Core Capabilities in Genetics & Genomics for Medical Graduates

As genomic investigation and medicine are increasingly integrated into mainstream healthcare, the Human Genetics Society of Australasia has undertaken a timely review of existing competencies and capabilities in genetics and genomics for medical graduates.

Both research into and our subsequent knowledge of genomics have advanced rapidly over the past decade. The translation of reseach discoveries and genomic testing into clinical care is impacting all areas of medicine. In order to provide care in the new era of genomic medicine, medical graduates require a genomic knowledge base that is both deep and broad. [1,2] The current rapid changes and steep trajectory of genomic medicine has demonstrated that what we consider to be emerging knowledge today will become essential knowledge in three to five years time.[3]

This document is intended to provide a genomic compentency roadmap for current and future medical workforces. It is intended to guide medical school curricula and inform credentialling of postgraduate education bodies and specialist training colleges.

The capabilities are structured to:

1. outline key knowledge domains, aligned with the Australian Medical Council's graduate outcome statements, and propose an associated capability level expected of genomics-literate medical graduates and future practitioners;
2. reflect requirements for mainstreaming genetic and genomic practice by classifying competencies as emerging, desirable or foundational. It is anticipated that higher order competiencies will become foundational knowledge in the near future as genomics expands across all areas of medicine. Medical practitioners of all persuasions will be involved across the clinical capabilities associated with genomic testing including: selection of appropriate genomic tests; genomic screening; leading the genomic consent process; discussing testing with patient and their families; and managing outcomes. These practical activities form the majority of the new learning objectives in this document; and
3. highlight implications for related fields as genomics crosses disciplinary boundaries. Genomic implications for population health and screening; health policy, bioethics; and emerging models of medical care such as precision medicine, pharmacogenomics, microbial genomics, and direct-to-consumer (online) genetic testing are also included in this document.

The following competencies represent the shared work of Australian and New Zealand geneticists and genomic educators. This group revised and updated the capabilities, with reference to the peer-reviewed literature.[4–9] This document and its recommendations have been reviewed and endorsed by the ten members of the HGSA Core Capabilities in Genetics & Genomics for Medical Graduates Working Group (see appendix).

I commend this document to inform the modernised curricula of all Australaiasian medical schools.

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On behalf of the HGSA Core Capabilities in Genetics & Genomics for Medical Graduates Working Group
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Human Genetics Society of Australisia
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Structure of the capabilities tables

The capabilities are primarily divided into two of the four major educational domains prescribed by the Australian Medical Council (AMC):[10]

A. **Science and Scholarship: the medical graduate as a scientist and scholar**
B. **Clinical Practice: the medical graduate as a practitioner**

Those capabilities that align to the remaining two AMC domains have been additionally identified

C. **Health and Society: the medical graduate as a health advocate**
D. **Professionalism and Leadership: the medical graduate as a professional and leader**

Each capability has been assigned a '**knowledge set**' to provide guidance where a capability may sit within a medical curriculum. Additionally, each capability has been assigned a '**knowledge level**', reflective of the depth of understanding the HGSA recommends medical school graduates acquire.

Knowledge set
Science
Public Health
Communication
Ethics, Legal & Social Issues

Knowledge level	
Foundational	Knowledge, abilities and attitudes the HGSA considers necessary for all medical students to acquire by graduation
Desirable	Knowledge, abilities and attitudes the HGSA strongly recommends medical students acquire by graduation
Emerging and/ or specialist	Knowledge, abilities and attitudes the HGSA recommends medical students have an awareness of upon graduation

Within each table, competencies are then classified into a **knowledge**, **ability** or **attitude**. Within this classification system, competencies are clustered into additional topic-specific fields.

A note around deliberate use of language

Specific verbs have been chosen to reflect the level of each capability specific to medical school graduates, referencing Bloom's taxonomy,[11] with 'awareness' delineated as a more basic form of knowledge relevant to graduates who have minimal clinical experience:

- *Be aware:* respect, appreciate, locate, be ready to, maintain
- *Know:* identify, select, define, list, recognise, recall
- *Comprehend:* describe, explain, discuss, communicate, 'be prepared', compare, associate
- *Apply:* demonstrate, conduct, construct, focus, produce, estimate, identify, document, use, order, engage, maintain, incorporate, provide, facilitate, ensure, 'be able to', 'advocate for…'
- *Analyse:* interpret, critique, measure, formulate

The last two levels in Bloom's taxonomy (*synthesise* and *evaluate*) were deemed to be beyond the expectations of medical school graduates.

SCIENCE AND SCHOLARSHIP: THE MEDICAL GRADUATE AS A SCIENTIST AND SCHOLAR

	DOMAIN	SET	LEVEL	COMPETENCY
				KNOWLEDGE
1	A			Recall the structure, organisation and function of nuclear DNA, genes and chromosomes
2	A			Recall the structure, function and transmission of mitochondrial DNA, genes and chromosomes
3	A			Recall the replication of DNA, genes and chromosomes, including their transmission through mitosis and meiosis
4	A			Recall the process and regulation of gene transcription and translation
5	A			Recognise that DNA variability can influence physiological and pathological processes
6	A			Recognise the difference between germline and somatic genetic variants
7	A			Recognise the process and consequence of X-inactivation
8	A			Recognise the transcription and function of non-coding RNA
9	A			Recognise epigenetic regulation of gene expression and epigenetic inheritance
10	A			Recognise defects of DNA replication and DNA repair
11	A			Explain core concepts of genetic epidemiology including population genetics, genetic drift, Hardy Weinberg equilibrium and genetic association
12	A			Recognise the effects of mutagens and teratogens on the developing human
				ABILITY
13	A			Interpret the difference between *de novo* and inherited DNA variants
14	A			Explain genetic variation between humans, including polymorphisms, benign and pathogenic genetic variants
15	A			Explain the aetiology and pathogenesis of common genetic disorders
16	A			Explain cancer pathogenesis: tumour evolution through a series of epigenetic and genetic changes, proto-oncogenes, tumour suppressor genes
17	A			Explain how molecular variants impart disease or disease predisposition
18	A			Apply knowledge of epigenetic regulation to disease pathogenesis
				ATTITUDE
19	A			Recognise molecular biology is a rapidly evolving field and participate in continued professional development as necessary

CLINICAL PRACTICE: THE MEDICAL GRADUATE AS A PRACTITIONER

	DOMAIN	SET	LEVEL	COMPETENCY
				KNOWLEDGE
1	B			Describe the basic patterns of Mendelian inheritance
2	B			Explain the differences between Mendelian, mitochondrial and multifactorial inheritance
3	B			Explain the difference between clinical diagnosis of disease and genetic predisposition to disease
4	B			Explain genotype-phenotype correlations; understanding of how gene variations can influence disease presentation, its severity, and clinical manifestation (anticipation, incomplete penetrance, variable expressivity)
5	B			Describe how both germline and somatic variation result in disease risk and the subsequent risk of recurrence

	DOMAIN	SET	LEVEL	COMPETENCY
6	B			Explain the genomic aetiology of cancer aetiology and the 'multi-hit' model
7	B			Describe common inherited cancer predisposition syndromes
8	B			Explain the aetiology of common inborn errors of metabolism and their general clinical manifestations
9	B			Describe common dysmorphic and non-dysmorphic syndromes
				Precision medicine
10	B			Explain how genotype influences precision/personalised medicine
11	B			Describe how germline and somatic genotype may influence disease management
12	B			Recognise that there is variability in the phenotypic expression of genetic variants and in response to therapy
13	B			Describe the principles of pharmacogenetics/pharmacogenomics and the relationship with personalised medicine
14	B			Explain emerging therapeutic strategies (e.g., gene therapy, mitochondrial donation, stem cell transplantation, personalised medicine)
				Genetic/genomic testing
15	B			Recognise the importance of documenting family history
16	B			Recognise when a patient presentation may have an underlying genetic cause
17	B			Describe genetic databases, guidelines and other resources used in the diagnosis and explanation of genetic and familial disorders
18	B			Recognise the limitations of genomic databases that will affect the interpretation of genomic results (e.g. lack of normative genotypic data of minority groups, including Aboriginal and Torres Straight Islander peoples and Māori)
19	B			Recognise the effect of ethnicity on interpretation of results (founder effects, unknown shared common ancestry)
20	B			Describe the key clinical settings and indications for genetic testing (screening, preimplantation, prenatal, diagnostic, predictive/pre-symptomatic, personalised medicine)
21	B			Describe the key technologies of genetic investigation (e.g., karyotype; single gene and genomic sequencing; RNA sequencing; cell free DNA sequencing)
22	B			Describe how microbial genomics can aid investigation and management of infectious disease
23	B			Recognise that genomic tests require interpretation with respect to the patient's clinical status (e.g., pathogenic, likely pathogenic, and benign)
24	B			Recognise the difference between benign and pathogenic DNA variants in the clinical setting
25	B			Describe the clinical and psychological consequence of a variant of uncertain significance
26	B			Ensure genomic consent includes informing patients that the clinical significance of variants can change with time
27	B			Explain the concepts of analytic reliability, clinical validity, and clinical utility as they relate to genomic testing
28	B			Explain how the admixture of genetic polymorphisms influence health and disease (polygenic scores)
29	D			Be familiar with the strengths and limitations of direct-to-consumer / personal genomic tests and potential risks
30	C			Explain the need for, and challenges to, obtaining informed consent when conducting genetic/genomic testing

	DOMAIN	SET	LEVEL	COMPETENCY
31	C			Recognise the personal and social implications genetic testing may have (e.g., effects on familial relationships and ability to obtain certain types of insurance products) for an individual and their family
32	D			Recognise the professional and legal obligations of medical practitioners to safeguard genetic information
33	C			Appreciate the diverse ethical, social, cultural, religious issues individuals may have when discussing personal genetic information
34	C			Appreciate the unique histories, cultural issues, and sensitivities when discussing genetic issues with indigenous peoples
				Genetic screening
35	B			Describe genetic screening programs, in particular newborn screening
36	B			Describe the meaning of false positive and false negative genetic screening results
37	B			Understand the difference between targeted group screening programs and coordinated public health screening programs (e.g. carrier screening, genetic screening for adult onset conditions)
				ABILITY
				Consultations and family history
38	B			Conduct patient interviews to compile a full and relevant family history
39	B			Construct a standard pedigree with symbols to document family history
40	B			Measure genetic risk of individuals in pedigrees demonstrating Mendelian inheritance
41	B			Differentiate between patterns of autosomal, sex-linked, mitochondrial and multifactorial inheritance
42	B			Explain to a patient the significance of their family history in order to estimate their chances of heritable disease
43	B			Appreciate the potential of family history information to reveal unexpected family relationships such as consanguinity or misattributed paternity
44	B			Critique/interpret a family history suggestive of complex inheritance
45	B			Recognise variable penetrance
46	B			Describe genetic risk/polygenic risk scores for individuals in pedigrees demonstrating complex inheritance
47	D			Demonstrate how to perform an adult or paediatric clinical examination, recognising the clinical features of common Mendelian diseases, common chromosomal disorders, dysmorphic syndromes, malformation syndromes, and/or clinical indicators that suggest an inherited predisposition to cancer
				Seeking information and referring
48	C			Explain when and how to refer individuals to clinical genetic services
49	C			Identify sources of information on genetic disorders and gene variant interpretation to aid with patient management
50	D			Identify and demonstrate an ability to apply the most recent national and international guidelines to manage patients with genetic conditions
				Ordering and using genetic/genomic tests
51	B			Appropriately apply the different types of genetic tests (diagnostic, predictive, test for carriers)
52	B			Demonstrate an ability to understand the results of genetic tests ordered and their clinical implications
53	B			Identify or facilitate identification of patients who may benefit from genomic testing

	DOMAIN	SET	LEVEL	COMPETENCY
54	C			Discuss with patients the benefits and risks of, and alternatives to, genetic/genomic testing
55	B			Order, interpret, and communicate the results of genomic tests, within scope of practice
56	B			Ensure that tissue biopsy procedures are coordinated to make certain that appropriate and sufficient material is obtained for testing
57	B			Maintain a dialogue with the clinical laboratory to ensure that the appropriate test(s) are ordered and interpreted in the context of the patient's clinical status
58	D			Recognise utility of testing multiple family members (segregation studies, trio testing, maternal lines for X-linked disorders)
59	B			Use appropriate repositories of information to aid interpretation of genomic test results
60	B			Recognise the utility of reanalysis and review of previous genetic/genomic results in light of new information
				Genetic/genomic test results
61	C			Demonstrate how to coordinate the information obtained from different sources (personal and family history, examination, investigation) into a coherent and rational action plan for genetic disorders
62	B			Critique the clinical impact of genetic variation on risk stratification and individualised treatment
63	B			Describe how genotype can inform medical or surgical management (e.g., pharmaceutical intervention, prophylactic surgery)
64	B			Explain how genomic testing can be used to guide the choice of medical and/or surgical therapy
65	C			Recognise that genomic results may have implications for other family members
66	D			Identify appropriate genetic support for the patients and their family in light of test results
67	C			Make appropriate referrals to specialists and other health providers and support the patient in ongoing care
				Genetic/genomic screening in public health
68	B			Recognise the differences between genetic testing and screening
69	C			Order appropriate genomic screening investigations in high-risk populations
70	B			Critique genomic screening results and develop an appropriate management plan
				Communicating genetics and genomics
71	C			Demonstrate how to communicate genetic information to patients in a respectful, compassionate, understandable, non-directive manner, being aware of the impact genetic information may have on an individual, family and society
72	C			Explain genetic information, the concept of genetic risk, susceptibility and influence of genetic factors on maintenance of health and development of disease to patients
73	C			Explain genetic health care options to patients without coercion or inflicting personal biases
74	C			Demonstrate how to explain the process of tissue sample collection and analysis to patients
75	D			Ensure that undergoing genomic testing is a joint decision of the patient and the physician
76	B			Explain the benefits and limitations of somatic genomic testing to the patient, including implications regarding treatment of the condition and clarification of their prognosis

	DOMAIN	SET	LEVEL	COMPETENCY
77	C			Explain the potential psychological and social impact of identifying genetic status in a child / young person / asymptomatic adult and their relatives
78	C			Discuss the possibility of incidental or secondary findings and how they will be managed
79	C			Explain to the patient issues of costs and financial coverage of genomic testing
80	C			Demonstrate the ability to obtain informed consent for genomic testing, including explaining to the patient how it will be stored and who will have access to it
81	B			Discuss with the patient the importance of genomic testing of their tissue sample, including potential implications for treatment and prognosis, and the limitations of genomic testing
82	A			Explain that interpretation is based on current knowledge and may change in future as new gene-disease information becomes known
				Communicating results and follow-up
83	B			Explain and document findings from genomic testing to the patient, including implications for other family members
84	C			Demonstrate an understanding of the impact of diagnosis of a genetic condition on the individual and the family
85	C			Facilitate access to resources to enhance patient learning about the results of genomic testing
86	C			Demonstrate how to interpret the results of a genetic test reports for patients and their families
87	D			Be able to identify team members' roles and work in a multidisciplinary team to deliver genetic diagnosis and counselling
88	D			Demonstrate an awareness of reproductive genetic counselling issues and options
89	B			Communicate clearly with other medical professionals involved in the care of patients about the therapeutic implications of the genetic information garnered about the patient
90	C			Discuss benefits, risks, and alternatives of preventive and therapeutic approaches that are informed by genomic findings
				Genetic and genomic issues and consequences
91	C			Explain who could have access to a patient's genetic/genomic information
92	B			Explain the implications of when genomic test results are placed into a patient's medical record or care plan
93	C			Demonstrate an ability to be aware of and responsive to patients' concerns about genetic discrimination
94	C			Demonstrate an ability to empathically respond to the various ethical, social, cultural, and religious issues that may arise when dealing with genetic issues of people from diverse backgrounds
				ATTITUDE
95	D			Recognise limits of own knowledge and rapidly changing advances in genomic technologies and medicine (e.g., RNA sequencing diagnostics), refer when appropriate, and participate in continued professional development when necessary
96	B			Remain aware of new genomic testing methods, their clinical applications
97	B			Recognise the potential advantages and pitfalls in ordering a genetic or genomic test
98	B			Recognise potential involvement of multiple organ systems in genetic disorders and therefore appreciate the need to seek appropriate consultation with experts in each field
99	D			Recognise the importance of medical research in clinical genomics

	DOMAIN	SET	LEVEL	COMPETENCY
100	C			Be prepared to refer patients to clinical studies or trials based on genomic test results
				Communication issues in genetics and genomics
101	D			Maintain awareness of the sensitivity of genetic information and the need for privacy and confidentiality while delivering genetic education and counselling
102	C			Acknowledge the needs of the patient as an individual as well as the needs of family members
103	C			Recognise sensitivity of exploring family relationships (such as adoption, paternity and consanguinity) and culturally nuanced topics (such as consanguinity, ethnicity, and causes of death)
104	C			Describe the range of emotional responses individuals and families may experience when confronted with having to discuss heritable health issues
105	C			Recognise that genomic test results can have benefits beyond aiding diagnosis and management. For example, findings may have personal utility for the patient and their family
				Societal considerations in genetics and genomics
106	C			Advocate for genetic health equity
107	D			Maintain nonjudgmental decision making and explanations and respect patient autonomy when discussing genetic information
108	D			Recognise self-discomfort if it arises when a patient's beliefs are discordant with one's own and refer to another practitioner if required
109	C			Recognise the distress an uninformative genomic test result may cause to the patient and or their family
110	D			Recognise when to seek legal or professional advice if/when requested to breech confidentiality
111	C			Maintain awareness of the importance of accurate communication; counselling without coercion or personal bias; and the provision of information appropriate to the culture, knowledge, and language level of the patient
112	C			Maintain awareness of the ethical, social, cultural and religious issues that may challenge clinical care when diverse groups and communities are served, including in including Aboriginal and Torres Straight Islander peoples and Māori

Appendix: Human Genetics Society of Australasia Core Capabilities in Genetics & Genomics for Medical Graduates Working Group Members

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