills, attitudes, confidence, the social environment (i.e. management/supervision) and utilization of genetic/genomics in nursing practice.
- Multiple question formats (select all that apply pick lists, multiple choice, yes/no, true/false, and Likert scales) administered online or on paper.
- Refined and validated using the following methods:
  - ✓ test/retest reliability
  - ✓ structural equation modeling
  - ✓ construct, content, & face validity
  - ✓ confirmatory factor analysis[1, 2, 3]
- Permission is granted on an individual basis.
- Release includes the instrument, instrument instructions, validation papers, and directions that the instrument can be modified for local context but that could impact instrument performance, validity, and reliability.

Building the Evidence

Developing instruments from scratch is challenging and hinders timely assessment.

With the formation of the Global Genomics Nursing Alliance (G2NA), the GGNPS was made open access to support countries (and regions of large countries) in assessing genomic capacity of their nursing workforce.

The GGNPS has been modified, validated, used, and resulted in publications from countries in North and South America, Europe, Northern Europe, the Middle East, Asia, and Africa.

★ Has used, is using, or has requested the GGNPS instrument and/or GGNPS item(s)

Findings

Published GGNPS studies document:

- Low faculty genomic knowledge and teaching capacity.
- Low practicing nurse genomic knowledge, skills, and abilities.

Large countries such as China have chosen to use the Chinese GGNPS in multiple regions/cities facilitating the development of common sharable interventions.

Analysis of GGNPS data from 5 countries is being presented at the 2025 meeting of the International Council of Nurses.[4]

Opportunities

There are ~28 million nurses practicing globally. Most nurses have limited genomic capacity with roles that span from bedside care to independent practice with prescriptive authority.

Identifying common, shared deficits can inform global solutions. Building workforce capacity demands interprofessional and international collaboration.

G2NA's global analysis will inform nursing genomic education globally. In parallel, G2NA's ongoing work will help determine what interventions have been effective in increasing capacity and integrating genomics into nursing practice.

References

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4. Yesilcinar, I., Laaksonen, M., Calzone, K., Halkoaho, A., Honghong, W., Limoges, J. (2025). Insights from five countries using the GGNPS survey to drive leadership strategies. International Council of Nursing. Abstract, accepted.

Cutting-edge genomics in the classroom

Kelly M. East, MS, CGC, HudsonAlpha Institute for Biotechnology, Huntsville, AL, USA

Classroom resources and educator experiences make genomics concepts and skills broadly accessible to students early and often in their educational pathway. Investment in these activities inspires, teaches, and trains the next generation to enter the STEM workforce and builds a genomically informed population.

HudsonAlpha Institute for Biotechnology

The HudsonAlpha Institute for Biotechnology is a non-profit genomics research institute dedicated to human health and agricultural challenges. HudsonAlpha is also deeply committed to an educational mission.

HudsonAlpha Education provides genomics-focused student programs, educator training, workforce development, and public outreach, reaching >1.5M learners annually.

Classroom Kits and Resources

- More than two dozen hands-on classroom genomics and biotech activities
- Real-world storylines and aligned with K-12 curriculum standards
- Reach >100,000 students annually

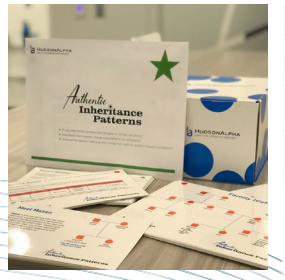
Examples

Gaudy Goldfish *(inheritance patterns)*

- ask questions and define problems
- analyze and interpret data
- construct explanations
- design solutions
- communicate information
- recognize patterns

How do Polygenic Risk Scores Stack Up?

- analyze and interpret data
- explain variation of expressed traits in a population
- construct distribution model
- explain what polygenic risk score is and how it is used
- discuss the impact of sample diversity on clinical utility

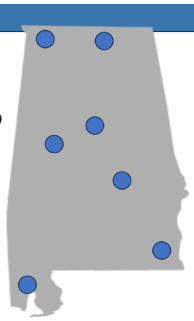


Educator Professional Learning

- Workshops for grade 7-12 life science teachers
- Held across the state of Alabama
- Brand-new content developed and deployed every two years
- No cost to teachers through State funding
- Teachers take home classroom materials
- Reach more than 400 teachers each year

Round	Content themes
2012-13	Karyotyping, Multifactorial Inheritance, Central Dogma
2013-14	Cell Division, Chromosome Segregation, Cancer
2015	Resources aligned to the then new Alabama Science Course of Study
2016-18	Intersection of Agriscience and Biology
2021-23	DNA Phenotyping, Authentic Inheritance Patterns
2023-25	Karyotyping revisited, Polygenic Risk Scores

Round 6 workshop locations across Alabama



Co-development of learning materials and experiences with scientists and educators can bring real-world science into classrooms at scale.

Footnotes

More information about HudsonAlpha classroom kits: https://knowledge.carolina.com/our-brands/hudsonalpha/
HudsonAlpha Educator Resource Hub: https://www.hudsonalpha.org/educatorhub/

The TRIG Model: Team-Based, Vetted Genomics Education Across Specialties

Richard L. Haspel, MD, PhD, FRCPath(Hon)
Professor of Pathology
Beth Israel Deaconess Medical Center
Harvard Medical School
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rhaspel@bidmc.harvard.edu
https://www.pathologylearning.org/trig

Check out the website!

Download free resources!

Oncology-focused

TRIG Training Residents In Genomics **Pathology**

TFIG Training Fellows In Genomics

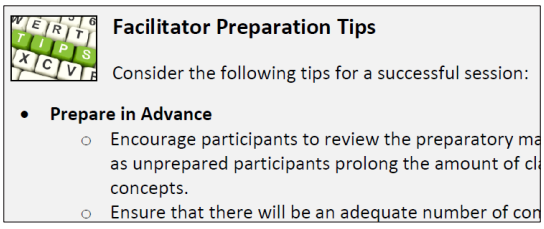
UTRIG Undergraduate Training in Genomics **Medical Students**

Practical Exercises Using Online Tools

NCBI Resources How To · ClinVar · PolyPhen-2 · COSMIC Catalogue of somatic mutations in cancer · gnomAD · ClinGen Clinical Genome Resource · OncoKB · cBioPortal for Cancer Genomics

Free Instructor Handbooks and Toolkits

- **Comprehensive guidance for local site implementation**
  - Workshop Instructor Handbooks
  - Toolkits: Slide sets; Activity handouts; Google forms
- **Used by ~37% US Pathology residency programs**
- **Resource downloaded: >4000 registrants > 90 countries**
  - Used w/>3,700 learners
  - >90% of respondents would recommend
  - Many used for own learning



Facilitator Preparation Tips

Consider the following tips for a successful session:

- **Prepare in Advance**
  - Encourage participants to review the preparatory ma
    as unprepared participants prolong the amount of cl
    concepts.
  - Ensure that there will be an adequate number of con

12 Publications: Some examples

Development of a validated assessment tool
– *Arch Pathol Lab Med. 2021;145:453-456*

Using national in-service exams
– *Genet Med. 2021;23:1151-1157*

Educational innovation: TBL at annual meetings
– *J Grad Med Educ.* 2016; 8:80–84

Non-oncology

Universal Genomics Instructor Handbook and Toolkit

Plug-and-Play (e.g. Cardiology, Neurology, Ophthalmology)

- Collaborating organizations/Expert developed
- >50 workshops >2,000 learners
- Team-Based Learning (TBL) = Interactive

Workshop Evaluations: Highly Rated

USCAP 2024 (n=49): 98% would recommend to other medical students

Participant-provided adjectives (large font = more frequently used)

THANKS!

- **Association for Academic Pathology (AAPath)**
  - PRODS Council/UMEDS Council
- **TRIG/UTRIG-Supporting Organizations**
- **Working Group Members**
  - TRIG/UTRIG/TFIG
  - ISCC-PEG Innovative Approaches (genome.gov/iscc)
- **American Society for Clinical Pathology (ASCP)/American Society of Clinical Oncology (ASCO)**
- **Supported by grant from the NIH (R25CA168544)**

GENOMICS EDUCATION TRIG model

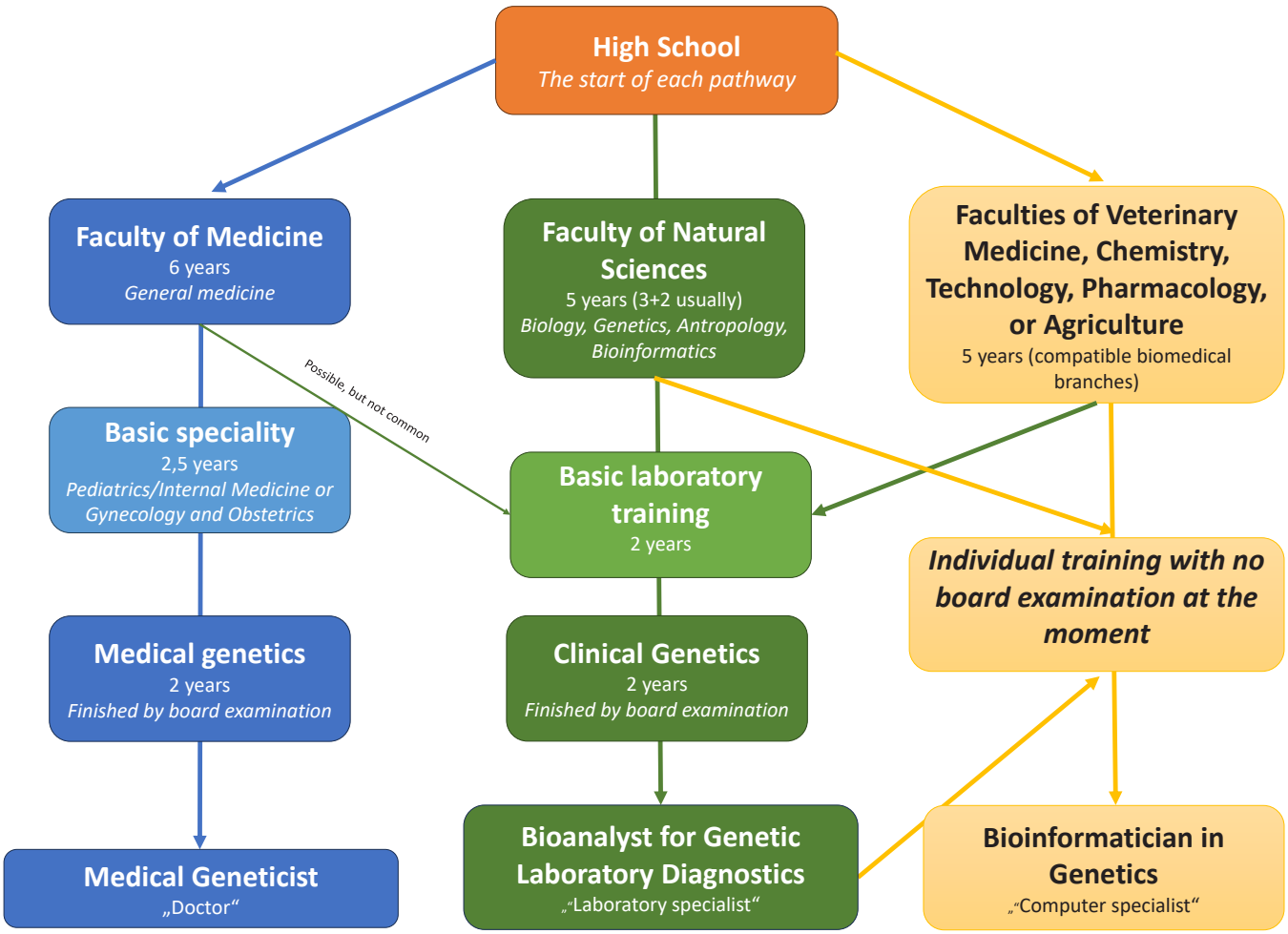
The Genetic Landscape of the Czech Republic: Career Pathways in Medical Genetics and Laboratory Diagnostics

Dr. Antonin Sipek[1], prof. Sarka Pospisilova[2,3], prof. Milan Macek[4]

1) *Institute of Biology and Medical Genetics, 1st Faculty of Medicine, Charles University and General University Hospital, Prague, Czech Republic*
2) *Center of Molecular Medicine - Central European Institute of Technology, Masaryk University, Brno, Czech Republic*
3) *Institute of Medical Genetics and Genomics, Faculty of Medicine, Masaryk University and University Hospital Brno, Brno, Czech Republic*
4) *Institute of Biology and Medical Genetics, 2nd Faculty of Medicine, Charles University and Motol University Hospital, Prague, Czech Republic*

Introduction:

Currently, a variety of professions contribute to the provision of genetic and medical services in the Czech Republic. This includes physicians certified in the field of medical genetics, laboratory diagnostics experts who interpret the results of genetic tests, genetic laboratory technicians and nurses, as well as bioinformatics specialists. Integral members of these multidisciplinary teams also include research and academic professionals.



Email: antonin.sipek@lf1.cuni.cz

The Estonian Biobank's MyGenome Portal
A platform for participant engagement

Leitsalu L., Pervjakova N., Metsalu K., Kruusmaa K., Pärkson P., Teder K., Kaasla K., Sissas LJ., Fischer K., Kukushkina V., Kleemann P., Reigo A., Alavere H., Krebs K., Reisberg S., Hudjashov G., Pankratov V., Nelis M., Lind S., Maal K., Läll K., Sokurova D., Mägi R., Kasela S., Luitva LB., Metspalu A., Metspalu M.*, Milani L.*
Institute of Genomics, University of Tartu, Tartu, Estonia

Objectives of the portal

1. Provide **individual results** to all interested participants of the EstBB (over 200,000);
2. Serve as a **platform for research.**
3. Ensure **transparency** regarding the use of participants' data;
4. Improve public **health and genomic literacy**;

Portal Contents

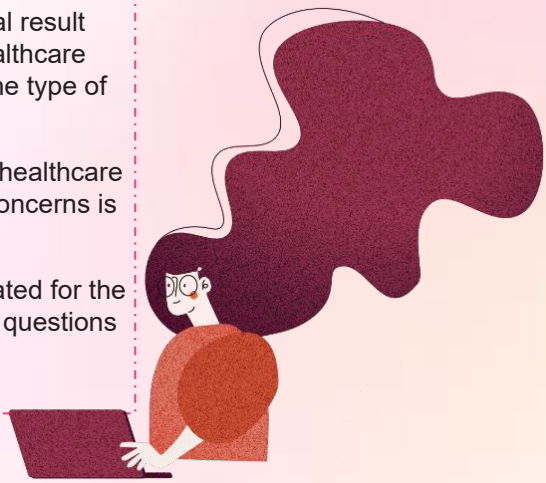
The MGP is structured into three sections: personalized results, educational content, and studies. The results section currently includes **polygenic risk scores, pharmacogenomics and ancestry**. The educational content complements the results section providing more detailed background information.

Support

Video tours of the individual result reports are available for healthcare workers to be informed of the type of contents offered.

Helpline for participants or healthcare workers with questions or concerns is available.

FAQ are collected and updated for the helpline, and most frequent questions added to the end of reports.



Unique considerations

Portal serves as a **tool** for the biobank to fulfil its objectives and is **not a research** project in itself**.**

Dynamic consent applied before entering MGP.

For data and measurements considered in risk score calculation the sources for information are shown for **transparency**.

Some reports from the individual results section registered as **inhouse medical devices.**

Launch

1000 invites sent for user testing phase. **374 respond**. Feedback collected and contents improved.

10 000 invites sent for load testing. **3659 respond**. Feedback collected and contents improved.

Open for all biobank participants. 70 000 participants visit portal within first week, 80K within three months, and over **90K** within 6 months. Feedback collected from all portal users.

First impressions

- Participants appreciate simple language, information conveyed in various ways, interactive tools, recommendations on how to modify their overall risk. Negative feedback largely regarding results not received, topics not covered, or time taken for offering results.
- After MGP was introduced in nightly news it soon became a hot topic on social media – the recreational information in particular (ancestry and coffee metabolism). Similarly, becoming a biobank participant became a hot topic again (top 3. searches for google in 2024).

For more information 1. biobank.ee 2. portaal.geenidoonor.ee

The CoMorMent project has received funding from the European Union's Horizon 2020 Research and Innovation Programme under Grant agreement 847776

Genomics education in low/middle income countries: The Golden Helix Conferences

Christina Mitropoulou [1], George P. Patrinos [1,2],

[1] The Golden Helix Foundation, London, UK; [2] University of Patras School of Health Sciences, Department of Pharmacy, Patras, Greece

Genomics conferences: Only for the lucky few?

There are significant discrepancies in University curricula in the field of Genomic and Personalized Medicine among different regions worldwide. For example, despite the fact that well-structured continuous medical education is only available in certain countries, such as the UK, the USA, the Netherlands, etc, there are no such options in other countries.

Furthermore, prestigious Genomic Medicine conferences are organized in developed countries, contrary to low/middle income countries. As such, accessibility of biomedical scientists and healthcare professionals from low/middle income countries to prestigious Genomic Medicine conferences is often limited due to budget constrains and logistics and travel restrictions.

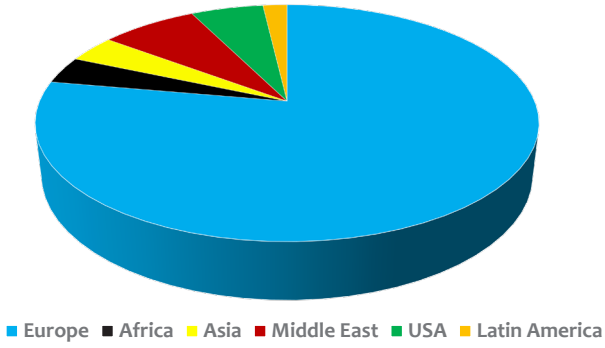
The Golden Helix Conferences

The Golden Helix Conferences is an international high caliper conference series in the field of Genomic and Personalized Medicine, organized mostly in resource-limited settings to facilitate access of healthcare professionals and researchers from low/middle income countries. There are 3 types of the Golden Helix Conferences:

Golden Helix Symposia, which are research scientific meetings, with a 2 to 4 days duration and average attendance 450 participants (ranging from 250 to 900). Conference venues are usually prestigious locations in major cities or summer retreats. **Sixteen Golden Helix Symposia** have been organized since **2008**.

Golden Helix Pharmacogenomics Days, which are half-to-single day educational events, with an average attendance of 150 participants. Conference venues are usually major cities or academic hospitals to advance pharmacogenomics knowledge. **Thirty five Golden Helix Pharmacogenomics Days** have been organized since **2009**.

Golden Helix Summer Schools with a duration of 4-to-5 days, and an average attendance of 90 participants, are organized in Greek islands, spanning across various Personalized Medicine themes. **Six Golden Helix Summer Schools** have been organized since **2014**.



Impact and major outputs of the Golden Helix Conferences

Since **2008**, there have been **57 Golden Helix Conferences** that have been organized in **40 cities in 22 countries in 4 continents** worldwide. These conferences have attracted more that **8000 delegates** from all over the world, from which approximately 10% are recurrent participants.

Interestingly, more than **60%** of these conferences have been organized in **low and middle income countries**.

Also, apart from advancing genomics education of a large number of healthcare professionals and biomedical scientists especially from LMICs, these conferences

(a) have encouraged the **formation of regional research networks**, some of which led to European-funded projects, such as **Serbordis-Inn** (FP7) and **PharmGenHub** (Horizon-Europe),

(b) catalyzed the establishment of two major regional Personalized Medicine projects (**Euro-PGx** in Europe and **SEAPharm** in Southeast Asia)

(c) have been conceptually successfully **replicated** in three major European-funded projects (**U-PGx**; Horizon 2020, **PharmGenHub**; Horizon Europe) and **Genome of Europe** (Horizon Europe).

Conclusions and future perspectives

The Golden Helix Conferences have significantly contributed in **enriching genomics knowledge** and expanding genomics education worldwide. In addition, these conferences have **developed research collaborations** between PIs, especially bridging developed and low/middle income countries from various regions worldwide. Lastly, these conferences have **encouraged student and junior researchers' mobility** from low/middle income countries to centers of excellence in developed countries through the **Golden Helix scholarships**.

BioSTEM: Raising genomics awareness for the next generation of citizens

George P. Patrinos [1,2], Christina Mitropoulou [2]

[1] University of Patras School of Health Sciences, Department of Pharmacy, Patras, Greece; [2] The Golden Helix Foundation, London, UK

STEM Education and Personalized Medicine

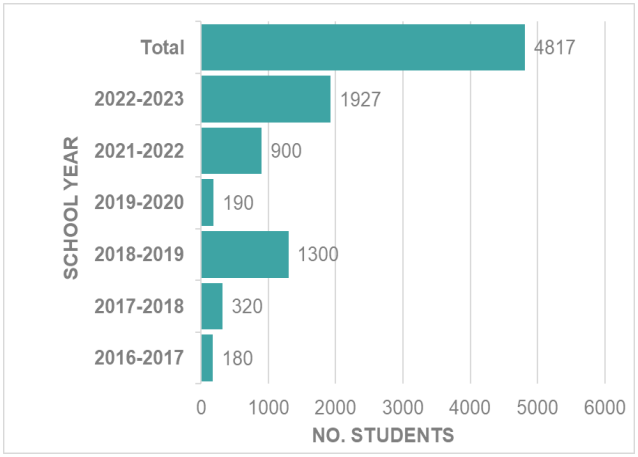
The STEM approach focuses on student-centered learning, with an emphasis on experimentation and inquiring and is considered best practice to develop students' skills in science, technology, engineering, and mathematics.

Scientific fields like Biology, Molecular Biology, Genetics, and Personalized Medicine are rapidly evolving. However, in Greece, the education of elementary, middle, and high school students in these fields is mostly limited to theoretical knowledge. To modernize biology education in schools, we have taken a bold step towards modernizing biology teaching. Our solution is BioSTEM – an innovative educational tool that focuses on education in the fields of Molecular Biology, Genetics and Personalized Medicine. It aims to bridge the gap between higher education and primary/secondary education by promoting inquiry-based and "hands-on learning" teaching methods in Molecular Biology and Genetics, while it also actively engages the students' parents, hence contributing to raising general public's genomics awareness. The initiative has received funding from the Hellenic Foundation for Research and Innovation.

The evolution of BioSTEM is a remarkable journey of growth and success

The BioSTEM initiative aims to enhance the existing curricula by focusing on the STEM approach, supporting school teachers in remote areas, and fostering collaboration between schools and society.

Students from elementary, middle, and high schools in Greece, as well as university students from Romania, Serbia, Moldova and the United Arab Emirates, have already participated in BioSTEM activities.



The graph above depicts the overall participation in BioSTEM activities from 2016-2023. In this period, a total of more than 5500 students from Greece and other countries participated in the various BioSTEM activities.

This is the first time that a University is fruitfully partnering with schools to implement a STEM approach in Molecular Biology and Genetics in more than 27 cities in Greece and 4 countries abroad, under the umbrella of the Golden Helix Foundation (London, UK), while our train-the-trainers initiative was also proven to be highly efficient in enhancing high school teachers knowledge in these modern disciplines.

Conclusions and future perspectives

According to our current research findings, students participating in BioSTEM activities are more engaged, think critically, collaborate more, and unleash their creativity when compared to the traditional teacher-centered approach (manuscript in preparation). BioSTEM was proven to be an important initiative to communicate scientific information effectively to the public, and hands-on learning is an effective way to understand biological science and laboratory findings.

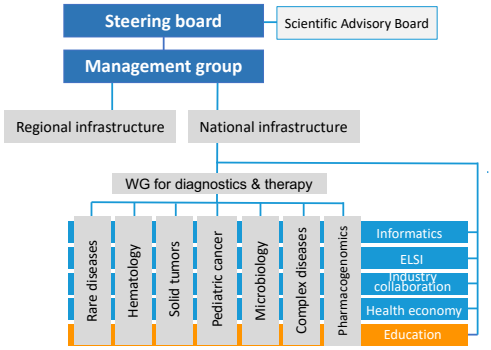
The future goal of BioSTEM is to continue to educate students and teachers in the fields of Genetics and Personalized Medicine, not only in Greece but also in other countries abroad, especially in low/middle income countries, through partnering with other academic entities and foundations. Participants have the opportunity to learn about genetic testing and the importance of human genome analysis, Personalized Medicine interventions, and their significance for the national health systems and the improvement of the quality of life of the patients.

Genomic Medicine Sweden: Advancing Education & Patient Involvement in Healthcare

Mikaela Friedman[1,2], Maria Johansson Soller[1,2,3], Charlotta Ingvoldstad Malmgren[4]
[1]Genomic Medicine Sweden, [2]Karolinska Institutet, [3]Uppsala University Hospital, [4]Karolinska University Hospital, Sweden

A National Initiative for Genomic Medicine

- Genomic Medicine Sweden (GMS) is dedicated to integrating genomic medicine into Swedish healthcare, ensuring equal access to personalised medicine for all patients.
- Collaboration between university healthcare regions and medical faculties drives this mission.
- Currently, Sweden lacks a national body for genomic medicine workforce education.



Empowering Education with the Patient Voice

In collaboration with **Network Against Cancer** and **Rare Diseases Sweden**, GMS co-develops educational initiatives in genomic and personalised medicine. Together, we:

- Integrate patient perspectives in seminars and healthcare dialogues
- Develop accessible information materials
- Emphasise the value of patient involvement in advancing genomic medicine

Digital & On-Demand Training for Healthcare Professionals

- Over **600 participants annually** (>50% from healthcare) attend monthly digital seminars, plus several hundreds taking part of each seminar through GMS YouTube.
- Topics cover **genomic and personalised medicine** in cancer, rare diseases, microbiology and complex diseases.
- Webinars are offered **live with discussions** and **on-demand**.
- A **2022 survey** of Swedish healthcare professionals highlighted the most desired but underutilised education formats:
  - 51.9% Online webinars & courses
  - 46.6% External courses
  - 44.4% External seminars & conferences
  - Only 28.6% preferred university courses
  - Many sought multiple education formats

GMS YouTube

Collaborative Development of Education & Training

GMS partners with universities, industry, and patient organisations to create educational materials, including:

- Massive Open Online Course (MOOC) – Introduction to Personalized Medicine
- E-learning module – Personalised Medicine in Breast Cancer

Scan to visit our website

gms Genomic Medicine Sweden

Contact: Mikaela Friedman, mikaela.friedman@ki.se

Global Genomics Education and Training Summit, Athens, 2025

"GENOMIC EDUCATION & TRAINING FOR INTEGRATED MULTI-DISCIPLINARY HEALTHCARE- INITIATIVES IN THE INDIAN SUBCONTINENT"

Dhavendra Kumar, South Asia Genomic Healthcare Alliance, The Genomic Medicine Foundation (UK)

South Asia Genomic Healthcare Alliance [SAGHA]

"To organise and lead a professional group for promoting and supporting the genomic applications and translations in medicine and healthcare across South Asia"

Current projects of SAGHA

- Indo-UK Genetic Education Forum
- South Asia Birth Defects Consortium
- Indian cancer genetics interest group
- South Asia cardiovascular genomics consortium
- Decipher India (South Asia) consortium
- South Asia Pharmacogenomics Consortium
- South Asia Genome Ethics consortium

UK INDIA GENOMIC MEDICINE ALLIANCE

Global Genomic Medicine Collaborative

THE GENOMIC MEDICINE FOUNDATION (UK)
FELLOWSHIP, SCHOLARSHIP, EDUCATION AND PROMOTION OF GENOMIC APPLICATIONS IN MEDICINE, HEALTHCARE AND SOCIO-ECONOMIC PROGRESS

For all Enquiries: www.genomicmedicine.org;md@genomicmedicine.org

Globally accessible and shareable genomics education-related resources, from the University of Glasgow (UK) and the HUGO, ESHG, ECMGG-examination, & APOGeE-e-textbook committees.

Prof Edward S. Tobias[1,2,3,4,5,6]

[1]Academic Unit of Medical Genetics, University of Glasgow (UofG), UK. [2]NHS Clinical Genetics, West of Scotland Centre for Genomic Medicine, Queen Elizabeth University Hospital, Glasgow, UK. [3]HUGO-International Education Committee. [4]ESHG Education Committee. [5]UEMS-ECMGG Steering Committee. [6]ERN-ITHACA-APOGeE Editorial Board.

Summary

Edward (Ed) Tobias co-chairs the Human Genome Organisation (HUGO) Education Committee (EC) & is an active invited member of several other international ECs (including the ESHG, ECMGG clinical professional exam & ERN-APOGeE e-textbook committees). He has created or co-created a range of educational resources, now shared globally & mostly free. The many challenges addressed include the need to provide educational resources for genetics & also non-genetics professionals. Although already accessed from over 140 countries, a remaining challenge is to facilitate access to the resources by the many professionals worldwide who could benefit from them.

(1) APOGeE: a free online medical genetics textbook for professionals, by ERN-ITHACA

https://my.ltb.io/www/#/stack/ABPCO

Figure 1: APOGeE. An online interactive medical genetics textbook by authors from several countries. It is up-to-date, comprehensive (with >40 sections), detailed, illustrated & free to access upon online registration.

(2) EuroGEMS: a free online guide to >100 worldwide genomics resources: www.EuroGEMS.org

A free web-based guide to a wide range of selected international genetic and genomic educational sources, created for all audience levels. Peer-reviewed, scrutinised, endorsed & used by the Eur. Soc. of Human Genetics (ESHG) & HUGO. Now in English, Spanish, Portuguese & French, and accessed from 143 countries.

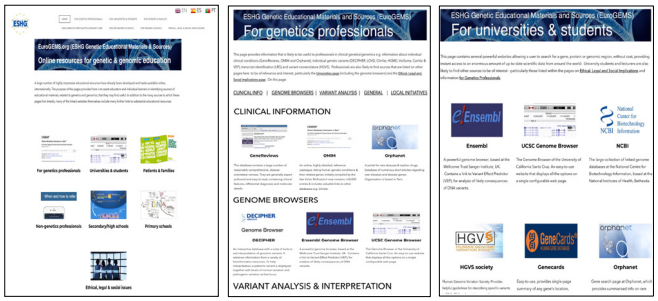


Figure 2: www.EuroGEMS.org (a) the English-language home page; and first sections of the web-pages for (b) genetics professionals and (c) universities and students.

(3) Free genomics apps for smartphones, tablets & 3D molecular visualisation virtual reality (VR) headsets

A set of free genetics quiz apps & more advanced clinical genomics apps, for students & professionals worldwide. These explain clinical genomics & bioinformatics terminology & permit self-assessment. The molecular visualisation education app enables molecular comparison & analysis in virtual reality (VR).

Figure 3 (left): Free smartphone guide & quiz apps, explaining genomics terminology. Used in over 70 countries, by >5000 people www.genomicsapps.org & (**right**) the molecular visualisation 3D virtual reality app, that can analyse >200,000 PDB protein structures. www.edify.ac

(4) Massive open online courses (MOOCs)

The set of FutureLearn massive open online courses (MOOCs) on clinical genetics, medical genomics and cancer genomics, developed together with the UofG Medical Genetics & Genomics MSc teaching team, have been used by >50,000 learners from >115 countries. Rated 4.9, 5.0 and 4.8, respectively.

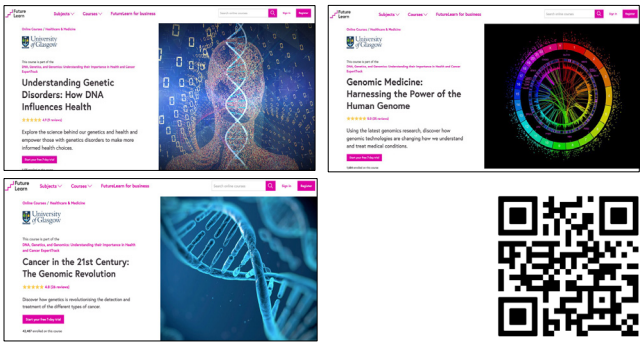


Figure 4: The set of MOOCs on genetics, genomics & cancer genomics, on the FutureLearn online platform.

(5) The European Certificate in Medical Genetics & Genomics (ECMGG) examination. www.uems-ecmgg.org

Figure 5: Examiners & candidates at the first ECMGG professional exam, held in Sweden, but which is now held annually online, with a detailed published syllabus. It is open to medical genetics candidates, worldwide, is UEMS-accredited, ESHG-supported & increasingly internationally adopted. 108 geneticists from 30 countries have now passed the exam.

(6) Human Genome Organisation (HUGO-International) Education Committee

The HUGO Education Committee consists of 6 active sub-committees, that run international courses & educational workshops; conduct global expert surveys; and publish educational curricula & resources.

Figure 6: The HUGO Education web pages, containing links to genetic & genomic educational resources, to the major professional genetics societies in each continent & to online courses on genetics & genomics.

Please get in touch

Efforts are underway to increase global access to these resources, including from LMICs. Please email Prof Edward (Ed) Tobias at Edward.Tobias@glasgow.ac.uk with any questions, suggestions or new ideas for collaboration or dissemination – these are always welcome!

Go to https://qrco.de/edtobias or scan this QR code, for all of the above links & FURTHER INFORMATION:

Advancing Genomic Education Globally:

Competencies and Credentialing for Genomics Nurse Educators

Deborah O. Himes[1] PhD, APRN; Jennifer R. Dungan[2] PhD, RN; Sarah Dewell[3] PhD, RN; Sarah H. Davis[1] MS, APRN; Linda D. Ward[4] PhD, APRN; Ruth F. Lucas[5] PhD, RN

[1]Brigham Young University, USA; [2]University of Florida, USA; [3] Thompson Rivers University, Canada; [4]Clemson University, USA; [5]University of Connecticut, USA

Aim:

- Advance genomic education for nurses globally.
- Develop a *Competency Framework for Genomics Nurse Educators*.

Methods:

- Framework developed using a structured, multi-phase approach (figure to the right).
- International Delphi panel validated framework.

Results:

- Expert panel included genomics nurse educators, researchers, and administrators (n=24) from 11 countries.
- Consensus reached on competency framework (figure below), and 42 competencies.

Discussion:

- Integration of genomics into nursing practice requires academic nurse educators equipped to deliver genomics instruction.
- This framework describes the role expectations for *Genomics Nurse Educators*.
- A portfolio-based credential for *Genomics Nurse Educators* is underway to:
  1. Support professional growth and career advancement internationally.
  2. Foster mentoring and peer review networks among genomics nurse educators.
  3. Ensure sustained expertise in genomics nursing education globally.

Plan: The International Society of Nurses in Genetics (ISONG) appointed a steering committee to develop competencies for *Genomics Nurse Educators*.

Identify Contexts of Practice & Explore Practice: Drafted competency framework through engagement with international stakeholders and literature review.

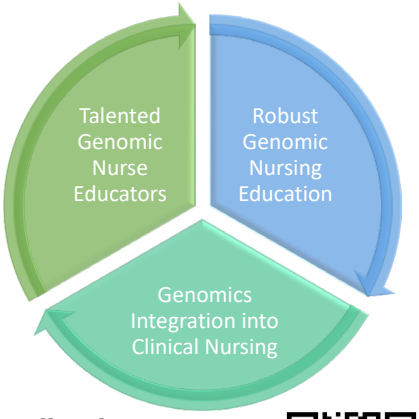
Translate & Test: Three-round Delphi study; Mar – Aug 2023. Expert panel reviewed, revised, and came to consensus on framework and competencies.

Portfolio-Based Credential: International academic nurse educators will have the opportunity to submit portfolios to become credentialed as *Genomics Nurse Educators*.

Evaluate, Update & Maintain: Ongoing evaluation will ensure competencies reflect current practice and assess the impact on nursing education.

Competency Framework for Genomics Nurse Educators

Domain 1	Domain 2	Domain 3
Foundational Knowledge for Genomics Nurse Educators	Teaching Expertise in Genomics Nursing Education	Advancing the Specialty of Genomics Nursing Education
Competency Area 1.1: Demonstrates Expertise & Experience in Academic Nursing Education	Competency Area 2.1: Expertly Designs & Teaches Genomics Nursing	Competency Area 3.1: Acts as a Change Agent & Leader in Genomics Nursing Education
Competency Area 1.2: Acquires & Maintains Genomics Knowledge	Competency Area 2.2: Facilitates Genomics-Informed Learner Development & Socialization	Competency Area 3.2: Develops & Maintains Academic-Practice Partnerships Related to Genomic Care
	Competency Area 2.3: Uses Assessment & Evaluation Strategies in Genomic Nursing Education	



Funding by:
ISONG and a grant from Brigham Young University

The Global Genomics Nursing Alliance (G2NA): Working to ensure wide representation in the development of Global Essential Competencies in Genomics for All Nurses

Emma Tonkin, PhD[1], Kathleen Calzone, PhD, RN, AGN-BC, FAAN[2], Andrew Dwyer, PhD, FNP-BC, FNAP, FAAN[3], Sarah Dewell, PhD, MSc., RN[4], Laurie Badzek, LLM, JD, MS, RN, FNAP, FAAN[5]

[1] Genomics Policy Unit, Faculty of Life Sciences and Education, University of South Wales, United Kingdom. [2] Genetics Branch, Center for Cancer Research, National Cancer Institute, National Institutes of Health, United States. [3] Connell School of Nursing, Boston College, United States. [4] School of Nursing, Thompson Rivers University, Canada. [5] Ross & Carole Nese College of Nursing, Penn State University, United States

G2NA Overview and Representation

G2NA is open to anyone with an interest in the 'Genomics within Nursing' agenda. We work across **Practice, Education & Training, Research, Policy & Leadership**

Vision: To serve as the unified international voice for advancing and integrating genomics into nursing practice.

Mission: To support nurses to realise their full potential through integrating genomics across nursing practice to improve healthcare for all.

Why Global Competencies?

- Competencies that describe the knowledge, skills and behaviours for professional practice are a key driver for change[1].
- They provide the basis for establishing education and training curricula and enable the measurement of competence to practice.
- Current competencies in genomics for nurses have been established in high income countries[2&3] and repurposing may not be appropriate.
- There is global variation in entry level nurse training, scope of practice, health systems and infrastructure to provide genomic based healthcare.

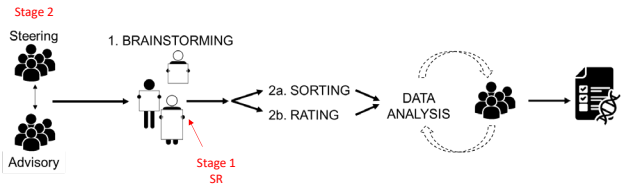
Aim

To define and promote the implementation of ***minimum competencies in genomics required by all nurses irrespective of professional role, academic preparation, country, healthcare system or population that they serve.***

- A framework that is harmonized across international contexts can be used to build capacity and can be adapted and built on to reflect country specific context and needs.

Approach

- **Group Concept Mapping (GCM) –** participatory mixed methods approach (qualitative data collection + quantitative analysis) using online social research platform groupwisdom™ [***groupwisdom.com***]



- A scoping review (SR) of established health professional competencies in genetics and genomics (Stage 1, completed) will be used to supplement but not lead or influence the participant's brainstorming component of the GCM (Stage 2)

- G2NA is actively developing links within Sigma (a professional nursing organisation with >100,000 members globally), WHO Collaborating Centres for Nursing and Midwifery, and the International Council for Nurses (a federation of >130 national nursing associations) to achieve geographically diverse participation from countries where nursing has not typically been associated with genomics.

References

1. Kirk, M., Calzone, K., Arimori, N. and Tonkin, E. (2011), Genetics-Genomics Competencies and Nursing Regulation. Journal of Nursing Scholarship, 43: 107-116. https://doi.org/10.1111/j.1547-5069.2011.01388.x
2. NHS England Genomics Education Programme (2023) The 2023 Genomic Competency Framework for UK Nurses
3. Calzone, K. A., Stokes, L., Peterson, C. & Badzek, L. (2024). Update to the essential genomic nursing competencies and outcome indicators. *Journal of Nursing Scholarship*, 56, 729–741. https://doi.org/10.1111/jnu.12993

How to Teach Genomics: A Workshop for Nursing and Midwifery Educators

Tonkin ET[1], Miller E[2], Pichini A[3], Coulson J[4], Creavin T[5], Hume J[5], Murphy JC[5], Bishop M[5]

[1] Genomics Policy Unit, Faculty of Life Sciences and Education, University of South Wales, United Kingdom; [2] Genomics Education Programme, NHS England, United Kingdom; [3] Genomics England, United Kingdom; [4] University of York, United Kingdom[5] Wellcome Connecting Science, United Kingdom

Context – Why this workshop is needed

Nurses and midwives comprise ~50% of the global healthcare workforce and play a crucial role in delivering the benefits of genomics to everyone.

Genomics has typically not been systematically incorporated into pre-qualification curricula. Internationally, numerous challenges are recognised including professional (lack of leadership or strategic direction from national bodies); organisational (lack of support from universities to revise curricula) and individual (lack of suitable educators).

Educators may not feel confident or be competent to teach genomics. This may include having no/limited experience of genomics within a clinical setting to draw upon in their teaching; be unsure of the genomics knowledge and skills required by tomorrow's nurses and midwives or be unaware of educational resources available.

Workshop Overview

- Four-day, residential workshop at the Wellcome Genome Campus, Cambridge, UK (2022 & 2024).
- Aimed at new and established educators of nurses (all fields) and midwives.
- Focus on undergraduate / 'pre-qualification' curricula (re)development, and the design and delivery of learning sessions to meet the knowledge, skills and behaviour requirements of nurses and midwives providing care that is informed by genomics.
- The course was not designed to teach genomics to participants.
- Learning outcomes were met through a mixture of presentations (including guest speakers), interactive sessions and group work.
- Evaluation of the 2022 workshop informed revisions to the 2024 programme.

Internationalization

Originally developed with a focus on incorporating genomics from a UK perspective, applications from non-UK-based educators evidenced the need internationally for this type of course.

Participants welcomed and identified additional benefits of the inclusion of more overseas educators and session content that had an international focus in 2024.

Evaluation

- At Level 1 of the Kirkpatrick Evaluation Model 'reaction', all respondents to the 2024 post-course survey, indicated high levels of satisfaction, usefulness and enablement as an educator, across all questions.
- Over the duration of the workshop, participants developed a more sophisticated understanding of genomics that moved from a focus on scientific concepts to one that included ethics and personalized medicine.
- Whilst participants were optimistic about the opportunities around genomics education, they identified many of the enduring challenges to incorporating genomics within pre-qualification health-professional curricula.

- To evaluate changes in 'behaviour' and 'results'/organisational benefit (Kirkpatrick Levels 3 and 4), participant's completed *Commitment cards* detailing what they intended to achieve on returning to their university. Six-month post-workshop follow-up is currently underway to understand what has or hasn't been achieved and why.

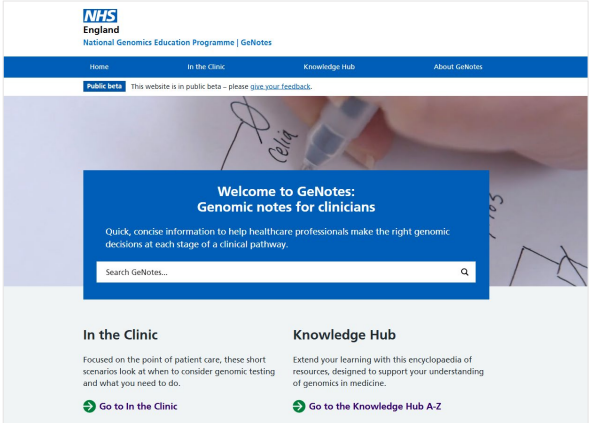
GeNotes: an NHS England genomics education resource for clinicians, with global reach

Amy Frost[1], Aine Kelly[1], Ben Armstrong[1], Elizabeth Barnett[1,2], Heidy Brandon[1,2], Amelia McPherson[1] Siobhan Simpson[1], Charlotte Szczepanik[1], Rhian Thomas[1,3], Elliott Williams[1] and Kate Tatton Brown[1,2,4]

1. NHS England's Genomics Education Programme; 2. St George's Hospital NHS Trust, London; 3. Great Ormond Street Hospital for Children NHS Trust, London; 4. St George's, University of London, UK

GeNotes (Genomic Notes for Clinicians) is an NHS England online, just-in-time, free to access, genomics education resource, supporting clinicians with requesting and receiving results of genomic testing.

GeNotes structure – two tiers



Tier 1 'In the Clinic' resources support clinicians at the point of patient care. They are:

- built around clinical scenarios;
- specialty-specific;
- written to a strict template to facilitate rapid access to consistent information;
- aligned to the NHS England National Genomic Test Directory; and
- Peppered with links through to Tier 2, providing 'hooks' for learning.

Tier 2 'Knowledge Hub' resources offer opportunities to extend genomics knowledge. They are:

- Varied, covering topic such as:
  - core concepts;
  - genetic conditions; and
  - genomic technologies.
- Engaging, with multimedia content including:
  - written articles;
  - infographics;
  - animations;
  - films; and
  - visual communication aids
- Collaborative, signposting to external links to relevant websites and information.
- Accessible, via links from Tier 1, standalone, or can be assembled to create bespoke learning packages according to training needs.

Co-design and user research

GeNotes has been co-designed and co-created with clinicians, allowing insight into the education needs, challenges and learning styles of end-users.[1] The utility of this approach was supported by excellent user feedback across the tested domains (Figure 1).

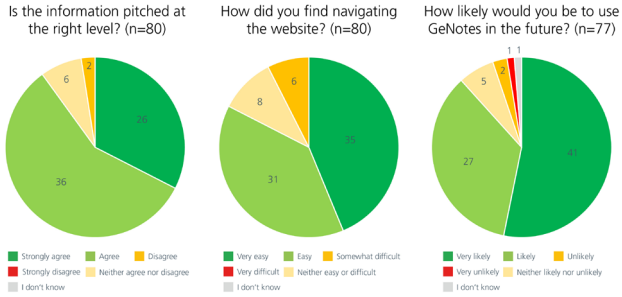


Figure 1: GeNotes user research feedback (content, navigation, likelihood of use)

Increasing use and global reach

Since launch in June 2022, GeNotes use has increased month on month, with a total of 478,449 visitors and 864,664 page views to date (December 2024).

GeNotes has found an international audience; in 2024 it was accessed by users from more than 190 different countries, the top 10 of which are shown below (figure 2).

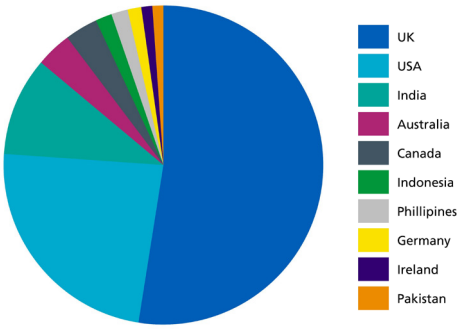


Figure 2: GeNotes top 10: percentage visitors by country, 2024

Take a guided tour of GeNotes:
bit.ly/GeNotes_tour

Footnotes

1. Frost A, Kelly A, Bishop M, Bogue D, Copson E, Gompertz L, Hay E, Hayward J, Hendriks E, McVeigh T, Simpson S, Tatton-Brown K. 'Genotes – a 'just-in-time' genomics education resource co-designed with clinicians'. BMC Med Educ 24, 1378 (2024). DOI: 10.1186/s12909-024-06059-w

GP with Extended Role Clinical Genetics / Genomics: A use case of the Genomics Advisors Framework for workforce development

Hayward J[1], Miller E[1], Evans W[2], Brooks M[1], Bunce S[1], Khan A[1,3], Norman M[1], Robinson N[1], Wong, J[1], Rafi I[1,5], Tatton-Brown, K[1,4,5].
1. NHS England's Genomics Education Programme; 2. Primary Care Stratified Medicine, University of Nottingham; 3. University of East London; 4. St George's Hospital NHS Trust, London; 5. St George's, University of London

Overview

General Practitioners (GPs) deliver clinical roles extending outside core Primary Care in a variety of specialties, captured in professional development infrastructure within the UK's Royal College of General Practitioners (RCGP) and annual GP appraisal.

The Genomic Advisors Framework (GAF) is a workforce development tool for non-genomics healthcare professionals to deliver leadership and clinical service roles. The GP with Extended Role (GPwER) Clinical Genetics / Genomics framework was developed using the GAF as an underpinning structure. This demonstrates:

1. The utility of the GAF as a generalisable role- and specialty-agnostic tool in supporting competency framework development within specialist professional body infrastructure through development of the GPwER Clinical Genetics / Genomics framework.
2. The GPwER Clinical Genetics / Genomics framework as a mechanism for GPs to contribute to NHS Genomic Medicine Service delivery in other non-genomic specialties and community settings, through provision of clear governance structure and processes.

The GPwER Clinical Genetics / Genomics framework

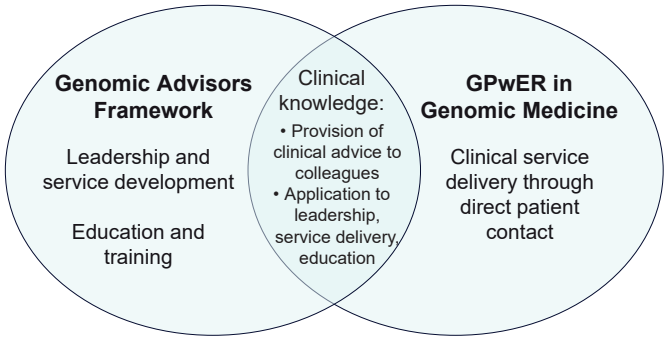
"Within the clinical genetics service, I deliver genomic testing for inherited cancer syndromes, care co-ordination for those with neurofibromatosis type 1, and advise the team on contraception and hormone replacement therapy in these groups."
Dr Jude Hayward

"Within the clinical genetics service, I assess children with learning difficulties and other health problems, deliver genomic testing and interpret results under consultant supervision, as well as support pathways for rare disease."
Dr Will Evans

The Royal College of General Practitioners (RCGP) has an established professional framework for GPs who deliver clinical roles within clearly defined service and governance infrastructure outside of Primary Care (GPwER roles)

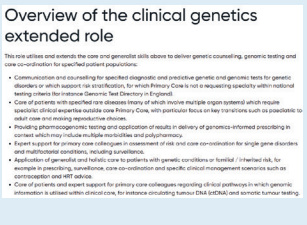
The working group developed the GPwER Clinical Genetics / Genomics framework aligned to both the Primary Care GAF and template RCGP GPwER frameworks, subsequently reviewed by clinical genetics consultant and within internal RCGP Professional Development processes.

The diagram depicts the relationship between the GAF and GPwER frameworks, with emphasis on clinical service delivery within the GPwER role:



The GPwER role

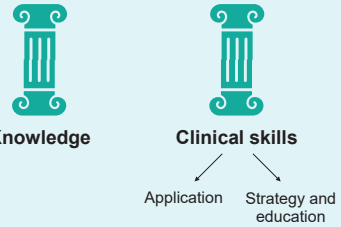
GPwER Clinical Genetics / Genomics framework captures the potential scope and activity of the role and governance, including accreditation and maintaining competence.


Overview of the clinical genetics extended role

Aims of the Genomics Advisors Framework (GAF)

1. Facilitate the **integration** of genomic medicine across the healthcare specialties and professions.
2. Support the **flow of information** from national to local and local to national level.
3. Develop a structure that will promote **consistency** across the UK, across the specialties and across the healthcare professions.

The GAF structure: two pillars



The GAF process

- Convening of working group consisting of GPs in leadership roles and clinical roles within clinical genetics, national pharmacy and nursing leads.
- Primary Care GAF is generalisable across GP and primary care allied healthcare roles, including pharmacy and nursing.
- Review and sign-off by NHS England's Genomics Education Programme.
- Excerpt from the GAF demonstrating alignment with curricula and other professional body frameworks:

2. Clinical skills: Application of genomics in clinical practice

Theme	Competencies		
	National	Regional	Local
Applying genomic principles to own practice.	Has an in-depth understanding of genomic medicine as detailed in the relevant professional body curriculum, e.g. RCGP Curriculum (including the Clinical Topic Guide Genomic Medicine) BPS or RPS post-registration foundational curriculum. Describes clinical pathways, referral criteria and further management of patients within other care settings. Can describe application of genomic medicine principles (detailed in section below) to clinical care of patients. Applies this to clinical decision-making in delivering expert advice and support to Primary Care colleagues, and throughout the activities delivered within the Primary Care Leadership for Genomics role.		

View the GPwER Clinical Genetics / Genomics framework here:

The UK Genomics Training Academy: a progress update

Jakob Whitfield[1], Sarah Clinton[1], Ed Miller[1], Melanie Watson[1], Áine Kelly[1], Alison Pope[1], Terri McVeigh[1], and Kate Tatton-Brown[1]
1. Genomics Education Programme, NHS England, United Kingdom

Key achievements

- **launch** of 'phase one' of the GTAC's virtual reality (VR) modules across UK Genomic Laboratory Hubs;
- **launch** of the new GTAC virtual learning environment (VLE);
- **development** of asynchronous, self-directed resources for training at pace and scale;
- **delivery** of multi-disciplinary synchronous training; and
- **training the trainer**: upskilling of GTAC educators in curricula mapping, online pedagogy and course design.

Why a genomics training academy?

Advances in the application of genomic technologies within UK healthcare have increased demand on the NHS specialist genomics workforce. In 2023, NHS England's Genomics Education Programme established the **Genomics Training Academy (GTAC)**[1] with the following aims:

increase training capacity

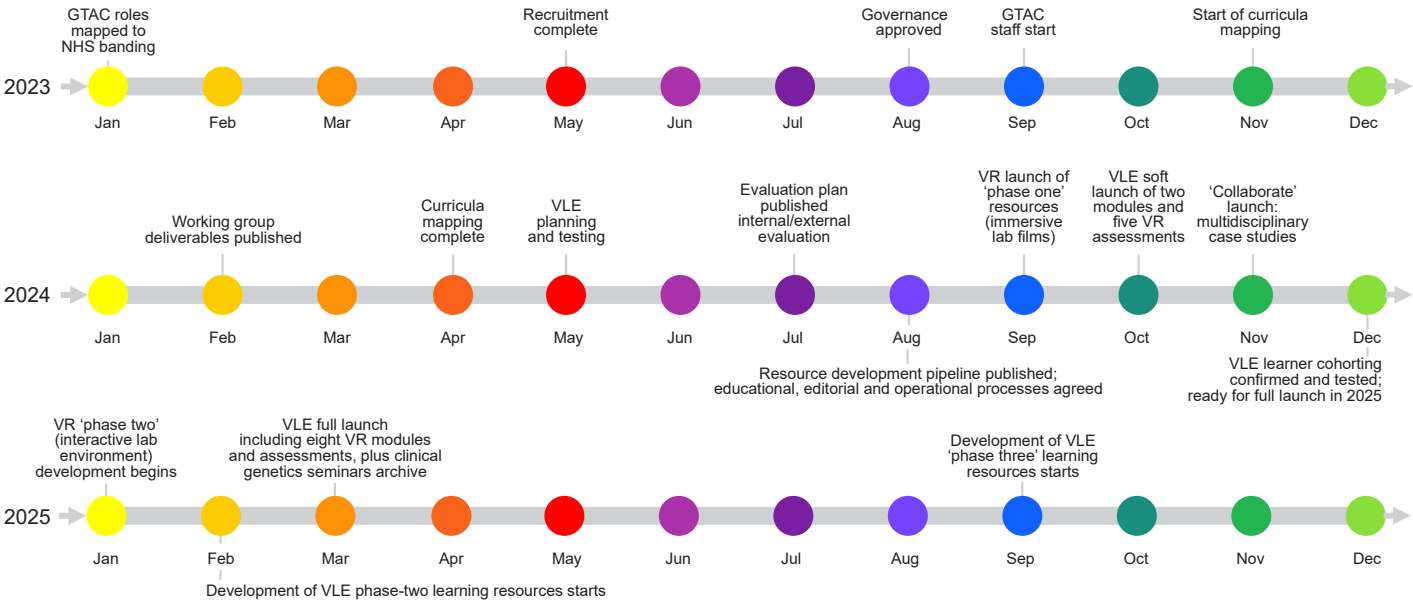
create high-quality education and training

foster innovation

establish a GTAC teaching faculty

improve delivery of genomic medicine

GTAC progress and roadmap



Case studies

Virtual reality

Scientific trainees must gain laboratory experience, but facilitating this is a training challenge for existing staff. To reduce this load, the GTAC has developed a series of immersive VR films and supporting training materials to familiarise trainees with the laboratory environment before they even step inside it. Each of the seven NHS Genomic Laboratory Hubs received a 16-headset kit (for a total of 112 headsets) from the GTAC. Development of the second phase of the VR project, which includes interactive immersive materials, is currently underway.

Collaborate: a clinical laboratory education initiative

This recurring webinar series is designed to support all genomics professionals from bench to bedside. This resource supports learners to address real clinical challenges and to:

- **learn** about genomic conditions and how they are diagnosed;
- **understand** the wider context of their roles within a clinical pathway;
- **recognise and respect** others' contributions to the stages in the pathway; and
- **build** a community of interprofessional learning.

Footnotes

1. www.genomicseducation.hee.nhs.uk/about-us/gtac-genomics-training-academy/

Scan to learn more

Leeds Clinical Genomics Service

The Leeds Teaching Hospitals NHS Trust

Culturally Competent Genomic Care - A Competence framework

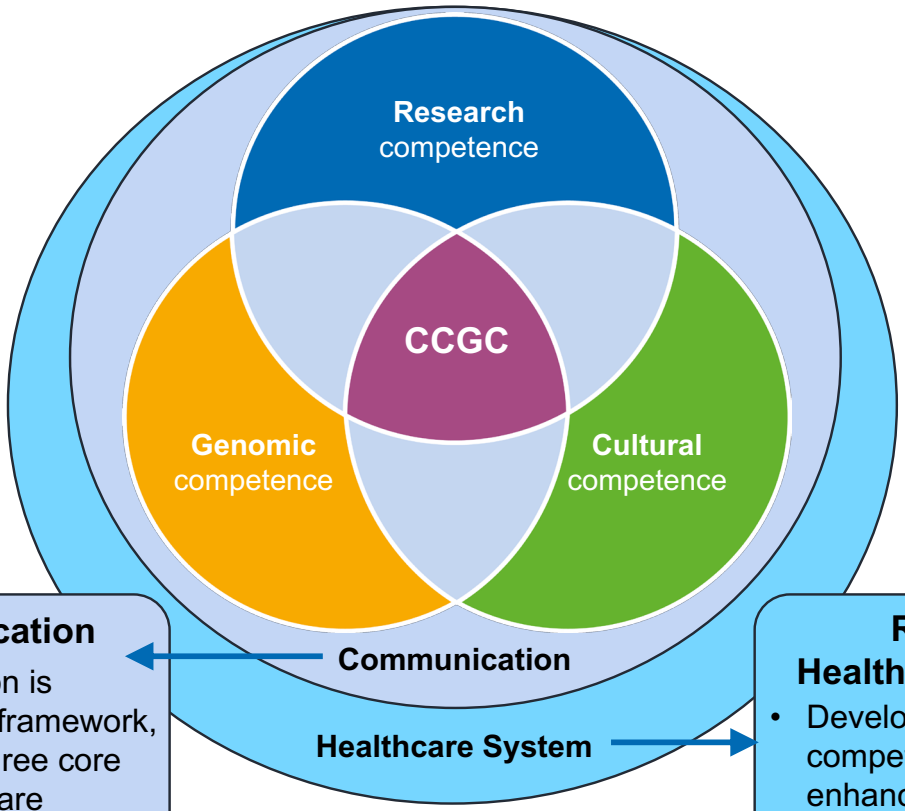
Dr Saghira Malik Sharif
Leeds Clinical Genomics Service, Leeds Teaching Hospitals NHS Trust, UK.

This framework comprises three core concepts:

● **Research competence** ● **Genomic competence** ● **Cultural competence**

CCGC

The area of overlap between the three core concepts ensures safe, ethical, and effective clinical practice in the delivery of CCGC



Communication

- Communication is central to this framework, in which the three core competences are embedded
- The synergy of these competencies relies on communication to facilitate CCGC

Roles of Healthcare System:

- Developing culturally competent strategies to enhance diversity and inclusion in genomics
- Provision of resources for genomic education and culturally competent training

References

Sharif SM, Blyth M, et al. Enhancing inclusion of diverse populations in genomics: A competence framework. J Genet Couns. 2020;29:282–292. https://doi.org/10.1002/jgc4.126

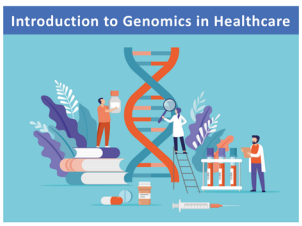
Genomics Training and Education Activities in Wales

Jonathan Hawken, Sophie Harding, Benjamin Player, Sian Nisbet, Nicola Taverner, Joanne Thomas, Ian Tully, and Alexandra Murray

Introduction

The benefits of genomic testing are well recognised and the indications for testing are expanding rapidly. However, adoption of genomic testing by non-specialist clinicians (i.e. mainstream testing) in Wales has been slower than in England. We are taking a collaborative approach to address this. Health Education and Improvement Wales (HEIW) have a dedicated Genomics team working on a range of projects. A Strategic Workforce Plan for Genomics has recently been developed, which includes a focus on mainstreaming. The HEIW team work in collaboration with Genomics Partnership Wales (GPW) and the All Wales Medical Genomics Service (AWMGS). This poster highlights some of the key projects related to mainstreaming in Wales.

HEIW Resources



HEIW are supporting mainstream clinicians to integrate genomic testing into their practice. Resources have been developed including online modules and videos covering the practical aspects of genomic testing. HEIW also fund MSc modules for healthcare professionals interested in making genomics a core part of their role. Each year HEIW hosts a series of lunchtime genomics webinars to educate the wider workforce. Topics include how to undertake testing and understand results, and pharmacogenomics.

MSc participant: *"The course was very informative and the assignments developed key skills that I have since found extremely useful."*

Non-Invasive Prenatal Testing (NIPT)

The All Wales Medical Genomics Service (AWMGS) is currently working with Antenatal Screening Wales (ASW) and Antenatal clinics from all Health Boards across Wales to provide Non-Invasive Prenatal Testing (NIPT) through antenatal services to couples who have had previous pregnancies or children diagnosed with one of the common chromosomal trisomies (Patau, Edwards & Down syndromes). Provision of NIPT via the antenatal clinics will streamline the antenatal journey for these couples, reduce the number of appointments they need to attend, and facilitate access to testing at the earliest possible gestations.

Breast surgeon: *"Patients have all been extremely keen to take up the offer of testing. In addition, more testing by us before they reach oncology without having to refer separately to AWMGS helps them with planning neoadjuvant/adjuvant treatments so they (oncology) are happy too!"*

Cancer Susceptibility Gene Testing

Mainstream testing of the breast cancer gene panel is now up and running in two health boards in Wales and we are looking to roll this out to all other health boards in 2025. Following feedback from the breast teams, standard letters have been developed for use which include advice about cascade testing for relatives. The breast teams have embraced testing and feel that patients are reaping the benefit of having access to testing earlier in their patient journey. A new pathway for Lynch syndrome testing is also under development as part of a project supported by the Moondance Cancer Initiative.

Cardiac nurse specialist: "It makes my job more interesting; I think it's working well for my patients."

Consultant-led triage

The All Wales Medical Genomics Service has recently introduced consultant-led triage of new referrals; advice letters are sent to referring clinicians when mainstream testing is appropriate. One of the aims of this initiative is to upskill the mainstream workforce in the process of choosing and arranging appropriate genomic investigations. This is supported by a video resource which explains the consent requirements for genomic testing. Feedback from mainstream clinicians about implementing testing has been largely positive.

Pharmacogenomics (PGX)

DPYD testing is available in Wales for patients who are due to receive specific chemotherapy. This pharmacogenomic test can identify those at risk of a drug reaction. It is requested directly by oncologists, which provides earlier results and avoid delays in starting treatment. This successful programme is a template for other pharmacogenomic tests including the identification of infants at risk of hearing loss from gentamicin, and the forthcoming CYP2C19 test for patients due to receive clopidogrel after an ischaemic stroke.

AWMGS

GIG CYMRU NHS WALES | Addysg a Gwella Iechyd Cymru (AaGIC) | Health Education and Improvement Wales (HEIW)

Partneriaeth Genomeg Cymru | Genomics Partnership Wales

GIG CYMRU NHS WALES

Outcomes of 3 Year Post-Doctoral Program & 15 Days Short Term Courses in Medical Genetics

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Sanjay Gandhi Postgraduate Institute of Medical Sciences, Lucknow, India
E mail: shubharaophadke@gmail.com

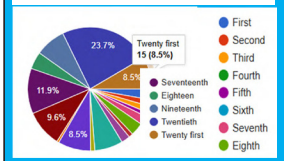
Introduction: As the first department of Medical Genetics in the country, we had responsibility of teaching. The outcomes of one 3 year program & one 2 week short program are presented.

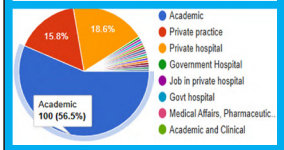
ACKNOWLEDGEMENTS: All my colleagues, students, research staff, administration, Indian Council of Medical Research have contributed in many ways to the work. And I thank the patients & their families who taught us.

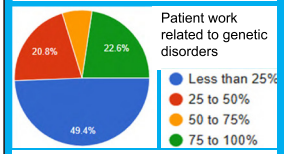
Course on Medical Genetics & Genetic Counseling
Held yearly since 2000; Duration – Two weeks
21st Course – 29th July to 10th Aug 2024
OBJECTIVES: [1] To teach basic principles of genetics & genetic diagnostics to clinicians of all specialities & at any stage of career [2] To create awareness of clinical presentations of genetic disorders [3] To empower clinicians in diagnosis & genetic counseling of common genetic disorders

Total participants- About 600 Feedbacks:177 responded (? more of those who are pursuing medical genetics)

Which course did you attend?



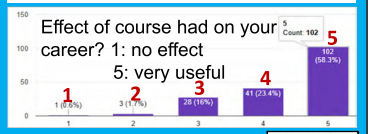




Recommended to colleagues & students – 67.4%

AFTER ATTENDING THE COURSE
54 [31.2%] did another course / program in medical genetics - DM-10, DrNB – 9, MS Genetic Counseling (Boston University)-1, Master trainer course UK-1, 4year speciality training (CCT)in Clinical Genetics- UK

CURRENT JOB
Academic – 100,NGS lab- 2
Clinical Assistant Professor, University of Nottingham-1
Fetal medicine specialist– 9
Psychiatrist New Zealand- 1



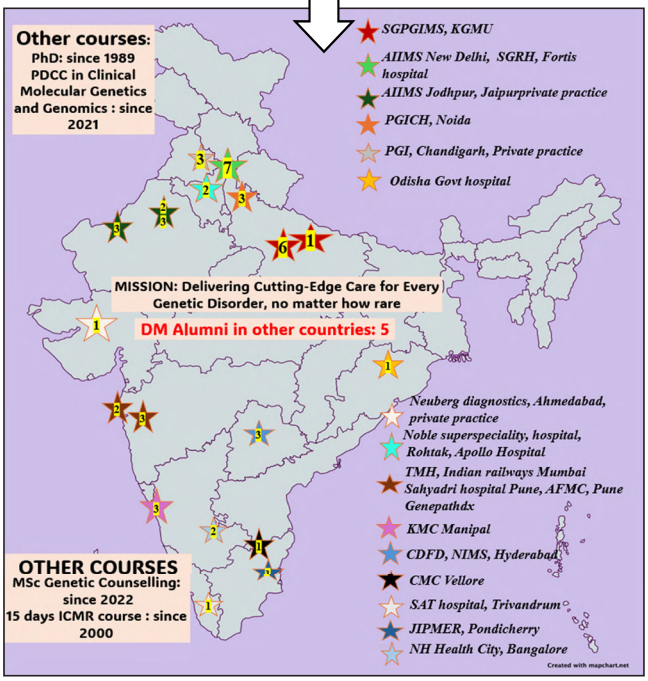
Attended any genetics related conferences, workshops, seminars after the course - 111

CONCLUSION: Well structured two week course can educate about essentials of genetics & genetic counseling. It also enthuses young clinicians to take up it as a career. Participants mostly felt that the course affected their patient care in a positive way.

Detailed program of 15 day course on MG & GC

Total	Pediatrics	Obstetrics	Fetal Medicine
255, The speciality wise distribution of last six courses held	67	87	15
	Non clinical	Med Genetics	Miscellaneous
	46	8	32

Doctorate of Medicine [DM] in Medical Genetics program- since 1990
DM pass outs- 50 (3 Years clinical + Diagnostics + 2 Research papers to be published)
SGPGIMS alumni heading various government and private institutes across the India



Representative Short Term Seminars / Symposia Organized by SGPGIMS, Lucknow Alumni

Dept of Medical Genetics KMC, Manipal – 9 Genetic Updates – 2012 to 2024
Developing brain, Bioinformatics Clinical Genetics, Exome data, Genetic disorders of Bone, Genetic counseling, Neuro -developmental, Cellular-Animal models of genetic disorders, Aortopathies

Video of Teaching courses & programs by SGPGIMS, Lucknow

Centre for DNA Finger-printing & Diagnostics, Hyderabad
Thirteen Laboratory Hands on Training Courses – 5 days each on Traditional cytogenetics, Molecular cytogeneics, Molecular diagnotics, Exome/ Long read sequencing,
Total participants - 394

CONCLUSION: Three year doctorate program trained 50 clinicians in medical genetics. This specialists not only established patient care services all over India but are also running 3 year DM /DrNB programs and short term courses / seminars/ workshops/ symposia to create trained manpower and prepare clinicians for the practice of molecular medicine.

UNIVERSITAS GADJAH MADA
FACULTY OF MEDICINE,
PUBLIC HEALTH, AND NURSING

BGS Kemenkes RS Sardjito

Lesson learnt from pioneering implementation of whole-exome sequencing for rare disease in Indonesia: role of education and training in resolving diagnostic odyssey

Gunadi[1], Pramana Adhityo[1], Petrus G Purwosatrio[1], Nova YP Budi[1], Nabila A Pramono[1], Ali Z Abidin [1], Dewiyani I Widasari[2], Kristy Iskandar[3]

[1]Pediatric Surgery Division, Department of Surgery, Faculty of Medicine, Public Health and Nursing, Universitas Gadjah Mada/Dr. Sardjito Hospital, Yogyakarta 55281, Indonesia;
[2]Department of Anatomical Pathology, Faculty of Medicine, Public Health and Nursing, Universitas Gadjah Mada/Dr. Sardjito Hospital, Yogyakarta 55281, Indonesia;
[3]Department of Child Health/Genetics Working Group, Faculty of Medicine, Public Health and Nursing, Universitas Gadjah Mada/UGM Academic Hospital, Yogyakarta 55291,Indonesia

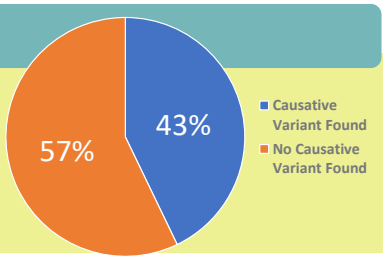
Insights from our cases

We pioneered whole-exome sequencing (WES) for rare disease in Indonesia using short reads by NextSeq 550 Illumina, which revealed genetic findings across 14 patients with rare disease: one pathogenic; one likely pathogenic; 12 variants of uncertain significance (VUS).

- 6 causative variants were identified: 3/6 patients were suspected to have causative variants requiring different diagnoses & management.
- 1 patient, a 34-year-old woman with periodic weakness & previously been diagnosed with unspecified spastic paraplegia. After years of diagnostic odyssey, the patient finally underwent WES and was found to carry a VUS in *CACNA1S* (p.Glu1488Lys), which led the clinician to suspect thyrotoxic periodic paralysis. Indeed, the patient showed dramatic improvement when treated with hyperthyroid medications, as the muscle spasms were absent after thyroid treatment.
- 2 patients were presented with spastic paraplegic type 8: a 25-year-old male who carried a *WASHC5* variant (NM_014846.4:c.2084T>C) classified as a VUS, and a 54-year-old female with the same *WASHC5* variant (NM_014846.4:c.2084T>C), also classified as a VUS.
- The latter 3 patients: 2 patients were confirmed to have Duchenne muscular dystrophy (DMD): a 30-year-old female, a manifesting carrier of DMD, who has a VUS in the *DMD* (X:g.31114692_31114695del), and a 10-year-old male with DMD who has both a pathogenic *DMD* variant (g.32809602_32809616del) and a VUS (DMD g.31114695del).
- 1 patient diagnosed with Limb-girdle muscular dystrophy type 2L (LGMD2L), a 10-year-old female, was found to carry an *ANO5* variant (NM_213599.3:c.649-27_649-26del), which was identified as a VUS.

WES or WGS? Which one provides the 'best of both worlds'?

- Our case series' diagnosis yield of WES was 42.8%, comparable to Czech population.
- Molecular genetics testing has been widely used in diagnosing rare disorders & promises a life-changing course of disease due to precision patient treatment.
- However, in our limited-resource setting, costs remain a barrier to implementing this approach. s
- Our healthcare facility's next-generation sequencing (NGS) method is WES.
- While whole-genome sequencing (WGS) may provide a more comprehensive view of genome & potentially highlight non-coding variants compared to WES, however, WES requires a lower operational cost and less complicated bioinformatic downstream analysis than WGS.



Inadequacy of continuing medical education in genomics

- In Indonesia, using WES has been pivotal in identifying genetic variants associated with rare diseases, leading to precise diagnosis and treatment.
- **Inadequate Continuing Medical Education (CME) in Genomics for Rare Diseases:** The scarcity of qualitative evidence on how healthcare workers in Indonesia are educated regarding rare diseases signifies a lack of awareness and a need for enhanced training in genomics to improve the diagnosis and management of these conditions.
- **Minimal Exposure Leads to Underutilization:** Studies indicate that a significant portion of medical students receive minimal exposure to genomics during their training; thus, despite the increasing relevance of genomic testing in clinical practice, many healthcare providers feel inadequately prepared to utilize these tools effectively.
- **Overlooked Benefits of Genomic Testing:** This further results in reluctance and hesitancy to order genomic tests, which may lead to missed diagnosis potential & overlooked benefits of genomic testing. Lack of comprehensive clinical phenotyping before genomic testing also may inadvertently lead to inappropriate tests, leading to low diagnostic yields, where overlapping clinical features complicate diagnostic processes.

What aspects should be improved in genomics education and training?

- **Integration of Genomics in Medical Curricula:** As genomic medicine becomes increasingly integrated into clinical practice, integrating genomic education into medical curricula can ensure the competency of healthcare providers in recognizing and managing rare diseases.
- **Rapid Advancements in Genomics**: Constant changes of variant interpretation classifications and guidelines mean that conclusions can quickly become outdated, potentially omitting newly identified causative genes or variants associated with rare diseases. In light of these swift developments, CME and training in genomics are essential. CME in genomics should include not only the basic concepts of genetics but also practical skills such as genomic data downstream analyses, ethical considerations and evaluation, and the application of genomic findings in patient care.
- **Phenomics**: or studies that aim to define the observable traits of genetic conditions, plays a crucial role in enhancing the diagnostic yield of genomic approaches. A thorough description of the patient's clinical presentation and history is essential to inform the genomic tests of choice. Thus, further training of phenotype-genotype correlations, the limitations of various sequencing strategies, and the importance of pre-test counseling to ensure that patients are adequately informed about the potential ethical, social, and other implications of genetic testing.
- **Role of Case-based and Practical Learning:** Moreover, integrating case-based learning and practical applications of genomic medicine into CME can enhance students' agility in navigating issues within complex cases.
- **Precision Medicine Initiative in Indonesia?** Additionally, the rise of the Indonesian Precision Medicine Initiative, known as the Biomedical and Genome Science Initiatives (BGSi), should support CME in genomics as it provides a platform for biomolecular research services.
- This initiative serves as a repository of genomic data, offers resources such as workshops and seminars, and provides a collaborative environment for various healthcare professionals, including students, geneticists, clinicians, and genetic counsellors, for sharing diverse perspectives and expertise.

Conclusion

- Genomic approaches have paved the way for precision treatment of rare diseases.
- However, physicians' lack of awareness and training concerning rare diseases and genomic approaches presents significant challenges to effectively utilizing genomic modalities.
- **Addressing these gaps through education and training initiatives is essential for enhancing healthcare providers' capacity to expedite the diagnostic odyssey in rare diseases.**

References

PANDU BIOSAINS

Challenges and Opportunities in Genetic Education in Nepal: A Qualitative Analysis of Healthcare Perspectives and Collaborative Initiative

Dr. Rupesh Mishra[1], Aassmi Poudyal[2] [1]Civil Service Hospital, Kathmandu, Nepal [2]University of New Haven, Connecticut, United States

Introduction

Genetic and genomic sciences are transforming global healthcare, driving advancements in personalized medicine, disease prevention, and treatment (WHO, 2022). However, in Nepal, the integration of genetics into healthcare and education remains in its early stages, presenting significant challenges for healthcare professionals and patients alike. This study aims to examine the challenges and opportunities in genetic and genomic education in Nepal.

Methodology

This study collected data from semi-structured, in-depth interviews with 25 participants, including healthcare professionals (HP), patient's families affected by genetic disorders, and representatives from national and international organizations, such as Rare Diseases Society Nepal (RDSN) and World Health Organization (WHO). Thematic analysis was conducted to identify the challenges and opportunities related to genetic education.

Results

The results of this study reveal both the challenges and opportunities in genetic education in Nepal, which are summarized in the following two tables.

Table 1: Challenges in Genetic Education in Nepal

Theme	Challenges	Quotes
Limited Genetic Knowledge and Training	• Healthcare professionals reported limited training in genetics. Many felt unprepared to manage genetic disorders. • Healthcare professionals lacked access to specialized genetic training programs.	• "We lack training in genetics, which makes it difficult for us to provide proper care to patients with genetic disorders." (Health Educator-1) • "We don't have access to genetic training programs. We have to rely on online resources, which are often inadequate." (Clinician)
Infrastructural and Resource Limitations	• Limited access to diagnostic tools like next-generation sequencing (NGS) creates barriers to diagnosing genetic conditions. • Rural patients face financial barriers, making it hard to travel to urban centers for testing.	• "Without access to NGS or genetic panels, our ability to diagnose rare conditions is severely constrained." (Genetic Expert) • "Traveling to cities for genetic testing is expensive." (Patient father).
Cultural and Ethical Barriers	• Misconceptions and stigma discourage families from seeking genetic counseling and testing.	• "There are still many families who are afraid of genetic testing due to cultural beliefs and social stigma." (Nurse)

Table 2: Opportunities in Genetic Education in Nepal

Theme	Findings	Quotes
Collaborative Workshops and Public Campaigns	• Workshops and public awareness campaigns organized by NMA, RDSN, and other partners have helped healthcare professionals improve genetic counseling skills and reduce stigma surrounding genetic disorders.	• "The workshops have equipped us to better explain genetic risks to families, while public campaigns have helped to reduced stigma in our community regarding genetic disorder." (Genetic expert)
Public Awareness and Patient Advocacy	• Increased trust in genetic counseling through public awareness efforts.	• "I didn't know what was wrong with my child until I attended an RDSN seminar. It gave me clarity about my child's condition." (Patient, mother)
Technology and International Collaboration	• Mobile applications, online platforms, and telemedicine can bridge the knowledge gap. International collaborations, offer expertise and resources to strengthen genetic education.	• "International partnerships have been critical in enhancing genetic education and capacity building." (RDSN-1)

Conclusion

Genetic education in Nepal faces several challenges, including insufficient training for healthcare professionals and limited resources. However, there are promising opportunities, such as collaborative workshops, public awareness campaigns, and international partnerships, to overcome these challenges and enhance genetic education.

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2) Kumar, D. (2022). Global perspectives of genomic education and training. Genomic Medicine Skills and Competencies, 107-129.

Workforce Development in Genomics in a Limited Resource Setting

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University of the Philippines Manila

BACKGROUND

Globally, there is a growing need for more trained medical geneticists and genetic counselors to support the growth of genetics services and genomic medicine. This shortage is particularly palpable in under resourced settings such as the Philippines.

The Philippines, a low middle income country in Southeast Asia, is an archipelago with 7,641 islands and a population of 119 million people. These pose unique challenges in health delivery and workforce development in the country, especially in highly specialized fields such as genomics.

CURRENT STRATEGIES TO ADDRESS GENOMICS WORKFORCE GAP

To address the severe shortage of a clinical genomics workforce, the Philippines has implemented several programs:

Post-Residency Fellowship Training Program in Clinical and Metabolic Genetics – A structured program aimed at training clinicians in genetic disease diagnosis, management, and counseling.

Master of Science in Genetic Counseling – A two-year degree program designed to train professionals who can assist patients in understanding genetic conditions.

Genetics Bootcamp for Non-Geneticist Clinicians – A flexible short-term training program providing clinicians with foundational knowledge in genomics.

Telegenetics Webinars – Virtual training sessions to improve access to genetic & genomic education & connect clinicians with experts remotely.

NATIONAL EFFORTS IN WORKFORCE DEVELOPMENT

The National Academy of Science and Technology Philippines (NAST PHL), through its Technical Working Group on Advanced Technologies in Healthcare, held a focus group discussion (FGD) on genomics in healthcare, highlighting the urgent need for workforce development. The FGD identified four key strategies:

1. **Integration of Genomics in Medical Education** – Incorporate genomics into medical school curricula to train future doctors on genomic medicine and its clinical applications.

2. **Development of a Professional Master's Program in Genomics for Clinicians** – Create an advanced training program that specializes in clinical applications of genomics for healthcare professionals.

3. **Mentorship Programs for Clinicians** – Establish structured mentorship networks that allow clinicians to learn and apply genomics in patient care and diagnostics.

4. **Advancing Genomics Research and Career Development** – Increase funding availability for genomics research, foster collaborations, and develop a structured career system for genomic researchers and professionals.

POLICY RECOMMENDATIONS FOR WORKFORCE EXPANSION

- **Scholarships and training programs** to attract and retain genomics professionals.
- **Expand workforce diversity** by training geneticists, genetic counselors, bioinformaticians, and laboratory scientists.
- **Enhance South-to-South collaborations** to build a robust genomics ecosystem.

CONCLUSION

The deliberate and intentional development of a genomics workforce is essential to accelerating equitable access to genomic healthcare. Strategic investment in **education**, **training**, **mentorship**, **policy reforms**, and **international collaboration** will be key to ensuring sustainable workforce growth and improving genomics-based healthcare in the Philippines.

ENTRUSTABLE PROFESSIONAL ACTIVITIES AS AN ASSESSMENT TOOL FOR MEDICAL SPECIALTY TRAINING PROGRAMME: IMPLICATIONS FOR MEDICAL GENOMICS

Meow-Keong Thong on behalf of The Malaysian National Postgraduate Paediatric Curriculum Committee

Introduction

Entrustable Professional Activities (EPAs) are used as assessment tools before a trainee is accepted into a formal postgraduate training program. An EPA is defined as a unit of professional practice that is fully entrusted to a trainee once the trainee has shown the necessary competence to carry out the activity unsupervised..

The use of EPAs in competency-based medical education has been validated in undergraduate medical studies. There is however little evidence of its use as an alternative assessment tool in postgraduate medical education or in the area of genomics education. The current assessment tools are knowledge-based with minimal emphasis on trainees' attitudes and professionalism

Following the initiative to develop a national unified postgraduate curriculum, each medical specialty was set up to write their respective curriculum.The EPAs were standardized using a specific template. The trainee's expected knowledge, skills, attitude and a set of positive and negative behaviors for each EPA were agreed upon by members of the paediatric writing group.

Aim

To determine the suitability of EPAs as an alternative assessment tool for postgraduate medical training program e.g. paediatric genomics in low resource settings..

Methods

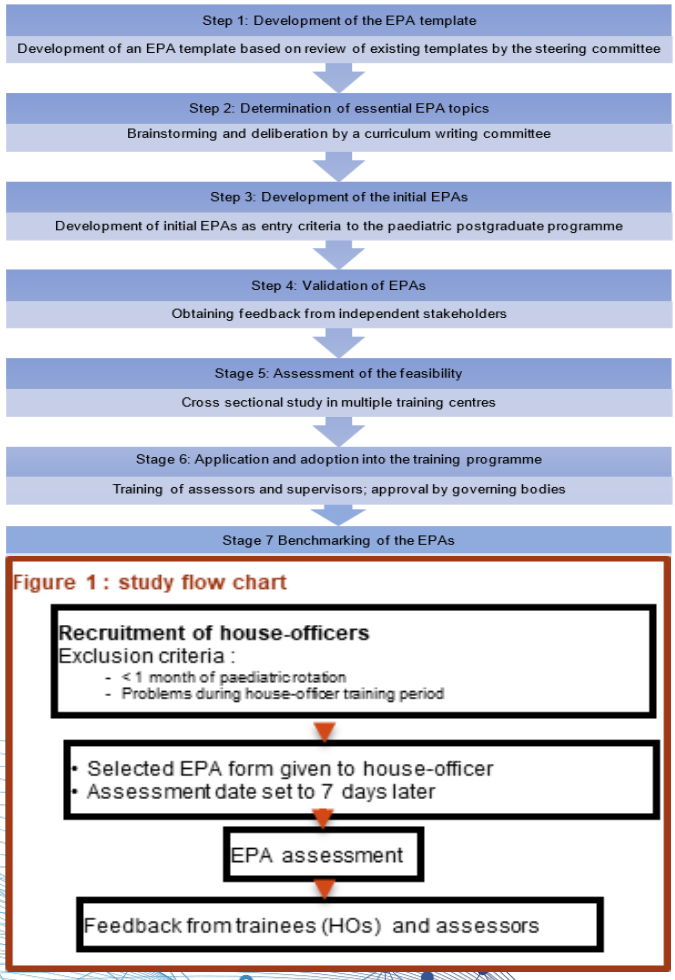

Figure 1 : study flow chart

Table 1. Feedbacks from assessors and candidates.

Total number of candidates	42				
Total number of assessors	21				
Mean time for assessment (minutes)	25.6 (SD 8.5)				
Candidates' feedback (%)	Completely Disagree	Disagree	Neutral	Agree	Completely Agree
a. The items assessed are appropriate for my stage of learning	0	0	0	42.9	57.1
b. I am now confident to perform the task independently	0	2.4	19	64.3	14.3
c. The preparation for this assessment process was difficult	21.4	38.1	14.3	19.0	7.2
Assessors' feedback (%)	Completely Disagree	Disagree	Neutral	Agree	Completely Agree
a. The items are easy to assess	0	7.1	14.3	33.3	45.3
b. The items assessed are relevant	0	0	0	28.6	71.4
c. The items assessed have clear objectives	0	0	11.9	26.2	61.9

Feedback from trainees

- Generally well received by the trainees. All trainees felt the items assessed were appropriate for their stage of learning.
- Comments included difficulties in getting new cases for EPAs that required a case-based discussions within the time given and difficulties in preparing for the EPA alongside their regular duties.

Feedback from assessors

- Majority were comfortable with using EPAs as an assessment tool
- All assessors agreed that the objectives of the EPAs were met at the end of each session.
- The EPAs were conducted with either a patient in the ward in real time, as case-based discussion of a simulated patient or of the trainee's previous encounter with a patient or as role-play.
- A few (7%) reported difficulties in assessing behaviour and skills objectively for sessions that was conducted as a viva or a case-discussion based on a simulated patient.

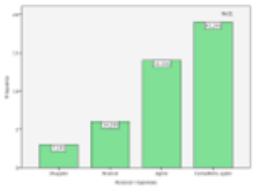

Figure 3 : Percentage of assessors agreeing on the ease of use of the EPAs.

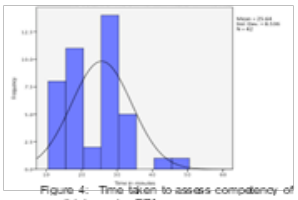

Figure 4: Time taken to assess competency of candidates using EPA

Conclusions

- Using EPAs as an alternative assessment tool for entry into the postgraduate medical training is feasible, acceptable and useful.
- EPAs is a realistic assessment tool in the working place to assess knowledge, skills, attitudes and values which are vital in shaping the professional life and behaviour of a medical specialist.
- A novel assessment tool for professionalism and attitudes in postgraduate trainees and creating new learning space in the workplace
- Cost-saving with potential to be used for genomic medicine training program in low resource settings

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Sitted L to R: Assoc Prof Noorizan A.M. (USM), Dr Noor Khatijah Nurani (Kem. Kesihatan Msia), Prof Haris Van Rostenberghe (USM), **Prof Thong Meow Keong (UM)**.
Standing L to R: Dr Farah Khalid (UM), Dr Wan Jazlah (KKM), Dr Anis Zainal (UiTM), Assoc Prof Tang Swee Fong (UKM), Dr Irene Cheah (UKM), Prof Norlijah Othman, Dr Thong Pui Ling (UPM). Not in pic: Dr Norzila MZ (KKM)

GENETIC COUNSELLING TRAINING IN LOW- AND MIDDLE-INCOME COUNTRIES; PERSPECTIVES FROM HUMAN GENOME ORGANIZATION (HUGO) AND MALAYSIA

Bin Alwi Zilfalil, Alison McEwen, Carolyn Applegate, Surabhi Aryal, Sultana Faradz, Maria Chimpolo, Annie Qurratulain Hasan, Punyaram Kharbuja, Catherine Lynn T. Silao, Milena Paneque, Merlene Peter, Kelly Ormond, Monisha Sebastin, Tilak R. Shrestha, Rapphon Sawaddisan, Thipwimol Tim-Aroon, Yoon Sook Yee, Narazah Mohd Yusuf

Introduction

With the increasing demand for genetic testing and genomic technologies, the necessity for skilled genetic counsellors has become essential, particularly in low- and middle-income countries (LMICs). Genetic counsellors inform, assess risks, and support individuals or families in understanding and adapting to genetic conditions. Worldwide, there are roughly 10,250 genetic counsellors practising in 45 countries, with a noticeably larger workforce found in high-income countries (HICs). Acknowledging this disparity, the Human Genome Organization (HUGO), through its Education Committee and the Genetic/Genomic Counselling subcommittee, has created curriculum guidelines aimed at enhancing global education and training, as standardized guidelines are not currently available worldwide.

Key Insight: Demand for genetics and genomics counselling far outstrips availability in LMICs, highlighting the need for workforce development.

HUGO's Role

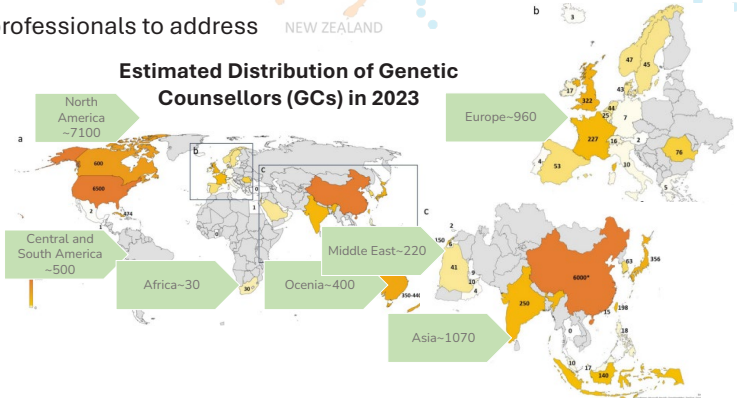
- Established the **HUGO Education Committee and Genetics/Genomics Counselling Subcommittee** to develop curriculum guidelines to standardize genetic counselling training worldwide.
- This subcommittee will also focus on equipping healthcare professionals to address genetic conditions while advancing global health equity.

Training Needs

- **Global Recommendation**: 6-12 genetic counsellors per million people.

Challenges
Limited resources
Lack of professional recognition
Insufficient job opportunities in LMICs

Estimated Distribution of Genetic Counsellors (GCs) in 2023



Malaysia's Initiatives

Educational Program: Master of Medical Science (Genetic Counseling)

- Universiti Kebangsaan Malaysia (UKM) offers the only postgraduate program in genetic counselling in Malaysia.
- Two-year, full-time program with coursework, clinical attachments, and research.

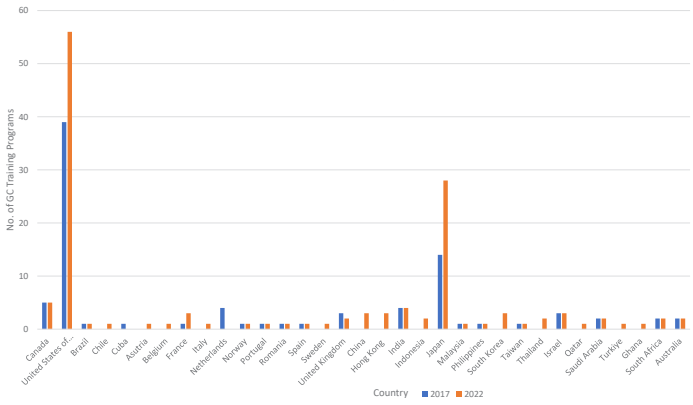
Achievements:

- Since its inception in 2015, the program has produced 15 graduates.

Challenges

- No official government recognition for genetic counsellors.
- Lack of public sector positions, discouraging student enrolment.

Genetic Counsellor Training Programme in HICs vs. LMICs



Future Directions

Recommendations	Goals
Government recognition and policy reform	Strengthen Malaysia's genetic counselling workforce.
Enhanced collaboration with healthcare institutions	Share insights with other LMICs to foster talent development.
Increase training capacity to meet global standards.	

Conclusion

- The curriculum guidelines developed by HUGO Education Committee and Genetics/Genomics Counselling subcommittee will contribute to HUGO's mission for global health equity.
- Foster collaboration and innovation for genetic counselling in LMICs.
- Engage with stakeholders, advocate for policy change, and foster genetic counsellor training collaborations.

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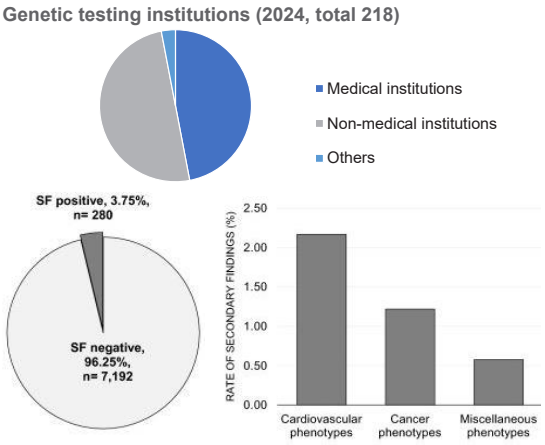
Return of Secondary Genetic Findings: Educational Challenges in South Korea

Hannah Kim (MD, PhD, College of Medicine, Yonsei University, South Korea)

Introduction

- National Health Insurance coverage: NGS-based gene panel testing (2017), whole genome/exome sequencing (WGS/WES) (2024)
- Development of the Guidelines for Reporting Secondary Findings (SF) in Genome sequencing for Koreans (Korean Association of Genetics Diagnostics, 2022 – 2024)
- SF rate: 3.75% among 7,472 participants (Kim *et al*., 2023)
- 62 certified-genetic counsellors in 2023
- This presentation introduces educational issues related to the return of SF in WGS/WES, particularly with a focus on the ethical and legal aspects.



Experts' consultation regarding SF reporting (2023)

Specialty	Clinician: 16	Diagnostics: 30
Board-certification	Genetic diseases: Adults 29; pediatrics 35 Rare genetic diseases: 35 Cancers :15 Cardiovascular: 7 Neurology: 7	
Working experience	-5 yrs: 4 11-20yrs:21	6-10 yrs: 11 20-yrs: 10

- Experience in WES/WGS: 87.2%
- Experience in reporting SF: 66.7%
- Institutions surveyed
  - Genetic counseling clinic (90.2%)
  - Genetic counseling personnel (80.5%)

<Barriers for SF reporting>

- Reimbursement
- Lack of systems in laboratories and clinical practice
- Psychological burden and social stigma
  - Pediatric patients
- Unclear clinical interpretation, making treatment planning difficult
- Limited awareness among clinicians

Issues in patient education in SF reporting

1. WGS and Request of SF

- Informed consent: Priotize interests of patients when obtaining informed consent
- Minors or individuals with limited decision-making capacity: e.g. newborn genome sequencing program
- Pre-testing genetic counselling: Provides support in understanding SF and decision-making during the process
- Re-confirmation: needed re-confirmation for medical doctors for the patient's decision regarding SF rejection
- Genetic discrimination: While it is prohibited by law, the risk remains, particularly in private insurance

2. Return of SF

- Psychological impact: Genetic counseling as a tool to alleviate the impact
- Post-testing genetic counseling: Crucial for minimizing the negative impact of SF and promoting shared decision-making, empowering patients in managing their health.
- Disclosure: No legal duty for clinicians to disclose SF to family members under the Medical Service Act

3. Follow up

- Re-evaluation: After receiving SF, individuals should be informed about future treatment and prevention plans by genetic specialists, including evaluation for family members.
- Quality of life: Recognition of the assessment of health outcomes, including health-related quality of life (QoL) in tracking the intervention process
- Data management as a subject of data donor/owner/user: understanding options, including withdrawal and request deletion of WGS data as needed

Footnotes

Kim Y, Kim JM, Cho HW, Park HY, Park MH. Frequency of actionable secondary findings in 7472 Korean genomes derived from the National Project of Bio Big Data pilot study. Hum Genet. 2023 Nov;142(11):1561-1569.
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*This research was funded by the Korea National Institute for Health (11-1790399-000216-01).