A Delphi consensus exercise to determine a genetics and genomics curriculum for primary medical degree in Aotearoa New Zealand

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Abstract

Introduction: Genetics and genomics are of increasing importance in diagnosis and treatment of patients. We aimed to determine relevant genetics and genomics curricular content and learning objectives for contemporary New Zealand medical graduates.

Methods: International and national undergraduate medical genetics curricula were identified and learning outcomes collated. Invited New Zealand subject experts (n = 58) contributed further learning objectives, with final pool of 73 learning objectives. A survey-based two-round Delphi process was used to gain consensus on the level of learning for each learning objective. Learning outcomes with consensus for learning greater than "in some depth" or "in detail" were included in the proposed curriculum.

Results: The response rate for the Delphi rounds were 41% (n = 24/58) and 29% (n = 17/58) for Rounds 1 and 2, respectively. Experts reached consensus on retaining 58/60 (97%) of proposed learning objectives that were to be learned to at least "some depth". Learning objectives in interprofessional skills, pharmacogenetics, clinical reasoning and information management were retained but refined. Learning outcomes specifically related to Indigenous health included taking an appropriate genetic history, understanding cultural tenets connected to whakapapa (genealogy), an understanding of DNA samples and genomic data being taonga (sacred), the application of genetic data to Māori and other Indigenous populations and the role of genetics in colonisation and racism and their impact on healthcare.

Conclusions: Learning objectives for contemporary medical genetics curricula should consider including those focusing on Indigenous health. Findings highlight the necessity of timely re-evaluation of medical curricula.

Keywords: genetics; genomics; education; medical curriculum; Indigenous peoples

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Introduction

Genomics is a rapidly advancing area of medicine. It is becoming essential to patient diagnosis and treatment and determining prognosis. The rapid expansion of the field of genomics demands doctors integrate genomic knowledge into their practice to a greater degree than ever before. It is therefore critical to contemporary practice that, by graduation, medical students are equipped with relevant foundational genomic knowledge and the ability to apply this with appropriate patient-centred skills.

The aim of undergraduate genomics curricula is to graduate students with genomic competencies to detect, diagnose, educate and treat patients and to have a deep enough understanding to apply advances in genomic understanding to practice. These competencies must be situated within a wider framework of clinical competencies. Previous studies have shown that while the scientific basis of genetics is widely taught in medical schools, the skills needed to apply the knowledge in practice are not (Plunkett-Rondeau et al., 2015; Telner et al., 2008; Thurston et al., 2007). The American College of Medical Genetics and Genomics proposes that the "best way to assure that non-genetic specialists are educated about genetics and genomics is to make it relevant to the care of their patients" (Bennett et al., 2017, p. 752). Relevance means that genomics curricula must include wider competencies, such as use of decision-making tools, teamwork and patient education with ability to undertake culturally appropriate care. In addition, medical curricula now emphasise Indigenous health, an area not previously addressed in genomics curricula. We aimed to describe a contemporary curriculum in genetics and genomics for primary medical degrees in Aotearoa New Zealand by finding the consensus of experts using a Delphi process.

Methods

The study consisted of a Delphi consensus exercise via email using expert participants in New Zealand. The statements for the Delphi were informed by a review of international literature on undergraduate genetics and genomics curricula, with the addition of relevant subject material from undergraduate medical curricula from both New Zealand medical schools (University of Otago, University of Auckland). A Delphi method was chosen as it confers an advantage compared to the two other main methods of achieving expert agreement (nominal group process and consensus development panel). In this study, an a priori decision was made to limit the Delphi to two rounds. A threshold of ≥ 66% (two-thirds) agreement on a 5-point scale was used to define consensus.

Literature review

To identify genetics and genomics medical school curricula, we searched the MEDLINE, SCOPUS and Google Scholar databases, including the following search terms: "curricula or curriculum", "genomics or human genetics", "teaching", "genetics curriculum medical school", "medical school curriculum", "genetics curriculum", "genetics outcomes medical

school", "genetics objectives medical school", "genetics teaching curriculum medical school" and "medical school curriculum". The search was limited to articles in English published from 1 January 2000 to 1 November 2019. We also checked references to find additional curricula using a snowball approach.

Learning objectives were extracted from curricula published in Australasia (n = 2), Europe (n = 5) and the United States (n = 16) and included objectives recommended for all health practitioners and general practitioners (Alkuwaiti, 2011; Bankier, 2008; Bennett et al., 2017; Burke, Martyn, Stone, et al., 2009; Dhar et al., 2012; Houwink et al., 2011; Hyland et al., 2013; Jenkins et al., 2001; Korf et al., 2004; Magee et al., 2001; Burke, Martyn, Thomas, & Farndon, 2009; Plunkett-Rondeau et al., 2015; Riegert-Johnson et al., 2004; Rubanovich et al., 2018; Salari et al., 2013; Skirton et al., 2010; Talwar et al., 2017, 2019; Telner et al., 2008; Thurston et al., 2007; Tognetto, 2019; Walt et al., 2011; Wolyniak et al., 2015).

Pre-Delphi generation of objectives

A matrix method (Garrard, 2020) was used to organise learning objectives from the literature search together with learning objectives from the two New Zealand medical school current curricula (University of Auckland, University of Otago). The 303 collated learning objectives were grouped into six domains, which were used to organise the MB ChB curriculum at University of Otago at the time of this Delphi: medical science, professional practice, clinical skills, diagnostics and therapeutics, Hauora Maori (Indigenous health) and population health. The 303 objectives were then thematically analysed by a geneticist (MT), an educator (DK) and a student (NK). This was done by these authors meeting in person with learning objectives printed on individual pieces of paper. The team carefully read the 303 learning objectives as grouped in the six domains. Learning objectives that were conceptually similar were grouped. Then learning objectives that addressed the same learning were aggregated and the most comprehensive retained and duplicates discarded. Next, the groups were aggregated into similar areas of learning or themes. This process of consolidation resulted in 109 objectives, which were organised into 13 themes (Table 1). This list was provided by email to Delphi exercise participants, who were asked to add learning objectives that they thought were missing and valuable, and a further 35 were nominated. Free-text comments about the learning objectives were invited, and learning objectives receiving free-text comments were refined accordingly by the lead author (DK). Of these 35, eight were in the domain of Hauora Māori, which had not had any representation from learning objectives in curricula in published literature. The Hauora Māori objectives were formulated by the Otago Hauora Māori curriculum domain group, composed of leading Māori health academics from Te Ika-a-Māui (North Island) and Te Waipounamu (South Island). The research team considered the 144 potential learning objectives, eliminated repeats and substantial overlaps, and consolidated this to 73 learning objectives for consideration in the Delphi process.

Table 1

The Themes Generated by the Pre Delphi Literature Review and Consultation

Structure and function of the human genome
Genetics concepts, including inheritance
Mechanisms leading to genetic mutation
Carcinogenesis
Research, scholarship and continuing professional development
Communication skills within the doctor–patient consultation
Conducting culturally appropriate and sensitive consultations
Clinical skills and clinical reasoning
The function and role of team members within interprofessional teams
Population genetics
Pharmacogenomics and therapeutics
Laboratory tests
Ethics

Delphi expert panel

Broad definitions of "expert" have been proposed, with a general agreement that an expert is "someone who possesses the relevant knowledge and experience and whose opinions are respected by fellow workers in their field" (de Villiers et al., 2005, p. 640). Delphi participants were selected using the two following criteria, with both required for invitation to participate. First, the person had professional expertise in medical genetics/genomics, or closely related fields. The scope of this criteria included works as a clinical geneticist or genetic counsellor, works in another medical role or clinical laboratory scientist role that uses/critically requires medical genetics/genomics, works as a medical researcher in a field that critically requires medical genetics/genomics, teaches genetics to medical students and/or has curriculum expertise in genetics. Second, the person had direct knowledge of/involvement in the learning of New Zealand medical students in the fields of medical genetics/genomics and had voiced an interest in improving the New Zealand medical genetics/genomics curriculum. On the basis of these criteria, 58 people were invited to participate in the study based on the research team's networks and invitations disseminated through the University of Otago Medical School, University of Auckland's Faculty of Medical and Health Sciences and Genetic Health Service New Zealand. Broadly, these people were categorised as being either clinical geneticists, scientists or clinician educators. This study received ethical approval from the University of Otago Human Ethics Committee (Reference D18/397).

Delphi survey structure

The Delphi was administered to experts using the Qualtrics (Qualtrics International Inc., Provo, UT) online survey platform. Participants were asked to rate the depth to which

each learning objective should be learned by graduating medical students. A Likert-type scale with five categories was used, with descriptors for level of learning including (1) superficially, (2) in little depth, (3) in some depth, (4) in detail and (5) in great detail. A 5-point scale was chosen to allow sufficient fidelity in categorising learning objectives without overwhelming participants with too many arbitrary options (Thangaratinam & Redman, 2005). In the second round of the Delphi, participants were provided with anonymised summary data from the previous round, containing a comment regarding why the learning objective may not have reached consensus or explaining the meaning and context of the objective. This was included to limit the effect of question ambiguity as a potential source of bias.

Analysis of Delphi ranking data on learning objectives

Data were entered into a spreadsheet (Microsoft Excel, Microsoft Corp., Palo Alto, CA), where frequencies and percentages for each learning objective were calculated. The mean and standard deviation for participant Likert responses for each learning objective were calculated after each round to check if any participants consistently gave very high or very low scores. Consistently high- or low-rating participants would be eliminated from the analysis to minimise undue influence from these participants. Learning objectives that reached consensus for inclusion, defined as those that were rated "3, 4 or 5" (to be learned "in some depth", "in detail" or "in great detail") by ≥ 66% of participants, were not included in the second round of the Delphi. Similarly, those that were rated "1 or 2" (to be learned "superficially" or "in little depth") by ≥ 66% of participants were excluded from the second round and were eliminated from the final list of learning objectives. Mean and median scores for ranking of objectives were compared between expert categories using frequency distribution tables.

Results

Delphi responses

In total, 24 experts (41% of n = 58 invited) participated in the first Delphi round, comprising 10 scientist/researchers, eight clinical geneticists and six "other" clinician educators. The response rate for the second round of the Delphi was 29% (17/58) of invited and 71% (17/24) of first-round participants. No participants consistently rated statements high or low. There was no difference in mean score or standard deviation between clinicians and non-clinicians.

Learning objectives

After the first Delphi round, three quarters (78%, 57/73) of learning objectives reached a consensus that they should be learned to "some depth" or greater. Eleven learning objectives reached consensus that they should only be learned to "little depth" or "superficially" and were eliminated. Five learning objectives had not reached consensus and entered the second Delphi round.

In the second Delphi round, one learning objective reached consensus that it should be learned by undergraduate medical students to "some depth" or greater. Two learning objectives reached consensus that they should be learned at "little depth" and were eliminated. The remaining two learning objectives did not reach consensus. One of these, "understands how common micro-organism genetics affects natural history, response to therapy (resistance) and prevention strategies", was decided by the participants as being better suited to another curriculum area, such as microbiology. After two Delphi rounds, 52 learning objectives reached consensus for inclusion in the curriculum, with 97% consensus (Table 2).

Table 2

Consensus Curriculum After the Delphi Process Was Complete

Biomedical Science

Understand and apply in practice knowledge about the structure of genes and the human genome

- Structure of genes, including the non-coding regions
- The nature and types of genetic variation and benign polymorphisms
- The mitochondrial genome
- X-inactivation

Understand and apply in practice knowledge about DNA replication, repair and recombination

- DNA replication and repair
- Meiosis, crossing over, recombination and linked genes

Understand and apply in practice knowledge about the gene expression

- One gene can give rise to many proteins
- Regulation of gene expression
- Role of epigenetics in gene expression

Understand and apply in practice knowledge about the patterns of inheritance and calculating risk

- Inheritance patterns and risk calculations in common Mendelian diseases (autosomal dominant/recessive, X-linked dominant/recessive and mitochondrial)
- For complex (multifactorial) medical conditions, interpret polygenic risk scores and use empirical risk figures to provide appropriate information

Understand and apply in practice knowledge about genetic mutation

- Mechanisms leading to genetic mutations (mis-sense, insertion, deletion, duplication, micro-deletions and micro-duplications), including those in meiosis (chromosomal translocations, inversions non-homologous chromosomal crossover)
- Environmental causes of mutation (radiation, chemical) and the effects of accumulation of mutations
- Principles of teratogenesis and the role of genes and teratogens (including alcohol, drugs and infection) in congenital anomalies

Understand and apply in practice knowledge about the genetic and phenotypic heterogeneity in genetic disorders

- Factors that affect the phenotypic expression of genetic disorders: penetrance, expressivity, sex limited inheritance, genomic imprinting, co-dominance, x chromosome inactivation, anticipation, gene dosage

Understand and apply in practice knowledge about the genetic testing and pharmacogenomics

- Innate metabolic errors, pharmacogenetic variations and their general clinical manifestations

Understand and apply in practice knowledge about population genetics

- History of genetics, impact of eugenics, racialised thinking and the use of pseudoscience in racial oppression and discrimination
- Critique biological genetics principles (e.g., genetic drift, founder effect) and their impact on interpretation of genetic variation in and between human populations
- Genetic diversity in humans and the abundance of genetic variants in each individual genome

Understand and apply in practice knowledge about the genomics of neoplasia

- Cancer aetiology: genetic principles including role of environment, familial cancer, epigenetics, genetic testing
- Cancer pathogenesis: molecular basis including proto-oncogenes and tumour suppressor genes, errors in the DNA mismatch repair genes, dysplasia to carcinoma sequence
- Cancer treatment: genetics and genomics in the diagnosis and treatment of cancer

Understand and apply in practice knowledge about the genomics of core conditions

- Genomic basis of common conditions
- Basic principles of innate metabolic errors, pharmacogenetic variations and their general clinical manifestations

Interpret advances in genetic research in a clinical context and apply them appropriately to patient care

Understand the health informatics challenges of new genomic technologies, such as data storage and re-analysis and national patient management systems

Professional Practice

Commitment to life-long learning and self-improvement

- Maintain competence in practice by evaluating learning needs in genomic medicine and addressing these

Decision-making tools

- Access and use specialist genetic databases and guidelines in the diagnosis and treatment of genetic and familial disorders

Interprofessional teamwork and team functioning

- Collaborate with different clinical genetics professionals in patient care and refer appropriately

Interprofessional communication

- Communicate family history and medical history pertinent to genetics with an interdisciplinary team of healthcare professionals

Ethical challenges specific to genetic testing

- Ethical principles of genetic testing and use of genetic information (e.g., Te Ara Tika: kaitiakitanga, tapu, rangatiratanga, mana, manakitanga, atuatanga; beneficence, non-maleficence, justice and autonomy).
- Implications and ethics of somatic and germline genetic modifications associated with emerging therapeutics (CRISPR, gene therapy, stem cell therapy, etc.)

Clinical Skills

History-taking skills

- Take a family history, using this to construct and interpret complex pedigrees of at least 3 generations using standard nomenclature

Examination skills

- When performing an adult or paediatric clinical examination, recognise the clinical features of common Mendelian diseases, common chromosomal disorders, dysmorphic syndromes and malformation syndromes
- When performing an adult or paediatric clinical examination, recognise the clinical features of common genetic cancer syndromes and clinical indicators that suggest an inherited predisposition to cancer

Communication skills within the doctor–patient consultation

- In a genetics-focused consultation, respect the patient's religious, cultural, social and ethical beliefs and act in accordance with these values
- When taking a family history, asks sensitively about culturally nuanced topics such as consanguinity, ethnicity and causes of death
- Apply the concepts of whakapapa and whanaungatanga when taking a genetic history

Ethical, legal and professional obligations

- Apply legal obligations that are relevant to genetic testing (e.g., consent, authority, insurance coverage and employment status, rights and custodianship for undertaking genetic testing and having access to test information)

Effective communication

- Communicate genetic information in a respectful, compassionate, understandable, non-directive manner, being aware of the impact genetic information may have on an individual, family and society
- In a genetics-focused consultation, respond appropriately to the emotional responses of patients
- Psychological and social impact of identifying genetic status in a child / young person / asymptomatic adult and their relatives

Consent

- Obtain age-appropriate consent and proxy consent for procedures and genetic testing

Diagnostics and Therapeutics

Clinical reasoning

- Coordinate the information obtained from different sources (history, physical examinations, biochemical, cytogenic, genomic, molecular) into a coherent and rational action plan for genetic disorders

Advise patients

- Advise patients on diagnostic testing, predictive testing and screening (including carrier screening and targeted group screening) and their respective value in genetic (core) conditions, including those detected prenatally.

Genetics testing

- Use laboratory tests to identify and characterise suspected (core) genetic disorders

Appropriate, safe, timely and effective testing

- Order appropriate genetic tests on relevant material on the right patient at the right time, with informed consent and with appropriate clinical information for the lab
- Concepts of analytic validity, clinical validity and clinical utility as they relate to genomic testing

Inclusion of the patient and appropriate explanation

- Discuss with patients the indications for genomic testing, specifically the benefits, risks and alternatives.
- Interpret results of genetic and genomic tests and laboratory reports and explain them to family members (including interpreting cytogenetic and molecular data, sequencing data and direct-to-consumer tests, especially assessing the probability of error and uncertainty in variant effect)
- Interpret genome sequencing results in the context of population genetics

Variance in dosing and clearance

- Apply pharmacogenomics and pharmacogenetics when prescribing and understand their role in the response of individuals to certain pharmacological agents
- Understand variants affecting drug responses found in a patient may also have implications for other family members

Hauora Māori

- Recognise race as a biological, yet socially constructed, category
- Recognise the origins of biological race, critique scientific naturalistic explanations and discuss human variations
- Recognise the political implications of biological categorisation as well as the scientific biases inherent in social construction
- Recognise the importance of context in the study of genetics: colonisation, racism, white supremacy, family histories and impact on healthcare
- Apply the relationships and concepts of whakapapa within genetic research
- Understand Māori responses to genetic research
- Understand inherited vs population genetics as theoretical constructs and their impact on Hauora Māori.
- Understand the role of genetic research in reinforcement of racist ideologies and current health inequities

Population Health and Epidemiology

Communication of key concepts

- Communicate and translate information to the public about genetics and genomics to support community understanding
- Critically reflect on the impact of the application and extrapolation of genetic data and the potential for misrepresentation of Māori and other Indigenous populations
- Understand difficulties in applying population genetics to individual health, including pharmacogenetics
- Critically appraise the potential harm of genetic research on communities (e.g., warrior gene)

Principles and practice of screening programs

- Local and national antenatal, newborn and population genetic disease screening programs: knows why they were developed and the factors that are important for their success

Discussion

New Zealand experts with experience relevant to medical curriculum or genetics and genomics reached consensus to include 52 learning objectives with indicative levels of learning in a medical degree curriculum in New Zealand. The learning objectives include novel objectives in Māori (Indigenous) health. Medical school genetic curriculum guidelines from around the world—including Australasian genetic guidelines for medical school graduates (Bankier et al., 2008)—have not included learning objectives related to Indigenous people and genetics/genomics previously. The addition of Māori health models, concepts and approaches is a small but vital step for addressing the current inequity in health outcomes experienced by Māori in New Zealand (Came et al., 2013; Phillips et al., 2017; Russel et al., 2013). Learning specifically how to engage appropriately with Indigenous culture in genetics-based consultations is important in achieving equitable health outcomes and aiming to provide culturally safe care.

The medical sciences, professional practice, population health and epidemiology, and diagnostics and therapeutics objectives are similar to previous curricula. The learning objectives under the "medical sciences" theme reflect what is traditionally taught as

the scientific basis of genetics. Neoplasia objectives are defined more specifically than previously, most likely reflecting the rapidly expanding number of mutation defined therapies coming into clinical practice. Learning objectives under the "professional practice" theme have an expanded focus on communication in the proposed curriculum, in addition to considerations of legal and ethical obligations.

More specific clinical skills learning objectives were nominated than in previously published curricula. Some "clinical skills"-based learning objectives are widely included in previous curricula, such as taking a family history and constructing a pedigree. The proposed learning objectives include wider adult and paediatric patient consultation skills and appropriate referral methods.

Practicality is an issue to consider when implementing new learning objectives. Curricula are full, and content must be removed in order to include more relevant material. The material removed by the expert panel related to specific laboratory methods and detailed aspects of cell biology. However, the expert consensus set of learning objectives may still be too comprehensive to incorporate into medical school curricula. Some of the objectives still overlap and may benefit from further consolidation to generate an implementable curriculum. Furthermore, learning objectives were included if there was expert consensus that the appropriate level of learning for the objective was "in some depth", "in detail" or "in great detail" and removed if consensus that appropriate level of learning was "superficial" or "in little depth". Definition of these Likert descriptors was not provided and was left to the interpretation of each expert, so the included learning objectives should be viewed as consensus on relative importance being sufficient for inclusion in a medical curricula. This consensus exercise did not seek to determine the details of the knowledge, skills or attitudes to be learnt, nor how acquisition would be measured. These details would be important in next steps of curriculum development and seem likely to also be a level of detail to be determined by the medical program itself. Future curriculum consensus exercises should carefully consider if further defining levels of learning is relevant or necessary for their objectives.

There may have been a limitation in the scope of the learning objectives generated for the Delphi, however feedback on acquiring additional objectives was requested from expert Delphi members to address this. Consolidation of objectives was also performed by three individuals, including a third-year medical student (NK), an anatomic pathologist and associate dean learning and teaching (DK) and a geneticist (MT). The small number of individuals involved in this process may have been a source of bias. The modest response rates of contributing experts (41% in first round and 29% in the second round) may also have contributed bias, however there were contributors from all three expert categories, which may balance opinion, and there was no difference between the mean scores on objectives between expert categories. The lack of definition of levels of learning, which was left to the interpretation of the experts may have led to differences in ratings by experts. This is a potential shortcoming of our work. There were only two Delphi rounds,

however most decision making occurs between the first and second round, and deciding a priori prevents participant fatigue and provides a definite endpoint for participants (Hasson, 2008). Finally, the consensus definition may have affected results, both in terms of using level of learning for inclusion in curriculum and 66% agreement in three categories as consensus. Delphi studies have a large range of acceptable participant agreement (between 50–80%) (Hasson, 2008), and a consensus of 66% was, therefore, within normal boundaries.

Conclusion

Using the consensus of opinions of scientist experts and genetics clinicians and drawing from previously published medical genetics curricula from around the world, we generated a set of curated learning objectives for medical genetics and genomics. Some were novel, particular those relating to Indigenous health. These learning objectives are comprehensive, and it remains to be seen how they are best implemented in an undergraduate curriculum. In particular, the exact content, level of demonstrated learning and how to best support students to achieve these will need to be addressed.

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