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**Prioritising the Pipeline: Updating Undergraduate Medical Genetics Education to Support
the Future of Genomic Medicine in Australia**

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Abstract

Advances in genomic medicine are transforming modern healthcare, with genetics playing an increasingly integral role across multiple medical specialties. Existing studies indicate that many practicing clinicians feel underprepared to integrate genomics into routine clinical care, highlighting deficiencies in early medical training. While postgraduate initiatives have sought to improve genomic skills among doctors and nurses, there has been limited investigation into how genetics is currently taught at the undergraduate level and whether medical graduates achieve the competencies necessary for contemporary practice.

This article examines the current state of genetics education in Australian medical schools, reviews international competency frameworks, and discusses the challenges of ensuring that genetics training remains relevant in an evolving healthcare landscape. A comparison of competency-based models from Australia, the United Kingdom, Europe, and the United States reveals common foundational principles but also key differences in the clinical integration of genomics. While international frameworks offer valuable insights, their direct application to Australia is limited by differences in healthcare delivery, workforce needs, and genomic testing policies.

A structured approach is proposed to improve genetics education at the undergraduate level: identifying the current intended learning outcomes across Australian medical schools, assessing the realised competencies of medical graduates, applying evidence-based educational strategies to integrate genetics effectively throughout medical curricula, and developing a nationally agreed-upon framework for genetics competencies. By addressing these gaps, medical education can ensure that graduates are adequately prepared to apply genomic medicine in patient care, contribute to precision medicine initiatives, and navigate the ethical, legal, and social implications of genetics in healthcare.

As genomic medicine continues to expand, undergraduate medical education must be proactive rather than reactive, ensuring that future Australian doctors enter the workforce equipped with the foundational knowledge and clinical skills necessary to integrate genetics into their practice. This article brings attention to undergraduate medical genetics education and recognises the role in bridging the gap between scientific advancements and clinical application to meet the demands of the current, and future Australian healthcare system.

I INTRODUCTION

Scientific advances in genetics and genomics have revolutionised how we approach health and disease (Hyland et al., 2019). Through the introduction of Next Generation Sequencing and the completion of the Human Genome Project the applications of genetics in clinical practice have grown exponentially (Levy & Myers, 2016; McGuire et al., 2020). While in most cases translation of scientific advances to practical clinical use takes time, genomic medicine has advanced at unprecedented speed, from widespread availability of sequencing to the development of targeted therapies and government funded rebates for genetic testing - all within the span of a decade. Today, genomics play a role in nearly every medical specialty. However, these rapid developments present challenge where resource availability, equity of access, and geographic spread are already barriers to delivery of specialist genetic services for many patients.

Demand has quickly outgrown the capacity of the specialist genetics workforce (Burns et al., 2019), and non-genetics physicians will hold an increasing role in genetic and genomic care for patients into the future. Already, these increased responsibilities range from general practitioners managing more patients with known genetic conditions owing to new additions to newborn screening panels and genetic carrier screening, to specialist physicians, such as oncologists, surgeons, and cardiologists, applying genomics to precision diagnostics and treatments for their patients. Furthermore, a growing number of conditions are being streamlined through mainstream pathways, where non-genetics specialists oversee genetic testing, with Medicare rebates for germline testing for breast, ovarian, and prostate cancer patients (White et al., 2020).

Acknowledging the need for genetics literacy across our healthcare system, building a skilled workforce was a key priority in the National Health Genomics Policy Framework, 2018-2021 (National Health Genomics Policy Framework 2018–2021, 2017). Consequently, a growing body of research has been published characterising the genetics knowledge and skills of the practising workforce (Nisselle et al., 2024). These studies indicate that many clinicians feel underprepared to integrate genomics into their practice, leading to underutilisation of mainstream pathways and delays in diagnosis and treatment, (Cormack et al., 2024; French et al., 2023; Nisselle et al., 2023; Young et al., 2024). In response, various educational interventions have been designed to address knowledge gaps among healthcare professionals, including continuing professional development and online modules (Cusack et al., 2021; Nisselle et al., 2019).

However, less attention has been paid to genetics education at the medical undergraduate level, despite the potential to lay the foundation in preparing the future workforce. Unlike postgraduate initiatives, undergraduate medical education offers an early and structured opportunity to standardise genetic competencies across the medical curriculum before students enter practice. Despite the importance of this stage in training, undergraduate genetics education in Australia has not been as thoroughly researched as postgraduate interventions. A current study by Australian Genomics is surveying medical students in Australia and New Zealand to assess their genetics knowledge and exposure to genomics education, and the results of this study will provide valuable insights into existing gaps (Australian Genomics, n.d.). Furthermore, only recently was a study published on pharmacogenetics content across Australian medical schools, and it found that current curricula were insufficient to prepare students for the integration of pharmacogenomics into clinical practice (Thomas et al., 2024). These findings highlight the urgent need for a comprehensive evaluation of genetics education across all Australian medical schools.

Undergraduate medical education has itself undergone significant transformations over the past few decades, and this has important implications to updating genetics teaching. The traditional Flexnerian model, which emphasized basic science education as a separate, preclinical phase, has gradually given way to competency-based education and integrated teaching approaches. These changes reflect the requirement for medical graduates to develop practical and applied knowledge, rather than rote learning. Given these broader shifts in medical education, updating genetics teaching requires not only identifying what should be taught, but also determining the most effective ways to teach it.

II WHAT SHOULD WE TEACH? COMPETENCY BASED FRAMEWORKS FOR THE AUSTRALIAN HEALTHCARE SYSTEM

In the early 1900's the landmark Flexner report advocated that an education in basic science should hold precedent in the training of physicians (McColl et al., 2012). The effect of the Flexner approach was that basic science was delivered as a preclinical curriculum in an academic environment, largely through traditional methodology including didactic lectures and rote learning of textbooks. However, as scientific knowledge grew in both volume and complexity, rote memorisation insufficiently prepares students to understand and therefore apply scientific principles in practice.

Genetics, specifically, has evolved from what was once considered a basic science to a core pillar across clinical practice. Unlike many other traditional 'preclinical' disciplines, genetic knowledge and technologies continue to change rapidly, requiring curricula that can adapt over time. Thus, rather than relying on static, rote-learning curriculum, genetics education must be competency-driven and clinically integrated. Learning outcomes that address comprehension help students apply and retain their knowledge. Such is well acknowledged by Bloom's taxonomy, which is a widely used classification system that describes six categories of cognitive skills from lower order (remembering) to higher order skills that require a greater degree of student understanding (analysing, applying, evaluating) (McColl et al., 2012).

However, questions remain regarding the appropriate breadth and depth of skills should be expected of non-genetics physician, and this presents a barrier to applying competency-based approaches in undergraduate medical education (Talwar et al., 2017). Internationally, several competency frameworks have been developed in attempt to answer this (Human Genetics Society of Australasia, 2022; Tognetto et al., 2019) (Box 1.). While common principles are shared between these frameworks, including molecular genetics knowledge, communication skills, and appreciation of the ethical, legal, and social implications of genetic testing, there are also important differences reflecting regional and cultural variations, patient demographics, and educational priorities. For example, the US model includes competencies relevant to a privatised healthcare model and direct-to-consumer testing (Quintero-Rivera et al., 2024). In UK and Australia, however, publicly funded testing models such as the NHS Genomic Medicine Service and Medicare funded testing requires knowledge of eligibility criteria, referral pathways, and appropriate test selection (Pichini & Bishop, 2022). Additionally, the intended population for which each of these frameworks has been developed for is important, and not all competencies listed are easily adapted for medical school curriculum. This is consistent with results from by Tognetto et al. (2019), who found that core genetics competencies differed between physicians and non-physician health professionals. Such may also true when comparing competency frameworks that developed for undergraduate medical education versus continuing medical education for postgraduate physicians. Additionally, for competencies to remain relevant, continuous review and updates are required. This is highlighted by the recent updates to the core competencies for medical students from the Association of Professors of Human and Medical Genetics Human Genetics (Quintero-Rivera et al., 2024) which introduced skills that reflect developments in precision medicine and diagnostic technology from the earlier publications. At present there is no formal, nationally recognised framework for genetics in Australian undergraduate medical education. While international frameworks provide a foundation, direct implementation without adaption will not fully address the needs of Australian healthcare. Likewise, the educational requirements of Australian medical education system must be considered.

Box 1: International competency frameworks for teaching genetics in undergraduate medical education

Region	Framework	Target population	Key Organisations	Reference
Europe	Genetic education and the challenge of genomic medicine: development of core competences to support preparation of health professionals in Europe	Non-genetics health professionals	European Society of Human Genetics	(Skirton et al., 2010)
United Kingdom	Cross-professional competency framework to facilitate genomic testing, Genomics Education Programme, Health Education England (2022)	Non-genetics health professionals	National Health Service	(Pichini & Bishop, 2022)
	Teaching of clinical genetics in Britain: A report from the Royal College of Physicians of London	Postgraduate physician trainees	Royal College of Physicians	(Johnston, 1990)
United States	Core competencies for undergraduate medical education in genetics and genomics Association of Professors of Human and Medical Genetics (APHMG) (2022)	Medical students	Association of Professors of Human and Medical Genetics	(Quintero-Rivera et al., 2024)
	Framework for development of physician competencies in genomic medicine: (2014)	Non-genetics health professionals	The National Human Genome Research Institute, Inter-Society Coordinating Committee for Physician Education in Genomics	(Korf et al., 2014)
Australia	Australian Medical Genomics Competency Framework (2022)	Medical students	Australian Society of Genetic Counsellors, Human Genetics Society of Australasia	(Human Genetics Society of Australasia, 2022)

The Australian Medical Council (AMC) are the governing body for accreditation and assessment standards for Australian medical schools (Australian Medical Council, n.d). This includes endorsement of the AMC graduate outcome standards – The essential skills expected of a graduating student prior to commencing clinical practice. These outcomes address four domains: Science and Scholarship, Clinical Practice, Health and Society, and Professionalism and Leadership, and serve to provide direction to medical schools in development of curriculum content, teaching and learning approaches, assessment programs and guide relevant governance structures. A well designed and unified genetics framework must integrate genetics across all four domains to ensure graduates are prepared not just in genetics knowledge, but in the clinical, professional, and societal applications in the Australian healthcare system. The Human Genetics Society of Australasia (HGSA) (2022) provide a useful starting point, defining key competencies under three areas – Knowledge, Abilities, and Attitudes. Knowledge refers to a student's ability to recall and interpret and understand core genetics concepts. Abilities refers to skills students are expected to master, such as counselling a patient considering genetic testing, or obtaining informed consent. Attitudes reflect a student's appreciation for the inherent ethical and social complexities within genetic testing (Tognetto et al., 2019). This structure can be adapted to align with the AMA graduate outcome domains (Box 2.).

Key similarities between the HGSA and international frameworks relate to genetics knowledge, primarily within the Science and Scholarship domain. Genetics concepts based on contemporary models and molecular biology improves student understanding of genomics applications and precision medicine (Redfield, 2012). However, unique considerations for Australian healthcare are easily identified across abilities and attitudes, and this is most evident in the remaining three AMC domains (Box 2.) For example, delivering genetic care within the Australian healthcare system is unique given the enormity of geographic and multicultural diversity across the population it serves. Therefore, although specialist centres exist, these are largely concentrated in metropolitan locations, and this may lead rural patients to seek genetic advice from their local general practitioner rather than requesting a referral. Integrating these considerations and communication skills will be increasingly important for medical schools to adequately prepare students (McClaren et al., 2020; Mladenović et al., 2024). Physicians must communicate complex genetics information to patients and their families in a culturally and socially sensitive manner, especially in migrant, linguistically diverse, and Indigenous populations. Genetic literacy across the Australian population requires physicians to educate and support their patients to make informed decisions and navigate genetic care. This requires an understanding of currently available genetic technologies, population screening programs, and eligibility for genetic testing under government funding. Medical school curricula should ensure students understand that genetic risk assessment varies given differences in allele frequencies between ethnicities, and importantly, Indigenous patients remain underrepresented in current population-based datasets. Attention must also be paid to the ethical and legal issues of genetic testing in Australia, such as equity of access in rural areas, and implications for life insurance eligibility (Cusack et al., 2021; Stark et al., 2019).

Box 2: Suggested framework for Genetics Competencies for Medical Students: adapted for the Australian medical education and healthcare system: Adapted from Core Capabilities in Genetics & Genomics for Medical published by The Human Genetics Society Australasia (Core Capabilities in Genetics & Genomics for Medical Graduates, 2022)

AMC Graduate Outcome Domains	HGSA Core Competencies	Specific Genetics Competencies for Medical Graduates	Australian Context & Relevance
Science and Scholarship (Medical Knowledge & Evidence-Based Practice)	Knowledge	- Understand the fundamental principles of molecular genetics, inheritance patterns, and genomic medicine.	- Ensuring that medical graduates have a strong foundation in genomic science, aligned with contemporary medical advances.
		- Recognise how genetic variation affects disease processes, pharmacogenomics, and precision medicine.	- Reinforcing the ability to interpret genetic literature and clinical guidelines, as required for evidence-based medicine.
		- Critically evaluate genetic research and apply evidence-based genomic knowledge in clinical practice.	- Ensuring graduates are equipped with skills to remain up to date with genomic advances throughout their career.
Clinical Practice (Patient-Centered Care & Clinical Decision-Making)	Abilities	- Identify clinical scenarios where genetics plays a role in diagnosis, prognosis, or treatment.	- Preparing future doctors to apply genomic principles in day-to-day clinical decision-making.
		- Take a family history and assess genetic risk using standard risk assessment tools.	- Ensuring graduates can effectively integrate genetic risk assessment into patient consultations.
		- Order, interpret, and communicate genetic test results appropriately.	- Promoting case-based and problem-based learning to support clinical reasoning in genetics.
		- Recognise the limitations of current genomic knowledge and testing technologies.	
Health and Society (Public Health, Population Health & Health Equity)	Attitudes	- Understand the role of genetics in public health, including population screening programs and epidemiology.	- Ensuring medical graduates are prepared to navigate Australian healthcare policies related to genomics.

		- Recognise health disparities in genomic medicine, particularly in Indigenous and rural communities.	- Addressing genomic health equity, particularly the challenges faced by rural and Indigenous populations.
		- Understand health policy and regulation related to genetics (e.g., Medicare-funded genetic testing, privacy laws, health/life insurance implications and laws).	- Reinforcing the importance of ethical and regulatory considerations in genetic service delivery.
Professionalism and Leadership (Ethics, Communication & Collaborative Practice)	Abilities & Attitudes	- Communicate genetic risks and test results clearly and compassionately to patients and their families.	- Ensuring that all medical graduates can engage in ethical and culturally competent discussions about genetics.
		- Apply ethical principles to genetic medicine, including issues of informed consent, genetic discrimination, and data privacy.	- Reinforcing professional boundaries—knowing when to refer to specialist genetics services while maintaining a foundational level of genetic literacy.
		- Recognise the limits of personal expertise and refer patients appropriately to clinical geneticists, genetic counselors, and other specialists.	- Promoting interdisciplinary collaboration and recognising the roles of other members of the team in genomic medicine.

III HOW SHOULD WE TEACH IT? CONTEMPORARY PEDAGOGICAL APPROACHES IN GENETICS TEACHING

Although integrated approaches have progressed since the Flexner era (McColl et al., 2012), most Australian medical schools continue to make some distinction between pre-clinical and clinical teaching. This often involves focusing on foundation science for earlier year students, with a progression to a clinical focus in the latter years. However, this separation between preclinical and clinical teaching can have important implications for pedagogical approaches relevant to teaching genetics. For example, genetics can be delivered as a standalone course that provide a dedicated view of the principles of molecular genetics and genomics. Commonly, introductory science teaching is undertaken by non-clinical faculty, and while this approach is effective in covering the fundamental genetics concepts, it limits the connection with clinical learning. Genetics is at risk of being introduced as a scientific field with limited practical applications. In turn this leads to poorer knowledge retention for medical students (Greb et al., 2009), and can be a barrier to their future understanding of genetic and genomic applications in their own practice.

Recognising the limitations of standalone basic science teaching, integrated approaches to curricular design have been increasingly popular in medical education (Brauer & Ferguson, 2015; Dasgupta et al., 2020; Mayer, 2010). Brauer and Ferguson (2015) describe two models of curricular integration in current medical education. Horizontal integration refers to integration of subject matter across disciplines, but within a finite time. For example, Problem Based Learning (PBL) activities are widely used in preclinical medical teaching. PBLs utilise a clinical case study to introduce students to scientific concepts within a clinical context. Vertical integration refers to integration longitudinally at different stages of the medical curriculum (Brauer & Ferguson, 2015; Wijnen-Meijer et al., 2020). This approach recognises that educational approaches change across the medical school continuum, with a transition from classroom-based to clinical-based activities (Wijnen-Meijer et al., 2020). While there are obvious differences in how these approaches may influence medical school curriculum design, there are common benefits in integrated approaches.

Firstly, integrated curriculums demonstrate clinical relevance of genetics principles. This leverages key components of Knowles' revolutionary theories of adult learning, through which adults require relevance to engage with and retain knowledge (Knowles, 1977). Given a common barrier to student engagement with basic science content is perceived relevance, it is logical that this could be addressed by both enhancing relevance during early-years teaching and reinforcing genetics as a clinical discipline in later-years teaching. Secondly, inter-disciplinary approaches, where genetics is imbedded in other disciplines such as population health or pharmacology, may allow medical schools to overcome the challenge of curriculum overload, time and resource limitations. This also addresses the important concept of Cognitive Load Theory (CLT) (Mihalca et al., 2011). CLT recognises that a learner can only retain a small amount of information in their working memory at any one time, and therefore educational approaches. This is especially important given the complex and abstract molecular genetics concepts that are required for a learner to engage with genomic and precision medicine approaches. Finally, by continued engagement with scientific principles in the latter phase of undergraduate medical training, students are encouraged to utilise evidence-based approaches to patient care. This skillset will be crucial for future physicians to remain up to date with scientific advances in an era of rapid technological growth and development.

IV CHALLENGES AND FUTURE DIRECTIONS

While genetics is now embedded across all areas of medicine, there remains a significant disconnect between the advances in genomic medicine and the how genetics is taught in Australian medical schools. Internationally there has been substantial interest in improving genetics education in undergraduate medical education, with several overseas studies evaluating genetics curricula (French et al., 2023; Whitley et al., 2020), student competencies (Kapur et al., 2024), and designing educational interventions specifically for undergraduate medical education programs. Through this research many creative methods have been applied, including active-learning and technology enhanced approaches (Alotaibi & Cordero, 2021; Bennett et al., 2017; Champion et al., 2019; Dasgupta et al., 2020; Haspel et al., 2021; McClaren et al., 2020; Plunkett-Rondeau et al., 2015). However, these findings are not always transferable to the Australian medical education system, where physicians must be prepared to work under a different healthcare model, navigate government funded genetic testing programs, and address specific challenges such as rural and Indigenous health disparities. Therefore, while international research provides valuable insights, Australia requires a tailored approach to ensure appropriate genetics training for the future workforce. Underpinning this remains the question of what should be considered core, versus specialised genetics knowledge for the future workforce of physicians. Indeed, scope of practice of both non-medical, and medical non-specialists is undergoing change not just in genetics, but in many areas of the Australian healthcare system. This is compounded by the unique service provision barriers for rural populations given the country's geographic spread, and where the role of a non-specialist may change between low- and high-resource available locations. While defining this complex and evolving target is beyond the scope of this

paper, the reality of current practice remains insufficient to meet patient demands. While the role of non-genetics physicians will undoubtedly change over time, current medical education programs have a responsibility to prepare students based on best available evidence at this time.

There are also many practical considerations that make implementing change to genetics education in Australian medical schools complex. Medical schools, even within Australia, vary widely in size, structure, curricular design, funding, faculty expertise, and access to research and clinically focused resources. Likewise, medical students are a diverse population, and the educational needs will vary between undergraduate and postgraduate student populations, which is a common distinction between medical schools in Australia. Consistent with this, the Medical Deans Australia and New Zealand, states it is not possible to have one framework applied to all universities due to the differences in curricula and lack of unanimous evidence of 'best' outcomes (Medical Deans Australia and New Zealand, n.d.). This implies that no one approach will be suitable for all schools, and for educational interventions to be sustainable and successful, there must be careful consideration of the target context and best available resources.

Efforts to improve genetics education through targeted, evidence-based interventions have already been made within the existing workforce. The Workforce & Education research program of the Australian Genomics Health Alliance have established frameworks that span from evaluating knowledge and skill deficits (Stark et al., 2019), to implementing needs-based interventions (Nisselle et al., 2024), to evaluation frameworks to identify the immediate-long term outcomes (Nisselle et al., 2024). These tools have been designed to be adaptable to different contexts, with emerging applications seen in the continuing medical education field for Australian general practitioners (Cusack et al., 2021), non-genetics physicians (Nisselle et al., 2019), and nurses (Wright et al., 2019). Although no equivalent national initiative currently exists for undergraduate medical education, these frameworks offer a solid foundation.

To address these challenges and ensure future medical graduates receive appropriate genetics education, a structured, evidence-driven approach is proposed. This process requires a comprehensive assessment of current genetics education in Australian medical schools, including an evaluation of intended learning outcomes, teaching methodologies, and curricular integration. This will identify existing frameworks and provide insight to where significant gaps or inconsistencies exist. Following this, it is important to determine the actual competencies achieved by graduating students, recognising that intended learning outcomes are not always compatible with what knowledge and skills a student retains (Thomas et al., 2024). This can be achieved through assessment data, and graduate surveys. By comparing intended learning outcomes with actual competencies, medical educators can identify where deficiencies lie, and where evidence-based pedagogical interventions may be most beneficial.

On a broader scale, developing a nationally standardised framework for genetics education that reflects the unique requirements of the Australian healthcare system is a priority. This framework should be informed by existing competency models such as the HGSA (Human Genetics Society of Australasia, 2022). It will be critical that this process engages medical schools, accrediting bodies such as the AMC, and clinical genetics professionals to collaboratively determine the expected competencies for all graduating medical students.

Finally, given the rapid and relentless progress within genomic medicine, commitment to continuous evaluation and intervention will be required. The success of the Australian Genomics Health Alliance in implementing competency-based interventions for postgraduate doctors and nurses underscores the effectiveness of structured educational reform. A similar national effort at the undergraduate level will be essential to ensuring that future Australian doctors enter the workforce already equipped with the foundational knowledge and practical skills required to integrate genomic medicine into their practice. Given the pace of advancements in genetics, medical education must be proactive rather than reactive in preparing students for a healthcare environment where genomics is increasingly integral to diagnosis, treatment, and personalised medicine.

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