Evidence Brief: Genomics

Contents

Produced by the Knowledge Management team Evidence Briefs offer an overview of the published reports, research, and evidence on a workforce-related topic.

Date of publication: August 2024

Please acknowledge this work in any resulting paper or presentation as:
Evidence Brief: Genomics. Katie Nicholas. (August 2024). UK: Workforce, Training and Education Knowledge Management Team

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- Complete Evidence Brief list – link for Workforce, Training and Education staff
- Complete Evidence Brief list – link for External staff

Key publications – the big picture

Ethnic inequities in Genomics and Precision Medicine
Source: NHS Race & Health Observatory
Publication date: June 2024
In the first report of its kind, the NHS Race and Health Observatory and the University of Nottingham have today published findings which reveal the lack of trust and targeted engagement by health commissioners, regulators and researchers.
See also Lay Summary and Appendices

A new national purpose: harnessing data for health
Source: Tony Blair Institute for Global Change
Publication date: May 2024
We are at a pivotal moment where the convergence of large health and biomedical data sets, artificial intelligence and advances in biotechnology is set to revolutionise health care, drive economic growth and improve the lives of citizens. And the UK has strengths in all three areas. The immense potential of the UK's health-data assets, from the NHS to biobanks and genomics initiatives, can unlock new diagnostics and treatments, deliver better and more personalised care, prevent disease and ultimately help people live longer, healthier lives.

Genomic Medicine strategy 2024 to 2029
Source: Scottish Government
Publication date: April 2024
Whole system workforce and education: We recognise that the knowledge and expertise of our workforce is our greatest asset. As genomic medicine becomes an integral part of the health and social care system we must ensure that we retain this knowledge and expertise to support the changes needed through service redesign. We must develop a fit-for-purpose sustainable genomic workforce for the future which is supported by robust education that also spans the wider healthcare profession to better help them support patients and families.

Pharmacy genomics workforce, education and strategic framework
Source: NHS England
Publication date: January 2024
Genomics will be at centre of the next generation of healthcare and is already being used to guide a personalised approach to medicines optimisation. In the NHS, this is underpinned by the world leading NHS Genomic Medicine Service that was launched in 2018 with the aim of embedding genomic medicine into mainstream care.

Improving access to genomics testing and medicine for cancer: Boosting outcomes and reducing inequalities
Source: Public Policy projects
Publication date: 2024
On 20 September 2023, Public Policy Projects (PPP) held a roundtable entitled Improving access to genomic testing and medicine for better outcomes and reduced inequalities. The roundtable was chaired by Parker Moss, (now Executive Vice President of Corporate Development at Exscientia), and was attended by: clinical oncologists; cancer alliance representatives; genomic laboratory hubs representatives; genomic medicine service alliance representatives; academics and professors in genomic medicine; health data experts; clinical geneticists/genomics leads; and clinical nurse specialists During the roundtable, participants explored the key challenges and opportunities that exist within the genomic testing landscape for cancer care. This report summarises the discussion and includes recent developments in this area such as the ICO Tech Horizon Report published in Feb 2024 and the recent policy paper on Engineering Biology, which is one of the 5 priorities for the UK's Science and Technology Framework and includes genomics.

Fit for the future: a modern and sustainable NHS providing accessible and personalised care for all
Source: Tony Blair Institute for Global Change
Publication date: July 2023
We have the technological and scientific means to transform health and care to safeguard the founding principles of Beveridge and Bevan. Without fully embracing the tech revolution, there is no future for the NHS. The NHS was founded on the principle that health should not be dependent on wealth. Of course, we know poorer people have a harder time living healthy lives; social deprivation begets health deterioration. But the NHS at least guarantees that if you are sick, particularly seriously sick, you're cared for irrespective of your bank balance.

NHS Long Term Workforce Plan
Source: NHS England
Publication date: June 2023
The first comprehensive workforce plan for the NHS, putting staffing on a sustainable footing and improving patient care. It focuses on retaining existing talent and making the best use of new technology alongside the biggest recruitment drive in health service history.
See p. 14 and p. 33 "Developments in science, research, technology, digital and data will continue" and pp. 101-2 "upskilling the workforce"

Topol Review: Progress on the recommendations
Source: Health Education England
Publication date: February 2023
Health Education England (HEE) has published a report into the progress it has made in delivering the recommendations of the Topol Review; four years on. Published in 2019, the Topol Review presented a compelling vision for preparing the healthcare workforce to deliver the digital future, where technology can help to tackle the big healthcare challenges, through enhancing the workforce, driving productivity, and releasing more time to care for patients.

Accelerating genomics medicine in the NHS
Source: NHS England
Publication date: October 2022
A strategy for embedding genomics in the NHS over the next 5 years.

Personalised prescribing: using pharmacogenomics to improve patient outcomes
Source: Royal College of Physicians
Publication date: May 2022
A new report from the Royal College of Physicians and British Pharmacological Society joint working party considers the opportunities provided by increasing pharmacogenomic testing.

Genome UK: shared commitments for UK-wide implementation 2022 to 2025
Source: Department for Science, Innovation and Technology et al.
Publication date: March 2022
The shared commitments set out how the genomics community across the UK will collaborate during 2022 to 2025 to progress the government's vision for genomic healthcare.

Introduction to digital healthcare technologies
Source: HFMA
Publication date: July 2021
Digital technologies such as digital medicine, genomics, artificial intelligence and robotics have a huge potential to transform the delivery of healthcare. These technologies can empower patients to participate actively in their care, with a greater focus on wellbeing and prevention. They also support the prediction of individual disease risk and personalise the management of long-

term conditions. Finance managers have a major role in investment decisions, and therefore need to be aware of how these technologies will transform services in future. This briefing provides NHS finance staff with an introduction to the role of digital in healthcare transformation.

Genome UK: The future of healthcare
Source: HM Government
Publication date: 2020
This strategy sets out a vision for the future focused on three key areas:

1. Diagnosis and personalised medicine: Incorporating the latest genomics advances into routine healthcare to improve the diagnosis, stratification and treatment of illness.
2. Prevention: Enabling predictive and preventative care to improve public health and wellness.
3. Research: Supporting fundamental and translational research and ensuring a seamless interface between research and healthcare delivery.

See also: Genome UK: 2021 to 2022 implementation plan
Healthcare staff must be empowered to know when a patient might benefit from genomic testing and feel confident that they have the skills to communicate complex results to patients and their families in a simple way. In addition to their technical and medical complexity, genomic and genetic test results often raise difficult ethical questions and dilemmas for both clinicians and patients. It is therefore important that clinicians are provided with a developing framework and guidance on how to navigate these issues with their patients, and empower patients to make informed decisions for themselves and their families.
Developing a workforce with these skills will require training and development of existing healthcare staff. We need to ensure that genomics knowledge and expertise is embedded in prospective education and training programmes.

The Topol Review: Preparing the healthcare workforce to deliver the digital future
Source: Health Education England
Publication date: 2019
The roll out of genomics across the NHS will have major implications for the clinical workforce. The pace of change may differ by speciality but, ultimately, genomics will impact on the practice of most, if not all, health professionals. While some aspects of care, such as the management of rare genetic diseases will remain largely within the domain of specialised colleagues, many aspects of genomics, including risk prediction for common diseases and pharmacogenetics, will become ‘mainstream’ and their application embedded in routine healthcare delivery. As practice in genomics evolves, individual workforce training needs will depend less on traditional role demarcations and more on specific responsibilities related to ‘real-world’ implementation based on genomic information.

Genomics Toolkit
Source: Royal College of General Practitioners
The Genomics Toolkit has been developed in partnership with Health Education England Genomics Education Programme to support increasing understanding, raise awareness of Genomics Medicine and support primary care with increased knowledge of how genomics can contribute to improving patient care in a 'genomics era'. It is a collection of resources that explain how Genomics Medicine can be incorporated in Primary Care, including training resources and audit suggestions, and providing links to relevant guidance and patient resources. The resources can be used as quick reference resources or as a package for CPD, to deliver awareness-raising and educational events, and in supporting trainees preparing for the MRCGP.

The Star for workforce redesign

More resources and tools are available by searching for **Genomics** in the Star

National Data Programme

Workforce, Training and Education staff can look at the National Data Warehouse (NDL) SharePoint site to find out more about datasets and Tableau products.

Published Peer Reviewed Research

Advanced Practice

The Advanced Practitioner's Role in the Rapidly Evolving Landscape of Precision Medicine
Item Type: Journal Article
Authors: MOORE, DONALD C. and GUINIGUNDO, ANDREW S.
Publication Date: 2023
Journal: JADPRO: Journal of the Advanced Practitioner in Oncology 14, pp. 39–48
Abstract: The advent of precision medicine targeting oncogenic mutations and other alterations has led to a paradigm shift in the treatment of many solid tumors and hematologic malignancies. For many of these agents, predictive biomarker testing is necessary to determine the presence of such alterations in order to select patients who are most likely to respond, and to avoid the use of ineffective and potentially harmful alternative therapy. Recent technological advancessuch as next-generation sequencing have facilitated the identification of targetable biomarkers in patients with cancer and thus help inform treatment decisions. Moreover, new molecular-guided therapies and associated predictive biomarkers continue to be discovered. For some cancer therapeutics, regulatory approval requires the use of a companion diagnostic to ensure proper patient selection. Advanced practitioners therefore need to be aware of current biomarker testing guidelines regarding who should be tested, how and when to test, and how these results can guide treatment decisions using molecular-based therapies. They should also recognize and address potential barriers and disparities in biomarker testing to ensure equitable care for all patients, and assist in educating patients and colleagues alike on the importance of testing and integration into clinical practice to enhance outcomes. The advent of precision medicine targeting oncogenic mutations and other alterations has led to a paradigm shift in the treatment of many solid tumors and hematologic malignancies. For many of these agents, predictive biomarker testing is necessary to determine the presence of such alterations in order to select patients who are most likely to respond, and to avoid the use of ineffective and potentially harmful alternative therapy. Recent technological advances such as next-generation sequencing have facilitated the identification of targetable biomarkers in patients with cancer and thus help inform treatment decisions. Moreover, new molecular-guided therapies and associated predictive biomarkers continue to be discovered. For some cancer therapeutics, regulatory approval requires the use of a companion diagnostic to ensure proper patient selection. Advanced practitioners therefore need to be aware of current biomarker testing guidelines regarding who should be tested, how and when to test, and how these results can guide treatment decisions using molecular-based therapies. They should also recognize and address potential barriers and disparities in biomarker testing to ensure equitable care for all patients, and assist in educating patients and colleagues alike on the importance of testing and integration into clinical practice to enhance outcomes.

Competencies

Update to the essential genomic nursing competencies and outcome indicators
Item Type: Journal Article
Authors: Calzone, Kathleen A.;Stokes, Liz;Peterson, Cheryl and Badzek, Laurie
Publication Date: 2024
Journal: Journal of Nursing Scholarship : An Official Publication of Sigma Theta Tau International Honor Society of Nursing
Abstract: INTRODUCTION: Genomic healthcare applications have relevance to all healthcare professionals including nursing, and most evidence-based clinical applications impact the quality and safety of healthcare. To guide nursing genomic competency initiatives, the Essential Nursing Competencies and Curricula Guidelines for Genetics and Genomics were established through a process of consensus in 2005. A 2009 update incorporated outcome indicators consisting of specific areas of knowledge and clinical performance indicators, to help support academic integration. Almost 20 years have elapsed since these competencies were first established, yet incorporating the competencies into general and specialty scope and standards of nursing practice is inconsistent, competency integration into curricula is highly uneven, continuing education in genomics for nurses is limited, and the genomic capacity of the nursing workforce remains low. These deficits have persisted despite substantial advances in genomic technology which substantially reduced costs and increased evidence-based clinical applications, including direct to consumer genomic tests, the integration of genomics into evidence-based guidelines, and evidence that genomics impacts the quality and safety of healthcare., DESIGN: The aim of this project was to update and achieve consensus on genomic competencies applicable to all registered nurses. This was a mixed methods study., METHODS: The update to the competencies was performed based first on a literature review to update the competencies based on the current state of the evidence. Using the updated content, a modified Delphi study was conducted with registered nurse panelists from clinical, academic, and research settings. Once consensus was achieved, the competencies were made available through the American Nurses Association for public comment. Public comments were then reviewed and integrated as needed., RESULTS: The literature review resulted in a transition from genetics to genomics, given the reduction in costs, which resulted in an expansion of the scope of testing in both the germline and somatic contexts. Two Delphi rounds were required to reach consensus prior to the public comment period. Public comments were solicited through the American Nurses Association, and each comment was reviewed by the authors and addressed as indicated., CONCLUSION: The Essentials of Genomic Nursing: Competencies and Outcome Indicators constitute the minimum competency in genomics required of all registered nurses regardless of the level of academic training, role, or specialty., CLINICAL RELEVANCE: Evidence-based genomic applications span the entire healthcare continuum and, therefore, are relevant for all registered nurses regardless of academic training, role, practice setting, or clinical expertise. These competencies serve as the guide for the minimum requirements for registered nurse practice as well as guide curricula and continuing education for all registered nurses, including but not limited to administrators, educators, nursing leaders, practicing nurses, and researchers. Copyright Published 2024. This article is a U.S. Government work and is in the public domain in the USA. Journal of Nursing Scholarship published by Wiley Periodicals LLC on behalf of Sigma Theta Tau International.

Competencies of the UK nursing and midwifery workforce to mainstream genomics in the National Health Service: the ongoing gap between perceived importance and confidence in genomics
Author(s): Carpenter-Clawson et al.
Source: Frontiers in Genetics 14
Publication date: 2023
The United Kingdom is recognised worldwide as a leader in genomics. The use of genomic technologies in the National Health Service (NHS) is expected to deliver faster and more accurate diagnoses, supporting personalized treatments to improve patient outcomes. The ambition of embedding genomic medicine in the diagnostic pathway requires involvement of the front-line clinical workforce, known as ‘mainstreaming’. Nurses and midwives are the largest professionally qualified workforce in the National Health Service thus, it is anticipated that they will play key roles in mainstreaming. This study investigated the level of competence/confidence of practicing nurses and midwives to support mainstreaming and their perception of the importance of genomics in delivery of patient care. A literature review of genetics/genomics competency frameworks, semi structured interviews of lead nurses and stakeholders were conducted to identify relevant competencies needed for mainstreaming. These were then used to survey four cohorts of nurses (*n* = 153) across England in four consecutive years (2019–22). The confidence level of these professionals in all aspects of genomics was 2.07 ± 0.47 measured on a 5-point Likert scale (1“*Low confidence*”; 5 “*High confidence*”). Intriguingly, these professionals all appreciated the importance of genomics for their patient care (4.01 ± 0.06). Whilst the importance scores increased, the confidence scores declined at the time when major genomic transformation took place in the NHS (e.g.: launch of the Genomic Medicine Service, the National Genomic Test Directory). To bridge this gap, relevant genomic education can play key roles. However, nurses and midwives were found to be grossly underrepresented in formal genomic education courses offered by Health Education England Genomics Education Programme since 2014. This may result from the lack of direct applicability of the currently offered courses for their practice and role. Thematic analysis revealed that nurses and midwives wish to support their patients by providing more information on their condition, inheritance, and treatment options in combination with the use of relevant genetic counselling skills. This study identified easy to follow competencies for embedding genomics into routine clinical care. We propose a training programme that addresses the gap that nurses and midwives currently have, to enable them to harness genomic opportunities for patients and services.

Competencies needed by the frontline clinical workforce for genomic mainstreaming Accessible to RCM MIDIRS subscription members
Item Type: Journal Article
Authors: Carpenter-Clawson, C.;Watson, M.;Lightfoot, A.;Barnes, D.;Abbas, W.;Pope, A.;Lynch, K.;Miles, T.;Bell, D.;Talbot, M.;Pichini, A.;Kirwan, D. and Varadi, A.
Publication Date: 2023
Journal: MIDIRS Midwifery Digest 33(4), pp. 326–332
Abstract: Background: Genomics is the study of a person's genetic material and how this influences the growth, development and working of the body. The UK is recognised worldwide as a leader in genomics, particularly after the recent completion of the groundbreaking 100,000 Genomes Project. The ambition is now to embed genomic medicine in the routine diagnostic pathway across the NHS, which requires involvement of the frontline clinical workforce, known as 'genomic mainstreaming'. Aim(s): This study investigated the level of competence/confidence of practising nurses and midwives, the largest professionally qualified workforce in the NHS, to support genomic mainstreaming. Method(s): A literature review of

genetics/genomics competency frameworks, and semi-structured interviews of lead nurses and stakeholders, were conducted to identify relevant competencies needed for genomic mainstreaming. These were then used to survey four cohorts of nurses and midwives (n=154) across England in four consecutive years (2019-2022). Result(s): The confidence level of these professionals in all aspects of genomics was 2.07+/-0.47 measured on a five-point Likert scale (1 'Low confidence'; 5 'High confidence'). Intriguingly, these professionals all appreciated the importance of genomics for their patient care (4.01+/-0.06). While the importance scores increased, the confidence scores declined at the time when major genomic transformation took place in the NHS, for example, the launch of the Genomic Medicine Service (NHS GMS) and the National Genomic Test Directory (NGTD) in October 2018. To bridge this gap, relevant genomic education can play key roles. However, nurses and midwives were found to be grossly under-represented in formal genomic education courses offered by the Health Education England Genomics Education Programme (HEE GEP) since 2014. This may result from the lack of direct applicability of the currently offered courses for their practice and role. We co-created with our stakeholders and students a postgraduate genomic training programme that successfully addressed the gap that nurses and midwives currently have by increasing their confidence from scale 2-4 within a 20-25 week period. Conclusion(s): This study identified easy-to-follow competencies for embedding genomics into routine clinical care. We demonstrated that our postgraduate course enables professionals with limited prior genomic knowledge to harness genomic opportunities for their patients and services. Copyright © MIDIRS 2023.

Equality, Diversity and Inclusion

Stark exclusion of ethnic minorities in genetic research must change, say experts
Author(s): Matthew Limb
Source: BMJ 385
Publication date: 2024
A new review has laid out a blueprint for tackling the poor representation of ethnic minority communities in genetic medicine research. Matthew Limb reports. Recently launched trials of a “personalised” cancer vaccine that uses mRNA technology to activate a patient’s immune response were not just a boon for the future of genomics and precision medicines but, at first glance, for diversity in UK science.12

Equity, diversity, and inclusion at the Global Alliance for Genomics and Health
Item Type: Journal Article
Authors: Skantharajah, Neerjah;Baichoo, Shakuntala;Boughtwood, Tiffany F.;Casas-Silva, Esmeralda;Chandrasekharan, Subhashini;Dave, Sanjay M.;Fakhro, Khalid A.;Falcon de Vargas, Aida,B.;Gayle, Sylvia S.;Gupta, Vivek K.;Hendricks-Sturrup, Rachele;Hobb, Ashley E.;Li, Stephanie;Llamas, Bastien;Lopez-Correa, Catalina;Machirori, Mavis;Melendez-Zajgla, Jorge;Millner, Mareike A.;Page, Angela J. H.;Paglione, Laura D., et al
Publication Date: 2023
Journal: Cell Genomics 3(10), pp. 100386
Abstract: A lack of diversity in genomics for health continues to hinder equitable leadership and access to precision medicine approaches for underrepresented populations. To avoid perpetuating biases within the genomics workforce and genomic data collection practices, equity, diversity, and inclusion (EDI) must be addressed. This paper documents the journey taken by the Global Alliance for Genomics and Health (a genomics-based

standard-setting and policy-framing organization) to create a more equitable, diverse, and inclusive environment for its standards and members. Initial steps include the creation of two groups: the Equity, Diversity, and Inclusion Advisory Group and the Regulatory and Ethics Diversity Group. Following a framework that we call "Reflected in our Teams, Reflected in our Standards," both groups address EDI at different stages in their policy development process. Copyright © 2023 The Author(s).

Improving Diversity of the Genomics Workforce: Proceedings of a Workshop-in Brief
Item Type: Proceedings from a Workshop
Publication Date: 2022
Abstract: In 2020, as part of its strategic planning initiative, the Roundtable on Genomics and Precision Health of the National Academies of Sciences, Engineering, and Medicine decided to explore four action-oriented focus areas for its activities over the coming three to five years: driving innovation, shaping the policy dialogue, spurring the adoption of tools and approaches, and achieving equity. The roundtable began its exploration of the fourth thematic area - achieving equity in genomics and precision health - by holding a public workshop on October 5, 2021. The workshop, Improving Diversity of the Genomics Workforce, examined the current state of diversity of the genetics and genomics workforce; the structural and social factors associated with the lack of workforce diversity; the impact of diversity in the workforce on access to genetic services and patient outcomes; and possible actions that could lead to a more diverse, equitable, and inclusive genomics workforce. This Proceedings of a Workshop-in Brief highlights the presentations and discussions that occurred during the workshop. Copyright 2022 by the National Academy of Sciences. All rights reserved.

Beyond inclusion: Enacting team equity in precision medicine research
Item Type: Journal Article
Authors: Jeske, Melanie;Vasquez, Emily;Fullerton, Stephanie M.;Saperstein, Aliya;Bentz, Michael;Foti, Nicole;Shim, Janet K. and Lee, Sandra Soo-Jin
Publication Date: 2022
Journal: PloS One 17(2), pp. e0263750
Abstract: PURPOSE: To identify meanings of and challenges to enacting equitable diversification of genomics research, and specifically precision medicine research (PMR), teams., METHODS: We conducted in-depth interviews with 102 individuals involved in three U.S.-based precision medicine research consortia and conducted over 400 observation hours of their working group meetings, consortium-wide meetings, and conference presentations. We also reviewed published reports on genomic workforce diversity (WFD), particularly those relevant to the PMR community., RESULTS: Our study finds that many PMR teams encounter challenges as they strive to achieve equitable diversification on scientific teams. Interviewees articulated that underrepresented team members were often hired to increase the study's capacity to recruit diverse research participants, but are limited to on-the-ground staff positions with little influence over study design. We find existing hierarchies and power structures in the academic research ecosystem compound challenges for equitable diversification., CONCLUSION: Our results suggest that meaningful diversification of PMR teams will only be possible when team equity is prioritized as a core value in academic research communities.

Improving racial diversity in the genomics workforce: An examination of challenges and opportunities
Item Type: Journal Article
Authors: Wicklund, Catherine;Sanghavi, Kunal;Coleman, Bernice;Johansen Taber, Katherine;Taylor, Jacquelyn Y.;Asalone, Kathryn C. and Beachy, Sarah
Publication Date: 2022
Journal: Genetics in Medicine : Official Journal of the American College of Medical Genetics 24(8), pp. 1640–1643
Introduction: The Roundtable on Genomics and Precision Health at the National Academies of Sciences, Engineering, and Medicine spent much of its meeting time in 2020 envisioning a new strategic plan to guide its work over the next few years.[1] The Roundtable's new vision, *realizing the full potential of health for all through genomics and precision health*, underscores the members' prioritization of the values of inclusivity and equity *for all*— a theme that emerged and recurred during strategic thinking discussions. As an effective convener of diverse stakeholders in genomics, the Roundtable fosters discussions, collaborations, and new ideas to shape the field with the longer-term goal of achieving equitable access to the benefits of innovations in genomics. Over the past year, the Roundtable has focused on underrepresentation and inequity in genomics research, workforce, and patient health care. The Roundtable has undertaken efforts such as a public workshop[2] and Roundtable discussions on workforce diversity to better understand issues the genomics field faces, including its history of racism. This commentary reflects key points the individual authors wished to highlight from the Roundtable workshop and discussions on workforce diversity, as well as from personal experiences.

The genomics workforce must become more diverse: a strategic imperative
Item Type: Journal Article
Authors: Bonham, Vence L. and Green, Eric D.
Publication Date: 2021
Journal: American Journal of Human Genetics 108(1), pp. 3–7
Abstract: The National Human Genome Research Institute (NHGRI) recently published a new strategic vision for the future of human genomics, the product of an extensive, multi-year engagement with numerous research, medical, educational, and public communities. The theme of this 2020 vision-The Forefront of Genomics-reflects NHGRI's critical role in providing responsible stewardship of the field of human genomics, especially as genomic methods and approaches become increasingly disseminated throughout biomedicine. Embracing that role, the new NHGRI strategic vision features a set of guiding principles and values that provide an ethical and moral framework for the field. One principle emphasizes the need to champion a diverse genomics workforce because "the promise of genomics cannot be fully achieved without attracting, developing, and retaining a diverse workforce, which includes individuals from groups that are currently underrepresented in the genomics enterprise." To build on the remarkable metamorphosis of the field over the last three decades, enhancing the diversity of the genomics workforce must be embraced as an urgent priority. Toward that end, NHGRI recently developed an "action agenda" for training, employing, and retaining a genomics workforce that reflects the diversity of the US population.

Healthcare Science

Evolving Careers in Genetics: Attitudes of Healthcare Scientists
Abstract all available [Abstract P19.28D]
Author(s):A J Clarkson
Source: European Journal of Human Genetics
Publication date: 2019
Studying the specialist workforce in the Genomics Service, particularly in Cytogenetics and Molecular Genetics, offers potential insights into the nature and impact of the changes on their occupational boundaries. Both disciplines have evolved as separate occupations with their own professional bodies in the UK. Recently, the distinction between them has become increasingly 'blurred' through the advancement of shared technology; the need to decrease service costs and find efficiencies; and the Modernising Scientific Careers policy programme which merges two previously separate scientist education and training programmes. As a result, the two specialities are being merged and the services rationalised. This is leading to changes in healthcare science careers and scientists' future working roles and relationships in this clinical domain.

Infrastructure and Implementation

Infrastructuring precision medicine: Making gene therapies for rare diseases workable in practice Abstract only*
Item Type: Journal Article
Authors: Wadmann, Sarah;Johansen, Anna Brueckner;Born, Alfred Peter and Kessel, Line
Publication Date: 2024
Journal: Social Science & Medicine 351, pp. N.PAG
Abstract: Long viewed by social scientists as a future imaginary, precision medicine is now materializing in many healthcare systems in the form of new diagnostic practices and novel treatment modalities, such as gene therapies. Based on an ethnographic study of the introduction of the first two clinically available in-vivo gene therapies in the Danish healthcare system, we investigate what it takes to make these therapies workable in practice. Drawing on social science literature on infrastructuring, we describe the many forms of mundane work required to fit these therapies into regulatory frameworks, political processes and daily work practices in the healthcare system. Further, we observe how the processes of infrastructuring required to introduce the gene therapies into clinical practice had transformative implications as they redistributed roles and responsibilities among clinicians, pharmacists, procurement agencies and pharmaceutical manufacturers. • The effects of gene therapies depend on their translation into clinical practice. • Securing facilities and staff to handle GMO constitute major challenges. • Local innovations in work practices are made to overcome regulatory challenges. • Such innovations can have transformative effects on healthcare systems.

"A very big challenge": a qualitative study to explore the early barriers and enablers to implementing a national genomic medicine service in England
Item Type: Journal Article
Authors: Friedrich, B.;Vindrola-Padros, C.;Lucassen, A. M.;Patch, C.;Clarke, A.;Lakhanpaul, M. and Lewis, C.
Publication Date: 2023
Journal: Frontiers in Genetics 14, pp. 1282034
Abstract: Background: The Genomic Medicine Service (GMS) was launched in 2018 in England to create a step-change in the use of genomics in the NHS, including offering whole genome sequencing (WGS) as part of routine care. In this qualitative study on pediatric rare disease diagnosis, we used an implementation science framework to identify enablers and barriers which have influenced rollout. Method(s): Semi-structured interviews were conducted with seven participants

tasked with designing the GMS and 14 tasked with leading the implementation across the seven Genomic Medicine Service Alliances (GMSAs) and/or Genomic Laboratory Hubs (GLHs) between October 2021 and February 2022. Result(s): Overall, those involved in delivering the service strongly support its aims and ambitions. Challenges include: 1) concerns around the lack of trained and available workforce (clinicians and scientists) to seek consent from patients, interpret findings and communicate results; 2) the lack of a digital, coordinated infrastructure in place to support and standardize delivery with knock-on effects including onerous administrative aspects required to consent patients and order WGS tests; 3) that the "mainstreaming agenda", whilst considered important, encountered reluctance to become engaged from those who did not see it as a priority or viewed it as being politically rather than clinically driven; 4) the timelines and targets set for the GMS were perceived by some as too ambitious. Interviewees discussed local adaptations and strategies employed to address the various challenges they had encountered, including 1) capacity-building, 2) employing genomic associates and other support staff to support the consent and test ordering process, 3) having "genomic champions" embedded in mainstream services to impart knowledge and best practice, 4) enhancing collaboration between genetic and mainstream specialties, 5) building evaluation into the service and 6) co-creating services with patients and the public. Conclusion(s): Our findings highlight the challenges of implementing system-wide change within a complex healthcare system. Local as well as national solutions can undoubtedly address many of these barriers over time.

Real-World Implementation of Precision Psychiatry: A Systematic Review of Barriers and Facilitators
Item Type: Journal Article
Authors: Baldwin, Helen;Loebel-Davidsohn, Lion;Oliver, Dominic;Salazar de Pablo, Gonzalo;Stahl, Daniel;Riper, Heleen and Fusar-Poli, Paolo
Publication Date: 2022
Journal: Brain Sciences 12(7)
Abstract: BACKGROUND: Despite significant research progress surrounding precision medicine in psychiatry, there has been little tangible impact upon real-world clinical care., OBJECTIVE: To identify barriers and facilitators affecting the real-world implementation of precision psychiatry., METHOD: A PRISMA-compliant systematic literature search of primary research studies, conducted in the Web of Science, Cochrane Central Register of Controlled Trials, PsycINFO and OpenGrey databases. We included a qualitative data synthesis structured according to the 'Consolidated Framework for Implementation Research' (CFIR) key constructs., RESULTS: Of 93,886 records screened, 28 studies were suitable for inclusion. The included studies reported 38 barriers and facilitators attributed to the CFIR constructs. Commonly reported barriers included: potential psychological harm to the service user (n = 11), cost and time investments (n = 9), potential economic and occupational harm to the service user (n = 8), poor accuracy and utility of the model (n = 8), and poor perceived competence in precision medicine amongst staff (n = 7). The most highly reported facilitator was the availability of adequate competence and skills training for staff (n = 7)., CONCLUSIONS: Psychiatry faces widespread challenges in the implementation of precision medicine methods. Innovative solutions are required at the level of the individual and the wider system to fulfil the translational gap and impact real-world care.

Genomics education for medical professionals – the current UK landscape
Author(s): Slade et al.
Source: Clinical Medicine 16(4)
Publication date: 2016
Genomic science, incorporated into the national healthcare landscape, offers the potential of improved patient care and disease prevention that can be fully exploited only by an educated health professional workforce. Cooperation and coordination will be required both between the multiple strategic and delivery players outlined in this review, as well as with the biomedical research sector, to effectively nurture such a workforce. Furthermore, any national educational strategy will need both persistence and substantial sustained resource commitment.

Leadership

The leadership behaviors needed to implement clinical genomics at scale: a qualitative study Full text available with NHS OpenAthens account*
Author(s): Best et al.
Source: Genetics in Medicine 22(8)
Publication date: 2020
Entrepreneurial leadership is needed to promote innovativeness, risk-taking, and proactivity, essential in these early stages of clinical genomics. Shared decision-making is required from a wide range of clinicians, calling for both clinical and distributed leadership. Sharing leadership, and the potential loss of positional status from formal senior positions, may prove challenging to genomics “migrants,” who are essential for nurturing genomic “natives.” Clinicians will need support from their organizations and professional bodies to manage the transition.

Nursing

Genomic medicine: the role of the nursing workforce Abstract only*
tem Type: Journal Article
Authors: Buaki-Sogo, Maria and Percival, Natalie
Publication Date: 2022
Journal: Nursing Times 118(8), pp. 36–38
Abstract: Genomic medicine can improve patient care through supporting quicker diagnoses and enabling more tailored care. In England, genomic testing has already been introduced for patients with some rare conditions and cancers, and, as understanding of the genome increases, it is likely to become part of more care pathways. This article discusses the key roles nurses may play in bringing the benefits of genomics to patients as it becomes part of routine care, including in identifying patients for genomic testing, discussing testing with patients, referring to genetic counselling services or the relevant clinicians for targeted therapies, and personalising care plans. We also explain how the NHS Genomic Medicine Service Alliances will work with nurses to help bring genomics into mainstream healthcare.

Canadian nursing and genomics: An engagement initiative
Item Type: Journal Article
Authors: Carlsson, Lindsay and Limoges, Jacqueline
Publication Date: 2022
Journal: Canadian Oncology Nursing Journal 32(4), pp. 559–570
Abstract: Advances in genome sequencing technologies and biomarker discoveries now inform individual risk assessments and treatment decision-making within cancer care. These advances have contributed to the emergence of precision healthcare where disease prevention and treatment recommendations are based upon individual genetic variability, as well as environmental and lifestyle considerations. To actively

participate in precision healthcare and support patients, oncology nurses require specific knowledge and skills in cancer genomics. Nurses are poised to contribute to the safe and equitable delivery of precision healthcare and further education and engagement will support this endeavour. The Canadian Nursing and Genomics (CNG) promotes collaboration between nurses from the five domains of practice, from different healthcare sectors and educational backgrounds, as a key strategy to prepare nurses for the genomics era. This article provides a case study to illustrate genomics informed nursing practice across clinical settings and provides leadership strategies across the domains of nursing practice to support genomic literacy within nursing practice.

Leading Canadian Nurses into the Genomic Era of Healthcare
Item Type: Journal Article
Authors: Limoges, J.;Pike, A.;Dewell, S.;Meyer, A.;Puddester, R. and Carlsson, L.
Publication Date: 2022
Journal: Nursing Leadership (Toronto, Ont.) 35(2), pp. 79–95
Abstract: Genomics is having a profound impact on every aspect of healthcare. To support nurses to develop genomic literacy and integrate genomics into care, an engagement framework was created. The framework uses principles of nursing intraprofessional collaboration, the knowledge-to-action cycle and the diffusion of innovations theory. This framework was used to identify six key priorities for action and leadership strategies to accelerate and sustain the nurses' engagement with genomics. With leadership and genomic literacy, nurses can fully participate in the creation and implementation of new care pathways, deliver education, advance research linked to genomics and improve patient experience and health outcomes. Copyright © 2022 Longwoods Publishing.

Essential genomic knowledge in graduate nursing practice
Abstract only*
Item Type: Journal Article
Authors: Connors, Laurie M.;Schirle, Lori and Dietrich, Mary S.
Publication Date: 2022
Journal: Journal of the American Association of Nurse Practitioners 34(9), pp. 1050–1057
Abstract: BACKGROUND: Genetics-informed nursing is essential to personalized health care. Advanced practice nurses will increasingly encounter genomic information in clinical care and are expected to have competency., PURPOSE: To examine genomic competency of advanced practice nursing students and faculty in a graduate nursing school., METHODS: A convenience sample of graduate nursing faculty and students were electronically sent a survey assessing genomic knowledge. In total, 13.98% of faculty (33/236) and 9.87% of students (82/831) completed a demographic questionnaire, perceived genomic competency items, and Genomic Nursing Concept Inventory (GNCI). The GNCI is a 31-question multiple choice questionnaire assessing 18 genomic concepts in four categories (Human Genome Basics, Mutations, Inheritance Patterns, and Genomic Health care). Percentage of correct items was calculated for faculty and students, as were correlations between demographics, perceived genomic competency, and GNCI scores., RESULTS: Students' GNCI overall scores were higher than faculty, 54.8% (interquartile range IQR] 38-72%) vs 48.4% (IQR 32-68%). Both groups demonstrated the lowest scores in Genomic Basics (students 41.7% IQR 25-67%] and faculty 33.3% IQR 16-50%]). Students' and faculty's perceived genomic competency correlated with GNCI scores (r = 0.49, p < .001 and r = 0.70, p < .001, respectively). Age (r = -0.40, p < .001), entering nursing school after 2010 (r = 0.47, p < .001), and previous genomics course (r = 0.52, p < .001) were significantly correlated with GNCI total score for students, but not faculty., CONCLUSIONS: This study indicates that faculty and students

in a graduate nursing school perceive and demonstrate low genomic knowledge, particularly in basic genomics. Further exploration into innovative methods to provide basic genomic education is needed., IMPLICATIONS: To provide access and equity for personalized genomic-based health care, we must prepare genomics-informed nurses. Copyright © 2022 American Association of Nurse Practitioners.

Current status and future directions of U.S. genomic nursing health care policy
Author(s): Kurnat-Thoma et al.
Source: Nursing Outlook 69(3)
Publication date: 2021
Without necessary genomic health care quality performance measurement tools for clinicians, purchasers, regulators and policymakers; an adequately prepared nursing workforce; frameworks, planning, and know-how for rapid cycle performance improvement/quality improvement; it will be difficult to move precision health beyond research trials at academic medical centers. Similar to other performance measurement health care policy 'calls to action', we opened the dialogue to defining a precision health care quality measurement strategy that is evidence based, harmonized, and multi-stakeholder (Lamb & Donaldson, 2011; National Quality Forum, 2019). Strategic integration of health care quality performance measurement principles and federal benchmarking infrastructure including development of genomic outcome measures and nurse contributions to genomic health care will facilitate broader expansion of precision health into routine health care operations.

A Maturity Matrix for Nurse Leaders to Facilitate and Benchmark Progress in Genomic Healthcare Policy, Infrastructure, Education, and Delivery
Author(s): Tonkin et al.
Source: Journal of Nursing Scholarship 52(5) pp. 583-592
Publication date: 2020
The essential elements that need to be in place for nurses to be able to deliver effective care that integrates genomics into standard practice are explicit within ASIGN. The tool provides the basic framework to guide further development in genomics, including guiding academic and continuing education. By using the matrix as a framework for assessment, groups, organizations, or countries can use ASIGN to benchmark themselves at a starting point to inform a plan for progress. The tool is nimble, as users can be a country or region within a country, or an organization such as a hospital, professional body, or government. Focusing on outcomes, users of ASIGN can capture both the current status and change over time. Comparisons between indicators and stage of maturity (i.e., high vs. low maturity) can help users identify areas where work needs to be focused. Individuals can identify the most appropriate measures (evidence) for each indicator, thus offering some flexibility, in recognition that there will be wide variations in resources, infrastructure, and service provision across countries, regions, or organizations.

A Roadmap for Global Acceleration of Genomics Integration Across Nursing
Author(s): Tonkin et al.
Source: Journal of Nursing Scholarship 52(3) pp. 329-338
Publication date: 2020
The model espouses five broad and synergistic principles. The first principle advocates that integration of context (in this case genomics) should also place emphasis on understanding the broader social and contextual influences. The second principle advocates for inclusivity, particularly for marginalized, minority, and vulnerable populations. Complementary to this, the third principle focuses on ensuring accessible diffusion of innovations, and understanding the barriers and facilitators for this. Gaining

stakeholder perspectives embeds communication as a dialog and further supports inclusivity. The fourth principle focuses on effective utilization of communication technologies to promote efficient sharing of information, inclusivity, and health equity. The fifth principle emphasizes the need to promote specialized training and continuing education that also encompasses cultural and linguistic competence. This set of broad guidelines can be used by teams to consider how their implementation plans address the issues of equity and inclusivity.

Exploring the genomic education needs within the NHS workforce: a cardiac genetic nurse perspective Abstract all available [Abstract, p. 777]
Author(s): Braddel and Watson
Source: European Journal of Human Genetics
Publication date: 2020
This study aimed to explore the education needs and support required for CGNs to fulfil their role. A key theme identified was the skills associated with managing the uncertainty of genetic information. The interviews highlighted that effective learning requires immersion in the clinical environment and the need for a CGN-specific competency framework to be developed. The study demonstrates the need to define the competencies required for the role of a CGN, to standardise practice nationally and ensure that nursing and genetic counselling resources are applied effectively.

Genomic Literacy of Registered Nurses and Midwives in Australia: A Cross-Sectional Survey 2019, Journal of Nursing Scholarship *Athens log-in required**
This survey measured the genomic literacy of Australian registered nurses and midwives. The findings indicate that the genomic literacy of Australian registered nurses and midwives is low, a finding similar to that reported in international studies where nurses' knowledge of genomics is largely reported to be poor or, at best, moderate. Poor performance across the GNCI© indicates that Australian registered nurses and midwives are not achieving the competencies outlined in the Essentials of Genetic and Genomic Nursing: Competencies, Curricula Guidelines and Outcome Indicators document (Consensus Panel on Genetic/Genomic Nursing Competencies, 2009). Since these competencies represent the minimum standards required to deliver competent genetic and genomic-focused care, it can be inferred that Australian nurses and midwives are not prepared to deliver such care. Australian nursing and midwifery authorities must act if they are to equip their registered nurses and midwives with the genomic literacy required to deliver genomic healthcare.

Measuring physician practice and preparedness for genomic medicine: a national survey [Abstract, p. 776]
Author(s): King et al.
Source: European Journal of Human Genetics
Publication date: 2020
T wo-thirds of respondents felt genomics will soon impact their practice and, while a third recently completed CGE, only a quarter felt prepared. Respondents lacked confidence in genomic knowledge and ability to explain concepts to patients and incorporate test results in patient management. These data can inform genetic services (re test referral patterns) and support genomic education providers to best meet learner needs and develop a competent, genomic literate workforce.

Hospital Nursing Leadership Led Interventions Increased Genomic Awareness and Educational Intent in Magnet® Settings
Author(s): Calzone et al.
Source: Nursing Outlook
Publication date: 2018
The Precision Medicine Initiative is poised to accelerate genomic discoveries relevant to practice. Assuring the genomic

awareness of nurses in the workforce is an essential step to realizing the benefits of genomic discoveries on the public's health. Longer term interventions are required for successful practice integration. This necessitates an ongoing investment in leadership education, infrastructure, and policy development to enable genomic adoption enhancing healthcare safety and quality while reducing costs. Results provide policy makers and healthcare leaders a mechanism applicable to the interprofessional healthcare community for capacity building and integration of genomics to improve health outcomes.

Expanding RN Scope of Knowledge—Genetics/Genomics: The New Frontier
Author(s): Rogers et al.
Source: Journal for Nurses in Professional Development 33(2)
Publication date: 2017
A knowledge survey was used to assess nurses' knowledge following a yearlong genomics education initiative. Findings indicate that nurses benefit from repeated exposure to genetics-related content. Recommendations from this study include development and implementation of strategies that can be used to prepare nurses at all levels for the application of genetics and genomics. Clinical nurses with knowledge of genetics will be able to implement evidence-based interventions to manage acute and chronic illnesses. These nurses will then be able to engage patients more fully, thereby helping them to understand the relationship of genetics to healthy outcomes.

A strategy for implementing genomics into nursing practice informed by three behaviour change theories
Author(s): Leach et al.
Source: International Journal of Nursing Practice 22(3)
Publication date: 2016
The core philosophy of the strategy is that genomic nurse Adopters and Opinion Leaders who have direct interaction with their peers in practice will be best placed to highlight the importance of genomics within the nursing role. The strategy discussed in this paper provides scope for continued nursing education and development of genomics within nursing practice on a larger scale. The recommendations might be of particular relevance for senior staff and management.

Using clinical genomics in health care: Strategies to create a prepared workforce Abstract only*
Author(s): Williams and Cashion
Source: Nursing Outlook
Publication date: 2015
Benefits from genomic discoveries hinge in part on a prepared and knowledgeable HCP workforce who can access and apply these discoveries for individuals, families, and communities when the information is needed in clinical care. However, current graduate and continuing education is not meeting this need for HCPs across all health care settings.

Ophthalmology

Ophthalmic Services Guidance: Genomics Services
Author(s):
Source: Royal College of Ophthalmologists
Publication date: 2020
A long-term objective will be to work with the Training Committee, in conjunction with other Royal Colleges, to develop an appropriate curriculum to reflect on-going changes to clinical practice that result from the development of mainstream genomic medicine. While the move towards adoption of genomic medicine is imminent, it is likely that the speed of development of skills will differ between subspecialty groups, and objectives will be different for subgroups of clinical ophthalmology.

Pathology

Pathology workforce and training in genomic medicine
Source: Royal College of Pathologists
Publication date: 2021
This article briefly outlines the establishment of clinical whole genome testing in NHS England and describes the subsequent curriculum in development to train and upskill the pathology workforce. Although the model by which training needs will be addressed remains unclear, what is certain is that equipping the pathology workforce to integrate modern genomic analysis with traditional morphology is essential for the enhancement of patient care.

Pharmacy

Pharmacy and Genomic Medicine: A UK-wide survey of pharmacy staff assessing their prior education, confidence and educational needs
Author(s): Wickens et al.
Source: International Journal of Pharmacy Practice 31(Supplement 2)
Publication date: 2023
Introduction: Pharmacy teams are key in helping patients to get the most from genomic medicine.[1,2] However, genomics has only recently been included in undergraduate curricula, and it has been suggested that all healthcare professionals could benefit from education in pharmacogenomics[2]. We surveyed pharmacy staff to gather information on previous education, current practice and future educational needs in genomics and pharmacogenomics. Aim: This survey aimed to establish existing levels of education and confidence in genomics and pharmacogenomics in pharmacy staff working in any role, in any sector, across the UK, and to investigate respondents' preferences in delivery of genomic education. Methods: The survey was based on a 2021 survey of genomic knowledge among medical staff by Health Education England (HEE)[3], and amended to reflect pharmacy roles and practice following discussion with pharmacy leads from the 7 NHS Genomic Medicine Service Alliances in England, and from Scotland, Wales and Northern Ireland. SmartSurvey software was used to host the survey, with data held securely. The survey was open between 1st March and 16th May 2022, and was publicised via pharmacy groups including the Royal Pharmaceutical Society, National Pharmacy Association, Local Pharmaceutical Committees, chief pharmacists networks in primary and secondary care, and social media. This work was assessed using the NHS Health Research Authority Research screening tool and judged as 'not research'; therefore ethical approval was not required. Results: 1,552 responses were received from pharmacists, pharmacy technicians, dispensers and other pharmacy staff across the UK; 68% of responses were from England, 13% from Scotland, 10% from Northern Ireland and 9% from Wales. The majority of responses (69%) were from Pharmacists, with 24% from Pharmacy Technicians and 4% from Pharmacy support workers. Only 13% of respondents had received any formal training in genomics. Most respondents felt unprepared to use genomic testing in their practice; just 8% of pharmacists (including trainees), and 1% of pharmacy technicians (including trainees) felt prepared. However, 65% of respondents thought that genomics would change their practice within the next 5 years, and over 70% of pharmacists, and 56% of pharmacy technicians, could envisage ordering, advising on, or counselling patients on genomic testing in the future after

appropriate training. 29% of respondents (mainly pharmacy managers) did not currently see patients and therefore might not train personally in genomics. Discussion/Conclusion: This work suggests that pharmacy teams are likely to require educational support to embrace the opportunities of genomic medicine. High survey engagement suggested that respondents were keen to make their voices heard. Pharmacists appeared more confident in their ability to advise patients on genomics than Technicians, however both groups seemed keen to receive training. One limitation is that respondents were likely interested in genomics; those with no interest may not have completed the survey. Additionally, pharmacy managers who do not see patients might not train personally in genomics, but may influence strategy for pharmacy genomics service development and delivery. National bodies should capitalise on enthusiasm across the sector to help drive pharmacy genomics services forward through education and training.

A systematic review of pharmacogenetic testing in primary care: Attitudes of patients, general practitioners, and pharmacists
August 2022, Research in Social and Administrative Pharmacy

- The majority of pharmacists had an interest in implementing pharmacogenetic services. More training and education for pharmacists is needed.
- Pharmacists were aware that they need more knowledge about PGx, the solution could be an education or training with a team-based approach, and to have access to clinical resources and experts in PGx.
- There is a lack of guidelines for using PGx results in prescribing. Pharmacists and GPs agreed that education of healthcare personnel and collaboration is needed.

Pharmacists Leading the Way to Precision Medicine: Updates to the Core Pharmacist Competencies in Genomics
Author(s): Roseann et al.
Source: American Journal of Pharmaceutical Education 86(4)
Publication date: April 2022
Clearly defined pharmacist competencies in genomics that are refined as genomic medicine matures are essential to advance pharmacy education and practice in pharmacogenomics and precision medicine. Pharmacists are the health care professionals best suited to lead collaborative, interprofessional teams in the provision of pharmacogenomics-based care across practice settings and therapeutic areas. The competencies described in this report serve as a blueprint for clinical pharmacogenomics instruction as part of pharmacy school curricula and continuing education programs for practicing pharmacists. When implemented into pharmacy school curricula, these competencies will ensure that graduates are “practice-ready” to integrate pharmacogenomics into patient care. Additional postgraduate training is needed for advanced roles in pharmacogenomics implementation, education, and research.

Practice, Education and Training

Results from the Delivery of a Community Health Worker Training to Advance Competencies in Cancer Genomics
Item Type: Journal Article
Authors: Allen, Caitlin G.;Hatch, Ashley;Hill, Elizabeth;Qanungo, Suparna;Ford, Marvella;Price, Sarah Tucker and Umemba, LaQuisha
Publication Date: 2024
Journal: Public Health Genomics 27(1), pp. 83–95
Abstract: INTRODUCTION: Less than half of eligible Black women are assessed for genetic risk and only 28% engage in recommended hereditary breast and ovarian cancer (HBOC) risk-reducing interventions. CHWs are trusted individuals that

work as a liaison between health systems and the community to improve access to services and support cancer prevention efforts, though they are an overlooked resource to support genetic risk assessment. To address the need and training gaps for CHWs, we developed and assessed an online training program to build CHW's competencies in cancer genomics and use of health information technologies to navigate high-risk individuals to appropriate genetic services., METHODS: The curriculum and 10 training modules were developed through engaging a panel of experts in a three-round Delphi process. Recruitment focused on CHWs who worked in clinical settings or groups providing outreach or health services to Black women. We assessed: changes in knowledge and attitudes about HBOC and genomics, as well as the perceptions about the quality and implementation of the training., RESULTS: Forty-six individuals expressed interest in the training after recruitment. Thirty eight individuals were eligible for the training and 26 completed the course. We found improvements in knowledge and genomics competencies immediately post-course, but the majority of these improvements were not sustained at 3-month follow-up. The training was highly rated for its relevance to CHW work and overall delivery. Top rated sessions included HBOC overview and family history collection. On average, participants reported discussing HBOC with 17 individuals at 3-month follow-up., CONCLUSION: Championing a diverse cancer and genomics workforce can help address the goals of the National Cancer Plan to improve early detection and health equity. Through this training, CHWs gained critical cancer and genomics knowledge that was then applied to their primary roles. Copyright © 2024 The Author(s). Published by S. Karger AG, Basel.

Compassion and equity-focused clinical genomics training for health professional learners
Item Type: Journal Article
Authors: Berninger, T. J.;Rajagopalan, R. M. and Bloss, C. S.
Publication Date: 2024
Journal: Journal of Genetic Counseling
Abstract: There remains an urgent need for expanded genomics training in undergraduate medical education, especially as genetic and genomic assessments become increasingly important in primary care and routine clinical practice across specialties. Physician trainees continue to report feeling poorly prepared to provide effective consultation or interpretation of genomic test results. Here we report on the development, pilot implementation, and evaluation of an elective offering for pre-clinical medical students called the Sanford Precision Health Scholars Immersive Learning Experience (PHS), which was designed leveraging genetic counseling expertise as one means to address this need. This 9-week course, piloted in Fall 2021 at UC San Diego, afforded students the opportunity to build technical skills and competencies in clinical genomics while identifying, addressing, and engaging with pervasive health disparities in genomics. Interactive exercises focused students' learning on strategies for empathic and compassionate patient interactions while supporting the application of concepts and knowledge to future practice. Upon completion of the course, participants reported increases in confidence related to skills required for clinical genomics practice. Drawing on learnings from this pilot implementation, recommendations for refining the program include deepening pedagogical engagement with ethical issues, expanding the offering to trainees across health professions, including pharmacy students, and incorporating an optional experiential learning component. Educational offerings, like PHS, that are designed with the input of genetic counseling expertise may ease pressures on the genetic counseling profession by building a more genomic-literate healthcare

workforce that can better support efforts to expand access for patients. Copyright © 2024 The Authors. Journal of Genetic Counseling published by Wiley Periodicals LLC on behalf of National Society of Genetic Counselors.

Survey of the training needs of genetic assistants supports the creation of genetic assistant training programs Abstract only*
Item Type: Journal Article
Authors: Rider, R. A.;Cubano, L.;Madden, E. B.;Rowley, R. K. and Manolio, T.
Publication Date: 2024
Journal: Journal of Genetic Counseling 33(4), pp. 733–744
Abstract: Despite the increasing numbers of genetic assistants (GAs) in the genomics workforce, their training needs and how to best prepare GAs for their role have not been well defined. We sought to identify the current educational status of GAs, opinions on their training needs, and attitudes about GA training programs (GATPs). Survey links were emailed to NSGC members, 17 state genetic counseling (GC) professional organizations, and genomic medicine researchers. Respondents (n = 411) included GCs (n = 231) and GAs (n = 136). Like other studies, we found that the GA position is filled by a range of education levels and career aspirations. Most respondents supported the creation of GATPs, with 63% endorsing that GATPs would be helpful and half endorsing a short-term (3 months or less) program. Most believed GATPs should focus on general knowledge, with almost all practical skills learned on-the-job. If more GATPs are created, our survey provides evidence that graduates would be hired. Indeed, of those whose work setting required a bachelor's degree, the number of respondents who favored keeping that requirement was similar to the number who favored hiring a GA without a degree if they attended a GATP. However, there were concerns about GATPs. Many (44%) believed creating GATPs could discourage candidates from becoming GAs. We observed that there are two types of GAs: entry-level and bachelor's-level, with the entry-level being those who do not have and are not working to obtain a bachelor's degree and the bachelor's-level being those who do/are. GATPs could focus on the education of entry-level GAs, while gaps in the knowledge base of bachelor's-level GAs could be addressed by augmenting bachelor's curriculum or providing additional training after hire. Further research on the training needs of GAs and hiring practices of institutions will be vital to understanding their training needs and designing and implementing effective GATPs. Copyright Published 2023. This article is a U.S. Government work and is in the public domain in the USA.

Ensuring best practice in genomics education: A theory- and empirically informed evaluation framework
Author(s): Nisselle et al.
Source: American Journal of Human Genetics 111(8)
Publication date: August 2024
Implementation of genomic medicine into healthcare requires a workforce educated through effective educational approaches. However, ascertaining the impact of genomics education activities or resources is limited by a lack of evaluation and inconsistent descriptions in the literature. We aim to support those developing genomics education to consider how best to capture evaluation data that demonstrate program outcomes and effectiveness within scope. Here, we present an evaluation framework that is adaptable to multiple settings for use by genomics educators with or without education or evaluation backgrounds. The framework was developed as part of a broader program supporting genomic research translation coordinated by the Australian Genomics consortium. We detail our mixed-methods approach involving an expert workshop, literature review and iterative expert input to reach consensus and synthesis of a new evaluation framework for genomics education. The resulting theory-informed and evidence-based framework encompasses evaluation across all

stages of education program development, implementation and reporting, and acknowledges the critical role of stakeholders and the effects of external influences.

We need a genomics-savvy healthcare workforce
Item Type: Journal Article
Publication Date: 2023
Journal: Nature Medicine 29(8), pp. 1877–1878
Twenty years after completion of the Human Genome Project, genetics is rapidly being integrated into everyday clinical practice. But in this era of genomic revolution, genetically trained teams of healthcare workers are needed to optimize delivery of patient care.

Evaluation of two Massive Open Online Courses (MOOCs) in genomic variant interpretation for the NHS workforce
Item Type: Journal Article
Authors: Coad, Beth;Joekes, Katherine;Rudnicka, Alicja;Frost, Amy;Openshaw, Mark Robert;Tatton-Brown, Katrina and Snape, Katie
Publication Date: 2023
Journal: BMC Medical Education 23(1), pp. 540
Abstract: BACKGROUND: The implementation of the National Genomic Medicine Service in the UK has increased patient access to germline genomic testing. Increased testing leads to more genetic diagnoses but does result in the identification of genomic variants of uncertain significance (VUS). The rigorous process of interpreting these variants requires multi-disciplinary, highly trained healthcare professionals (HCPs). To meet this training need, we designed two Massive Open Online Courses (MOOCs) for HCPs involved in germline genomic testing pathways: Fundamental Principles (FP) and Inherited Cancer Susceptibility (ICS)., METHODS: An evaluation cohort of HCPs involved in genomic testing were recruited, with additional data also available from anonymous self-registered learners to both MOOCs. Pre- and post-course surveys and in-course quizzes were used to assess learner satisfaction, confidence and knowledge gained in variant interpretation. In addition, granular feedback was collected on the complexity of the MOOCs to iteratively improve the resources., RESULTS: A cohort of 92 genomics HCPs, including clinical scientists, and non-genomics clinicians (clinicians working in specialties outside of genomics) participated in the evaluation cohort. Between baseline and follow-up, total confidence scores improved by 38% (15.2/40.0) (95% confidence interval CI] 12.4-18.0) for the FP MOOC and 54% (18.9/34.9) (95%CI 15.5-22.5) for the ICS MOOC ($p < 0.0001$ for both). Of those who completed the knowledge assessment through six summative variant classification quizzes (V1-6), a mean of 79% of respondents classified the variants such that correct clinical management would be undertaken (FP: V1 (73/90) 81% Likely Pathogenic/Pathogenic LP/P]; V2 (55/78) 70% VUS; V3 (59/75) 79% LP/P; V4 (62/72) 86% LP/LP. ICS: V5 (66/91) 73% VUS; V6 (76/88) 86% LP/P). A non-statistically significant higher attrition rate was seen amongst the non-genomics workforce when compared to genomics specialists for both courses. More participants from the non-genomics workforce rated the material as "Too Complex" (FP n = 2/7 29%], ICS n = 1/5 20%]) when compared to the specialist genomics workforce (FP n = 1/43 2%], ICS n = 0/35 0%])., CONCLUSIONS: After completing one or both MOOCs, self-reported confidence in genomic variant interpretation significantly increased, and most respondents could correctly classify variants such that appropriate clinical management would be instigated. Genomics HCPs reported higher satisfaction with the level of content than the non-genomics clinicians. The MOOCs provided foundational knowledge and improved learner confidence, but should be adapted for different workforces to maximise the benefit for clinicians working in specialties outside of genetics.

Genomics education for advance practice nurses: Staying cutting edge Abstract only*
Item Type: Journal Article
Authors: Connors, L. M. and Wysocki, K.
Publication Date: 2023
Journal: Journal of the American Association of Nurse Practitioners 35(12), pp. 784–786
Abstract: As we celebrate the 20th anniversary of sequencing of the human genome and the rapid integration of genetics in health care, we pause to reflect on the status of genomic competency in nursing. The literature provides evidence that nurses do not feel prepared or confident in genomics. Genomic education for nurses and other health care professionals can support access and equity in the integration of genomics into practice. Resources are provided to support genomic education and clinical support. Copyright © 2023 Lippincott Williams and Wilkins. All rights reserved.

Understanding the application of genomics knowledge in nursing and midwifery practice: A scoping study Abstract only*
Item Type: Journal Article
Authors: Schluter, J. E.
Publication Date: 2023
Journal: Collegian 30(2), pp. 306–314
Abstract: Background: With the increased realisation of the benefits of genomic testing, nurses and midwives are being exposed to genomic care as a part of normal clinical practice. Aim(s): To explore how Queensland nurses and midwives are applying genomics knowledge in clinical practice to understand how best to support the workforce to meet patient needs in response to increased genomic testing rates. Method(s): A scoping methodology was used whereby the research question was defined, relevant studies were identified for the purposes of a literature review, followed by interviews with 32 nurses and midwives to support the interpretation of the literature review and to understand the implications for practice. Finding(s): Nurses and midwives are working in partnership with their patients and families to support genomic decision making. The emerging needs of patients to understand their diagnostic and treatment pathway is forcing nurses and midwives to self-educate to keep pace with current practice demands. This approach to upskilling is not adequate for those nurses and midwives currently who are regularly exposed to patients requiring genomic support. Discussion(s): Despite national and local policy documents identifying genomics workforce capacity as a strategic priority action and clinicians reporting their involvement in genomics care, there is a lack of succession planning, organisational support and educational opportunities to support these advances in practice. Conclusion(s): There is a need to address the emerging genomic workforce and education requirements to ensure nurses and midwives are capable of supporting patients undergoing genomic testing. Copyright © 2022

A Web Screening on Training Initiatives in Cancer Genomics for Healthcare Professionals
Item Type: Journal Article
Authors: Hoxhaj, Ilda;Beccia, Flavia;Calabro, Giovanna Elisa and Boccia, Stefania
Publication Date: 2022
Journal: Genes 13(3)
Abstract: The disruptive advances in genomics contributed to achieve higher levels of precision in the diagnosis and treatment of cancer. This scientific advance entails the need for greater literacy for all healthcare professionals. Our study summarizes the training initiatives conducted worldwide in cancer genomics field for healthcare professionals. We conducted a web search of the training initiatives aimed at improving healthcare professionals' literacy in cancer genomics undertaken worldwide by using two search engines (Google and Bing) in English language and conducted from 2003 to 2021. A total of 85,649

initiatives were identified. After the screening process, 36 items were included. The majority of training programs were organized in the United States (47%) and in the United Kingdom (28%). Most of the initiatives were conducted in the last five years (83%) by universities (30%) and as web-based modalities (80%). In front of the technological advances in genomics, education in cancer genomics remains fundamental. Our results may contribute to provide an update on the development of educational programs to build a skilled and appropriately trained genomics health workforce in the future.

Genetic and genomic learning needs of oncologists and oncology nurses in the era of precision medicine: a scoping review Abstract only*
Item Type: Journal Article
Authors: Rahman, Belinda;McEwen, Alison;Phillips, Jane L.;Tucker, Katherine;Goldstein, David and Jacobs, Chris
Publication Date: 2022
Journal: Personalized Medicine 19(2), pp. 139–153
Abstract: Genetic and genomic data are increasingly guiding clinical care for cancer patients. To meet the growing demand for precision medicine, patient-facing oncology staff will be a part of leading the provision of genomic testing. A scoping review was undertaken to identify the range of genetic and genomic learning needs of oncologists and oncology nurses. Learning needs were reported relating to interpretation of genomic data, clinical decision-making, patient communication and counseling, and fundamentals of genetics and genomics. There was a lack of empirical research specific to oncology nurses and their learning needs in tumor sequencing. Our findings suggest that oncologists and oncology nurses need tailored support, education and training to improve their confidence and skills in adopting genomic testing into clinical practice.

An international genomics health workforce education priorities assessment
Author(s): Johnson et al.
Source: Personalised Medicine 19(4)
Publication date: 2022
Aim: Global implementation of genomic medicine will require education of healthcare providers. There are limited international needs assessment data to guide curriculum development. Materials & methods: Genomics education experts developed and distributed a survey to individuals with knowledge of country-specific needs: 113 completed surveys (19% response rate) from 34 countries. A high percentage of respondents ranked non genetics physicians as the #1 target for genetics education. Over 70% indicated a need for moderate/extensive modification in physician training. The majority considered germline and somatic topics and targeting primary care and specialist providers equally important. Conclusion: Regardless of country economic level, there is a clear need for genomics education of healthcare providers. The study results can be used to focus future genomic medicine education efforts.

Preparing the genetic counseling workforce for the future in Australasia Abstract only*
Item Type: Journal Article
Authors: McEwen, Alison and Jacobs, Chris
Publication Date: 2021
Journal: Journal of Genetic Counseling 30(1), pp. 55–60
Abstract: Current genetic counseling students will graduate into a workforce involving more opportunities, diversity, and uncertainty than any previous generation. Preparing the future genetic counseling workforce is a dynamic challenge, both for the profession and for educators. The dominance of the medical model in the state funded Australian healthcare system creates a power imbalance between doctors and other health

professionals. As a result, professional regulation to protect the public from harm in line with the United States, the UK, and Canada only became mandatory in 2019. Professional regulation has the additional benefit of enhancing professional standing and autonomy, enabling genetic counselors to help shape the future of genetic health care in Australia and New Zealand. Within this rapidly evolving environment, we are establishing a new Masters' program and building a discipline of genetic counseling, working alongside other allied health professionals. Our program involves synchronous and asynchronous learning, greater accessibility, flexibility and, as we have learned in 2020, reduction in disruption during a global pandemic. In this program, we foreground the inherent knowledge, skills, and values of genetic counseling, shifting the focus from provision of genetic and genomic tests, to educating competent, person-centered, research enabled and culturally safe genetic counselors. As educators, we have a responsibility to prepare students to embrace the uncertainties, challenges, and potential of the genomic era, to seize the many possibilities that lie ahead, and to expand their thinking and vision. We ask our students to be courageous, to step into a deep exploration of their own identity, beliefs, understanding, and experiences of oppression, power, and privilege. We are pushing boundaries, and challenging ourselves and our students to remain always open to possibilities. Equipping students with open eyes and listening ears may be the single most important thing we can do to prepare the genetic counseling workforce of the future to provide the best possible care.

Ensuring best practice in genomics education and evaluation: reporting item standards for education and its evaluation in genomics (RISE2 Genomics)
Item Type: Journal Article
Authors: Nisselle, Amy;Janinski, Monika;Martyn, Melissa;McClaren, Belinda;Kaunein, Nadia;Barlow-Stewart, Kristine;Belcher, Andrea;Bernat, John A.;Best, Stephanie;Bishop, Michelle;Carroll, June C.;Cornel, Martina;Dissanayake, Vajira H. W.;Dodds, Agnes;Dunlop, Kate;Garg, Gunjan;Gear, Russell;Graves, Debra;Knight, Ken;Korf, Bruce, et al
Publication Date: 2021
Journal: Genetics in Medicine : Official Journal of the American College of Medical Genetics 23(7), pp. 1356–1365
Abstract: PURPOSE: Widespread, quality genomics education for health professionals is required to create a competent genomic workforce. A lack of standards for reporting genomics education and evaluation limits the evidence base for replication and comparison. We therefore undertook a consensus process to develop a recommended minimum set of information to support consistent reporting of design, development, delivery, and evaluation of genomics education interventions., METHODS: Draft standards were derived from literature (25 items from 21 publications). Thirty-six international experts were purposively recruited for three rounds of a modified Delphi process to reach consensus on relevance, clarity, comprehensiveness, utility, and design., RESULTS: The final standards include 18 items relating to development and delivery of genomics education interventions, 12 relating to evaluation, and 1 on stakeholder engagement., CONCLUSION: These Reporting Item Standards for Education and its Evaluation in Genomics (RISE2 Genomics) are intended to be widely applicable across settings and health professions. Their use by those involved in reporting genomics education interventions and evaluation, as well as adoption by journals and policy makers as the expected standard, will support greater transparency, consistency, and comprehensiveness of reporting. Consequently, the genomics education evidence base will be more robust, enabling high-quality education and evaluation across diverse settings.

Measuring physician practice, preparedness and preferences for genomic medicine: a national survey
Item Type: Journal Article
Authors: Nisselle, Amy;King, Emily A.;McClaren, Belinda;Janinski, Monika;Metcalfe, Sylvia and Gaff, Clara
Publication Date: 2021
Journal: BMJ Open 11(7), pp. e044408
Abstract: OBJECTIVE: Even as genomic medicine is implemented globally, there remains a lack of rigorous, national assessments of physicians' current genomic practice and continuing genomics education needs. The aim of this study was to address this gap., DESIGN: A cross-sectional survey, informed by qualitative data and behaviour change theory, to assess the current landscape of Australian physicians' genomic medicine practice, perceptions of proximity and individual preparedness, and preferred models of practice and continuing education. The survey was advertised nationally through 10 medical colleges, 24 societies, 62 hospitals, social media, professional networks and snowballing., RESULTS: 409 medical specialists across Australia responded, representing 30 specialties (majority paediatricians, 20%), from mainly public hospitals (70%) in metropolitan areas (75%). Half (53%) had contacted their local genetics services and half (54%) had ordered or referred for a gene panel or exome/genome sequencing test in the last year. Two-thirds (67%) think genomics will soon impact their practice, with a significant preference for models that involved genetics services ($p<0.0001$). Currently, respondents mainly perform tasks associated with pretest family history taking and counselling, but more respondents expect to perform tasks at all stages of testing in the future, including tasks related to the test itself, and reporting results. While one-third (34%) recently completed education in genomics, only a quarter (25%) felt prepared to practise. Specialists would like (more) education, particularly on genomic technologies and clinical utility, and prefer this to be through varied educational strategies., CONCLUSIONS: This survey provides data from a breadth of physician specialties that can inform models of genetic service delivery and genomics education. The findings support education providers designing and delivering education that best meet learner needs to build a competent, genomic-literate workforce. Further analyses are underway to characterise early adopters of genomic medicine to inform strategies to increase engagement.

Genomics Education for Medical Specialists – A Blended Learning Approach [Abstract, p. 68]
Author(s): Maher et al.
Source: Twin Research and Human Genetics 24 pp. 49-88
Publication date: 2021
We developed four self-directed, interactive online modules, e-book and infographic that introduced foundational genetics content and clinically-relevant concepts, processes and skills for germline and somatic genomics. Workshops (pediatric, adult and somatic/cancer) then focussed on clinical cases presented by clinicians with genomics expertise and small group discussions. Participants completed evaluation surveys at baseline, post-online modules and post workshops. Participants rated online modules helpful for learning more foundational and pragmatic concepts, and facilitated workshops more helpful for learning about clinical aspects such as identifying the right test for the right patient, interpreting reports and genetic counseling, yet the major gains in knowledge and confidence were seen after online modules.

Teamwork in clinical genomics: A dynamic sociotechnical healthcare setting Abstract only*
Author(s): Best et al.
Source: Journal of Evaluation in Clinical Practice 27(6)
Publication date: 2021
Challenges to teamworking that arise in the elongated adaptive context do not always fit traditional ways of working, and innovative strategies will need to be adopted to ensure the diagnostic advances of clinical genomics are realised. Provision of time and permission for team members to share knowledge and evolve, promoting capacity building, nurturing trustful relationships and establishing boundaries are amongst the practice recommendations for organisational and team leaders, even though these activities may disrupt existing ways of working or hierarchical structures.

Social and behavioral science at the forefront of genomics: Discovery, translation, and health equity
Author(s): Koehley et al.
Source: Social Science & Medicine 271
Publication date: February 2021
This special issue highlights the unique role that social and behavioral science has to play at the forefront of genomics. Through the introduction of papers comprising this special issue, we outline priority research areas at the nexus of genomics and the social and behavioral sciences. These include: Discovery science; clinical and community translation, and equity, including engagement and inclusion of diverse populations in genomic science. We advocate for genomic discovery that considers social context, neural, cognitive, and behavioral endophenotypes, and that is grounded in social and behavioral science research and theory. Further, the social and behavioral sciences should play a leadership role in identifying best practices for effective clinical and community translation of genomic discoveries. Finally, inclusive research that engages diverse populations is necessary for genomic discovery and translation to benefit all. We also highlight ways that genomics can be a fruitful testbed for the development and refinement of social and behavioral science theory. Indeed, an expanded ecological lens that runs from genomes to society will be required to fully understand human behavior.

A family systems approach to genetic counseling: Development of narrative interventions
Author(s): MacLeod et al.
Source: Journal of Genetic Counselling 30 pp. 22-29
Publication date: 2021
To what extent are family systems approaches relevant in the genomics era? What difference does it make to remember the wider social context within which ‘problems’ associated with a genetic diagnosis reside? How does this influence the conversations we have with our patients? These questions will be considered in relation to systemic approaches to genetic counseling practice. Narrative therapy with an emphasis on people's strengths, wishes, and ways of resisting the effects of a problem may be a particularly useful framework for genetic counselors. Narrative practice views people as multi-storied and is concerned with the question of how we encourage people to tell their stories in ways that make them feel stronger. Increased uptake of genomic testing and the number of people seeking genetic counseling present opportunities to consider new ways of working, particularly around support following a new genetic diagnosis. One option is to realize the potential of group interventions. Family therapy and narrative practices have the potential to encourage communication and for families to learn from each other.

Development of a novel, online training platform for genomics education in cancer care; preparing healthcare professionals for "Generation Genome" [Abstract, p. 511]
Author(s): Tripathi et al.
Source: European Journal of Human Genetics 28 pp. 141-797
Publication date: 2020
We developed an online platform (Nucleus) to address this need across for cancer healthcare professionals in our region. Modules in basics of cancer genomics, cancer genomics susceptibility, and precision oncology (technological and clinical applications) have been developed. Podcasts and blogs are currently in development. Initial user testing has been very positive with users rating the platform as relevant to practice, with appropriate length and pace.

Preparing Medical Specialists for Genomic Medicine: Continuing Education Should Include Opportunities for Experiential Learning
Author(s): McClaren et al.
Source: Frontiers in Genetics 11:151
Publication date: 2020
Our findings show that motivations to engage with continuing education about genomic medicine appear to be driven by a combination of: individual characteristics (interest in genomics, career stage, and medical specialty); perceptions of relevance to practice (current and future); and prior experience, such as that gained in research settings. We have shown that medical specialists contextualize their knowledge gained through formal education by engaging with their peers and seeking out opportunities for experiential learning. In fact, participants described how most genomics learning occurs outside of attendance at continuing education activities, which have been the previous focus of workforce development (Burton, 2011; Talwar et al., 2017).

From helices to health: undergraduate medical education in genetics and genomics Full text available with NHS OpenAthens account*
Author(s): Hyland et al.
Source: Personalized Medicine
Publication date: 2019
Integration of genetics and genomics into the clinical years can help students learn genomic medicine skills, and requires genomically literate teaching faculty. Genomic champions in different clinical specialties can be recruited to teach medical students and promote genomic literacy among their peers.

Ensuring Best Practice in Genomic Education and Evaluation: A Program Logic Approach
Author(s): Nisselle et al.
Source: Frontiers in Genetics 10
Publication date: 2019
The program logic developed in this paper is a versatile and useful tool for developing education interventions in different settings. Despite a "call to action" over a decade ago (Gaff et al., 2007), few papers published since have described use of program logic in their design or evaluation. This program logic model can be used to inform program development and redesign; it is not intended to be linear, but as with all program logic models, can be used through cycles, with the outputs and outcomes informing inputs and activities at different stages. As not all education providers will be familiar with a program logic model approach to developing interventions, we are developing a set of companion documents to support the use of the tool, including a "how to" guide, a glossary of terms, useful resources for both education and evaluation, and detailed definitions and examples throughout.

Readiness of clinical genetic healthcare professionals to provide genomic medicine: An Australian census
Author(s): Nisselle et al.
Source: Journal of Genetic Counseling 28(2)
Publication date: 2019
This is the first national audit of clinical genetic healthcare professionals, revealing the Australian workforce is motivated and prepared to embrace new models to deliver genomic medicine but consideration of education and training is required to meet demand. The majority of respondents (89.9%) were satisfied with their job and 91.6% planned to work in genetics until retirement. However, 14.1% of the genetic counselors in clinical roles and 24.6% of the clinical geneticists planned to retire within 10 years.

Current conditions in medical genetics practice Full text available with NHS OpenAthens account*
Author(s): Maiese et al.
Source: Genetics in Medicine 21(8)
Publication date: 2019
New genetic discoveries are creating new opportunities in prevention, health maintenance, and the management of heritable disorders. To fully realize the potential of these medical breakthroughs and to ensure that all Americans have access to genetic services will take new practice models, new residency training models, and better reimbursement. Long-term investments are also needed to encourage students to join the genetics profession, so that ten years from now patients and families affected by genetic conditions are more quickly diagnosed and more easily able to access the primary, secondary, and tertiary care services they need. Genetics professional capacity within the health-care system will need to be substantially expanded and wait times for nonemergency appointments greatly reduced.

Delivering genomic medicine in the United Kingdom National Health Service: a systematic review and narrative synthesis
Author(s): Pearce et al.
Source: Genetics in Medicine 21(12)
Publication date: 2019
Mainstreaming genomic medicine into routine clinical practice requires actions at each level of the health-care system. Our synthesis emphasized the organizational, social, and cultural implications of reforming practice, highlighting that demonstration of clinical utility and cost-effectiveness, attending to the compatibility of genomic medicine with clinical principles, and involving and engaging patients are key to successful implementation.

Interdisciplinary learning on a postgraduate genomic medicine course – help or hindrance? Abstract all available
Author(s): Alice Garrett
Source: Archives of Disease in Childhood
Publication date: 2019
Participants reported being affected in different ways and to different extents by social, individual and course factors related to interdisciplinary learning. These mirror the behavioural, personal and environmental determinants of Bandura's social cognitive theory. Several advantages and disadvantages identified by participants show parallels with adult learning theory and the interprofessional education literature. These results may be used to inform strategies for optimising the experience of participants on future interdisciplinary learning courses.

Genomics in Clinical Care: Preparing Non-genetic Health Professionals Abstract all available [Abstract, p. 411]
Author(s): Lynch et al.
Source: Twin Research and Human Genetics
Publication date: 2019

Melbourne Genomics has an upskilling strategy to meet education needs of practising non-genetic medical professionals: (1) internships; (2) blended learning short courses in clinical genomics; (3) workshops.

Driving the precision medicine highway: community health workers and patient navigators
Author(s): Ramos et al.
Source: Journal of Translational Medicine 17(85)
Publication date: 2019
Low levels of health and genomic literacy render the American public ill-equipped to make informed decisions, use and interpret genomic information, or appreciate the benefits afforded by genomics-based technologies. We propose that coordinated expansion of the roles of community health workers and patient navigators within the precision medicine space can be effectively used to disseminate the knowledge required for the public to benefit from precision medicine advances in healthcare. A well-organized and trained community health worker and patient navigator workforce will provide a voice for the disadvantaged, especially among recent immigrants likely to be experiencing social isolation, language barriers, and economic deprivation. Armed with this knowledge, community health workers and patient navigators can advance the precision medicine agenda and empower disadvantaged communities to take advantage of major advances in the precision medicine era.

Building a learning community of Australian clinical genomics: a social network study of the Australian Genomic Health Alliance
Author(s): Long et al.
Source: BMC Medicine 17(44)
Publication date: 2019
Social processes and self-directed modes of learning were shown to be powerful influences on members' genomic practice, underlining the significance of the strategy of building relationships to form a genomic learning community.

The 2019 US medical genetics workforce: a focus on clinical genetics
Author(s): Jenkins et al.
Source: Genetics in Medicine 23 pp. 1458-1464
Publication date: 2019,
To address the current shortfall and expected capacity needs of genomic medicine, a substantial increase in clinical genetics trainees will be necessary. However, there have been persistent deficiencies in filling training slots. Potential solutions, such as increasing the recruitment of clinical genetics trainees into the field, improving workforce diversity, and enhancing collaborative practice are a start, but will require a concerted effort and innovations across many stakeholders to fully realize. Salary enhancement and increased funding support for trainees will be integral in achieving this goal. If the workforce is successfully enhanced, genetics patients will benefit more from cutting edge research and therapies, in addition to a more collaborative approach to medical genetics care.

Using the Findings of a National Survey to Inform the Work of England's Genomics Education Programme
Author(s): Simpson et al.
Source: Frontiers in Genetics
Publication date: 2019
For those NHS staff that need to understand genomics and apply this to their practice, our findings suggest there are two levels of education and training resources required. The first is general information targeted to professional groups and the second is cross-professional resources on specific areas or activities that form part of the clinical pathway. However, the results from these surveys also emphasize the need for ongoing awareness raising about genomics in general, as there are still healthcare

professionals, as well non-clinical NHS staff, who do not know what genomics is, let alone how it can be applied to healthcare.

Genomic Education at Scale: The Benefits of Massive Open Online Courses for the Healthcare Workforce
Author(s): Bishop et al.
Source: Frontiers in Genetics
Publication date: 2019
MOOCs are an excellent vehicle for reaching large numbers of learners from across healthcare professions. The use of frontline practitioners as course mentors was successful in this setting: these mentors enhanced the learning experience, while the model itself developed frontline staff as educators. Further research is needed to see if this model, which may offer a sustainable way to deliver healthcare MOOCs, can be replicated, both in terms of using different professional groups as mentors and in healthcare settings outside of the NHS.

Preparing Medical Specialists to Practice Genomic Medicine: Education an Essential Part of a Broader Strategy
Author(s): Crellin et al.
Source: Frontiers in Genetics 10:789
Publication date: 2019
Findings from the very limited empirical studies conducted to date (largely in the field of oncology) suggest that medical specialists' perceptions of genomic medicine are likely to be complex. Mixed views on the clinical utility of genomic medicine currently exist, with perceived benefits frequently tempered by several concerns. At the same time, specialists generally consider the arrival of genomic medicine inevitable. Most do not feel prepared for this inevitability and perceive a lack of understanding and confidence. While little evidence exists, there is indication that CME in genomic medicine is likely to be broadly welcomed.

Challenges and Opportunities for Genomics Education: Insights from an Institute of Medicine Roundtable Activity Abstract only*
Author(s): Dougherty et al.
Source: Journal of Continuing Education in the Health Professions
Publication date: 2016
This commentary promotes continuing and graduate education – informed by an awareness of barriers, drivers, and best practices – as the most effective approaches for preparing the workforce for genomic medicine and ultimately improving patient care, and argues that the time for education is now.

Primary Care

Anticipating the primary care role in genomic medicine: expectations of genetics health professionals
Author(s): Carroll et al.
Source: Journal of Community Genetics 12(4)
Publication date: 2021
GHPs described a key role for PCPs in genomic medicine that could be enhanced if GHPs and PCPs worked together more effectively, making better use of GHPs as a scarce specialist resource, improving PCP knowledge and awareness of genomics, and increasing GHPs' understanding of primary care practice and how to provide PCPs meaningful education and support. Health system change is needed to facilitate the GHP/PCP relationship and improve care. This might include: PCPs ordering more genetic tests independently or with GHP guidance prior to GHP consultations, genomic expertise in primary care clinics or GHPs being accessible through buddy systems or virtually through telemedicine or electronic consultation, and developing educational materials and electronic decision support for PCPs. Our findings highlight need for change in delivering genomic medicine, which requires

building the relationship between GHPs and PCPs, and creating new service delivery models to meet future needs.

Opportunities for education and learning in primary care genomics Abstract only*
Author(s): Bishop et al.
Source: InnovAiT: Education and inspiration for general practice
Publication date: 2020
The continued evolution of the NHS Genomic Medicine Service, alongside improved accessibility to genomic testing through other routes, will increase consultations regarding genomics issues within primary care. GP trainees identify 'genetics' as a learning gap in their training, with anecdotal evidence suggesting that completion of this section in the curriculum is 'challenging'. To support trainees and trainers in meeting this aspect of GP training, this article outlines relevant educational theory, initiatives and resources in genomics from a primary care perspective. This information will equip the learner with approaches and tools to promote confidence in learning and managing issues involving genomic medicine in practice.

Primary Care Genomics: A roadmap for curriculum change and educational resource development Abstract all available
[Abstract, p. 140]
Author(s): Hayward et al.
Source: European Journal of Human Genetics
Publication date: 2020
Health Education England's Genomics Education Programme (GEP) has developed a roadmap for defining General Practitioners' (GPs) learning needs and identifying priorities for resource development, suitable for adoption by other specialties. The toolkit has become a go-to destination for GPs learning about genomics, and its development roadmap can be adopted by other specialties.

Research

Establishing an infrastructure to optimize the integration of genomics into research: Results from a precision health needs assessment Abstract only*
Item Type: Journal Article
Authors: Allen, Caitlin G.;Bouchie, Gwendolyn;Judge, Daniel P.;Coen, Emma;English, Sarah;Norman, Samantha;Kirchoff, Katie;Ramos, Paula S.;Hirschhorn, Julie;Lenert, Leslie and McMahon, Lori L.
Publication Date: 2024
Journal: Translational Behavioral Medicine 14(7), pp. 386–393
Abstract: Researchers across the translational research continuum have emphasized the importance of integrating genomics into their research program. To date capacity and resources for genomics research have been limited; however, a recent population-wide genomic screening initiative launched at the Medical University of South Carolina in partnership with Helix has rapidly advanced the need to develop appropriate infrastructure for genomics research at our institution. We conducted a survey with researchers from across our institution (n = 36) to assess current knowledge about genomics health, barriers, and facilitators to uptake, and next steps to support translational research using genomics. We also completed 30-minute qualitative interviews with providers and researchers from diverse specialties (n = 8). Quantitative data were analyzed using descriptive analyses. A rapid assessment process was used to develop a preliminary understanding of each interviewee's perspective. These interviews were transcribed and coded to extract themes. The codes included types of research, alignment with precision health, opportunities to incorporate precision health, examples of researchers in the field, barriers, and facilitators to uptake, educational activity suggestions, questions to be answered, and other observations. Themes from the surveys and interviews inform implementation strategies that

Technology

Digital interventions for genomics and genetics, empowerment, and service engagement: a systematic review
Author(s): Gasteiger et al.
Source: Journal of Community Genetics 14
Publication date: May 2023
Background: Patient-facing digital technologies may reduce barriers to and alleviate the burden on genetics services. However, no work has synthesised the evidence for patient-facing digital interventions for genomics/genetics education and empowerment, or to facilitate service engagement more broadly. It is also unclear which groups have been engaged by digital interventions. Aim: This systematic review explores which existing patient-facing digital technologies have been used for genomics/genetics education and empowerment, or to facilitate service engagement, and for whom and for which purposes the interventions have been developed. Methods: The review adhered to the Preferred Reporting Items for Systematic reviews and Meta-Analyses guidelines. Eight databases were searched for literature. Information was extracted into an Excel sheet and analysed in a narrative manner. Quality assessments were conducted using the Mixed Methods Appraisal Tool. Results: Twenty-four studies were included, of which 21 were moderate or high quality. The majority (88%) were conducted in the United States of America or within a clinical setting (79%). More than half (63%) of the interventions were web-based tools, and almost all focussed on educating users (92%). There were promising results regarding educating patients and their families and facilitating engagement with genetics services. Few of the studies focussed on empowering patients or were community-based. Conclusion: Digital interventions may be used to deliver information about genetics concepts and conditions, and positively impact service engagement. However, there is insufficient evidence related to empowering patients and engaging underserved communities or consanguineous couples. Future work should focus on co-developing content with end users and incorporating interactive features.

Competency Frameworks

Building a competency framework to integrate inter-disciplinary precision medicine capabilities into the Medical Technology and Pharmaceutical Industry
Author(s): Conway and Chisholm
Source: Therapeutic Innovation and Regulatory Science 58 pp. 567-577
Publication date: 2024
Introduction: Integration of precision medicine (PM) competencies across the Medical Technology and Pharmaceutical industry is critical to enable industry professionals to understand and develop the skills needed to navigate the opportunities arising from rapid scientific and technological innovation in PM. Our objective was to identify the key competency domains required by industry professionals to enable them to upskill themselves in PM-related aspects of their roles. Methods: A desktop research review of current literature, curriculum, and healthcare trends identified a core set of domains and subdomains related to PM competencies that were consistent across multiple disciplines and competency frameworks. A survey was used to confirm the applicability of these domains to the cross-functional and multi-disciplinary work practices of industry professionals. Companies were requested to trial the domains to determine their relevance in practice and feedback was obtained. Results: Four PM-relevant domains were identified from the literature review: medical science and technology; translational and clinical application; governance and regulation and professional practice. Survey results refined these domains, and case studies within companies confirmed the potential for this framework to be used as an adjunct to current role specific competency frameworks to provide a specific focus on needed PM capabilities. Conclusion: The framework was well accepted by local industry as a supplement to role specific competency frameworks to provide a structure on how to integrate new and evolving technologies into their current workforce development planning and build a continuous learning and cross-disciplinary mindset.

The 2023 Genomic Competency Framework for UK Nurses
Source: NHS England Genomics Education Programme
Publication date: 2023
This report describes the process of updating an competency framework for UK nurses undertaken by NHS England's National Genomics Education programme. The report outlines the healthcare context in which these competencies sit, the background to the work and the methodology used to produce the framework.

Personalised precision medicine for health care professionals: development of a competency framework
Author(s): Martin-Sanchez et al.
Source: JMIR Medical Education
Publication date: 2023
Background: Personalized precision medicine represents a paradigm shift and a new reality for the health care system in Spain, with training being fundamental for its full implementation and application in clinical practice. In this sense, health care professionals face educational challenges related to the acquisition of competencies to perform their professional practice optimally and efficiently in this new environment. The definition of competencies for health care professionals provides a clear guide on the level of knowledge, skills, and attitudes required to adequately carry out their professional practice. In this context, this acquisition of competencies by health care professionals can be defined as a dynamic and longitudinal process by which they

use knowledge, skills, attitudes, and good judgment associated with their profession to develop it effectively in all situations corresponding to their field of practice. Objective: This report aims to define a proposal of essential knowledge domains and common competencies for all health care professionals, which are necessary to optimally develop their professional practice within the field of personalized precision medicine as a fundamental part of the medicine of the future. Methods: Based on a benchmark analysis and the input and expertise provided by a multidisciplinary group of experts through interviews and workshops, a new competency framework that would guarantee the optimal performance of health care professionals was defined. As a basis for the development of this report, the most relevant national and international competency frameworks and training programs were analyzed to identify aspects that are having an impact on the application of personalized precision medicine and will be considered when developing professional competencies in the future. Results: This report defines a framework made up of 58 competencies structured into 5 essential domains: determinants of health, biomedical informatics, practical applications, participatory health, and bioethics, along with a cross-cutting domain that impacts the overall performance of the competencies linked to each of the above domains. Likewise, 6 professional profiles to which this proposal of a competency framework is addressed were identified according to the area where they carry out their professional activity: health care, laboratory, digital health, community health, research, and management and planning. In addition, a classification is proposed by progressive levels of training that would be advisable to acquire for each competency according to the professional profile. Conclusions: This competency framework characterizes the knowledge, skills, and attitudes required by health care professionals for the practice of personalized precision medicine. Additionally, a classification by progressive levels of training is proposed for the 6 professional profiles identified according to their professional roles.

A cross-professional competency framework for communicating genomic results Abstract only*
Author(s): Pichini et al.
Source: Journal of Genetic Counseling 33(1)
Publication date: November 2023
To ensure genomic medicine is delivered safely and effectively, it is crucial that healthcare professionals are able to understand and communicate genomic results. This Education Innovation describes a nationally agreed, cross-professional competency framework outlining the knowledge, skills and behaviors required to communicate genomic results. Using principles of the nominal group technique, consensus meetings with clinical, scientific and educational experts identified six stages in the return of results process, drafted and iterated competencies. Competencies were then mapped across three levels to acknowledge different degrees of experiences and scopes of practice. The framework was open for consultation with healthcare professionals and patient communities before being published. The finalized framework includes six core competency statements required to communicate genomic results. This framework is designed to be a guide for best practice and a developmental tool to support individuals and organizations. It can be used by healthcare professionals, such as genetic counselors, to identify individual learning needs or to structure the development of training for other healthcare professionals who are increasingly involved in requesting and returning results for genomic tests.

Computational personalised medicine professionals: explore competencies and career profiles in computational personalised medicine
Source: PerMedCoE
Publication date: August 2022
Discover and explore career profiles in computational personalised medicine.
The competency framework for professionals in the field of computational personalised medicine has been developed by PerMedCoE. A competency is an observable ability of any professional, integrating multiple components such as knowledge, skills and behaviours. The competencies that an individual might need to fulfil a particular role are listed in reference profiles, which can be used to guide career choices.

A nationally agreed cross-professional competency framework to facilitate genomic testing
Author(s): Pichini and Bishop
Source: Genetics in Medicine 24(8)
Publication date: August 2022
Purpose: The study aimed to develop a nationally agreed, cross-professional competency framework outlining the knowledge, skills, and behaviors required to facilitate genomic tests. Methods: Using principles of the nominal group technique, a consensus meeting with 25 experts mapped themes to an initial framework and voted on areas of inconsistency. A revised framework was open for consultation with health care professionals and patient communities before being published. An evaluation, using an online survey, was conducted to explore early use and factors to facilitate adoption of the framework. Results: The framework identified 8 competencies required to facilitate genomic tests. The evaluation (239 survey responses from health care professionals) indicated that the framework addresses a timely need among users and identified ways to improve awareness and accessibility for different health care professional groups. Conclusion: This framework can be used as a guide for best practice by health care professionals who request genomic tests. It can also provide a foundation to identify learning needs and structure training such that conversations about genomic testing can be delivered in a consistent manner across specialties. These competencies can also be used as a reference to evaluate how consent is facilitated in different specialty areas to enhance the responsible delivery of genomic medicine.

Genomics Education Programme
Source: Health Education England
Publication date: 2021
Delivering genomics education, training, and experience for the healthcare workforce.
See also: Facilitating genomic testing: A competency framework
This competency framework identifies the core competencies required to facilitate and consent patients for germline genomic tests. The Genomics Education Programme has developed this framework in collaboration with healthcare professionals from across the health service. Each competency is provided with links to supporting evidence-based educational resources.

Master's in Genomic Medicine framework
Source: Genomics Education Programme (Health Education England)
The Master's in Genomic Medicine framework is an educational programme designed to provide healthcare professionals with a multidisciplinary perspective on genomics and its applications in healthcare.

A nationally agreed cross-professional competency framework to facilitate genomic testing Abstract all available [Abstract, p. 767]
Author(s): Pichini et al.
Source: European Journal of Human Genetics
Publication date: 2020
The Genomics Education Programme has developed a nationally agreed cross-professional competency framework outlining the knowledge, skills and behaviours required to facilitate genomic testing. This framework can be used as a guide for best practice by clinicians involved in requesting genomic tests. For those delivering education, the framework provides a foundation to identify learning needs and structure training, such that consent conversations can be delivered in a consistent manner across specialties. In addition, these competencies can be used as a reference to evaluate how consent is facilitated in different specialty areas to enhance the delivery of genomic medicine.

The changing face of clinical genetics service delivery in the era of genomics: a framework for monitoring service delivery and data from a comprehensive metropolitan general genetics service Full text available with NHS OpenAthens account*
Author(s): Fennell et al.
Source: Genetics in Medicine 22(1)
Publication date: 2020
Development of a standardized method of determining workforce adequacy in clinical genetics, based on average work RVUs or similar metrics, would assist in clarifying acceptable international benchmarks. Combining this with modeling on training program outputs, as well as physician retention and retirement timelines, will be important to guide strategic workforce planning. Cancer genetics and subspecialties with significant practice structure variation may require customized frameworks. This framework will capture areas of practice that require modification in response to shifts in technology and workload. Longitudinal data gained by implementation of this framework and publication of the outcomes will assist services worldwide in refining best practices.

The first competency based framework in genetics/genomics specifically for midwifery education and practice Abstract only*
Author(s): Tonkin et al.
Source: Nurse Education in Practice
Publication date: 2018
This paper details a competency framework to help address the need for structured guidance around genetic and genomic education and training for midwives.
• Genomics is an integral component of the maternal and family health care pathway.
• Learning outcomes and practice indicators develop over time and with experience.
• Genomic competence can improve care, consistency and safety for women and families.

Methods of Genomic Competency Integration in Practice Full text available with NHS OpenAthens account*
Author(s): Jenkins et al.
Source: Journal of Nursing Scholarship 47(3)
Publication date: 2015
Champion dyads used creativity to design interventions that supported their staff to advance a new complex competency, genomics, within their facilities. They informed interprofessional colleagues about the value of nursing leadership in advancing genomic healthcare translation and supporting provision of safe, quality care delivery that integrates new knowledge. Developing policies to facilitate responsible translation of genomics into nursing practice is an effective strategy for nursing leaders who accept the responsibility to design systems and infrastructure that support the translation of genomics into health care.

eLearning

Genomics in the NHS
Source: eLearning for Healthcare
The Genomics in the NHS programme is a collection of elearning courses developed by NHS educational teams to support and educate healthcare professionals about genomic medicine and the benefit it will bring to patient care. With the launch of the NHS Genomics Medicine Service (GMS) in England, more healthcare professionals than ever before are likely to encounter genomics in their daily role. As it increasingly becomes a part of routine care, all healthcare professionals, not just those within specialist departments, will need some level of understanding of the impact genomics will have on the diagnosis, treatment and management of a wide variety of conditions. The elearning courses have been developed for those directly involved in the GMS and the wider healthcare workforce.

*Help accessing articles or papers

Where a report/ journal article or resource is freely available the link has been provided. If an NHS OpenAthens account is required this has been indicated. It has also been highlighted if only the abstract is available. If you do not have an OpenAthens account you can self-register here.

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