



May 2020



Genomic competence: workforce development, engagement and education in NZ

Dr Michelle Thunders, PhD
University of Otago, New Zealand



Global Genomic
Medicine Collaborative

New Zealand



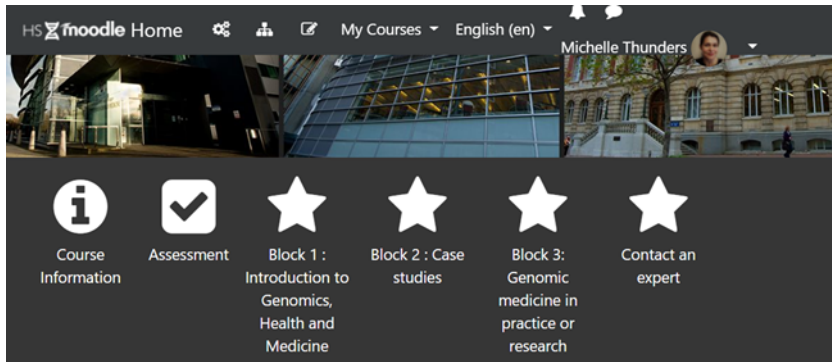
- Population 4,951,500
- Health system mainly funded from general taxation
- \$16.142 billion in 2016/17
- **2 Medical schools in New Zealand**; Eight Universities
- Indigenous population Maori, ~14%
- Health inequity- Maori **health** status is demonstrably poorer than other New Zealanders

Genetics workforce and Genomic Medicine in NZ

- Genetics Professionals and Training (3 Hubs)
 - Clinical Geneticist
 - Genetic Counsellor- currently no training in NZ, Australia only.
- UG/PG Medical Teaching
 - UG- ELM and ALM, Curriculum map and core conditions
 - PG- target GPs, Pharmacists, Health care practitioners
- MyDNA- Pharmacogenomics
- Genomics Aotearoa
- Genomic database NZ
- PHG Foundation - The \$37.8 million (NZ) Precision Driven Health Care Partnership



PG Cert Genomic Health and Medicine



The screenshot shows the Moodle course interface. At the top, there are navigation links for 'Home', 'My Courses', and 'English (en)'. A user profile for 'Michelle Thunders' is visible. Below the navigation, there are six main menu items, each with an icon and a text label: 'Course Information' (info icon), 'Assessment' (checkmark icon), 'Block 1: Introduction to Genomics, Health and Medicine' (star icon), 'Block 2: Case studies' (star icon), 'Block 3: Genomic medicine in practice or research' (star icon), and 'Contact an expert' (star icon).

PG Health Professionals:

- Student led
- Relevant
- Distance
- Practical and relevant

← Assessment

Block 2 : Case studies →

Block 1 : Introduction to Genomics, Health and Medicine

Block 1 overview - Weeks 1-4

In the last decade the knowledge of the human genome, and the associated impact of genetic mechanisms on



Patterns of inheritance



To interpret family histories you need to be able to recognise patterns of inheritance. The tables below remind you of the approach to analysing family pedigrees. They are also in the notes (the book symbol in the header bar) for easy reference.



Here is a pedigree tool you might find useful

Yr5 Med students: Genomic Medicine

- Competency based
- Skills focused
- Case based, NZ relevant
- Tool kits of resources

Mode	Features
AD	Vertical transmission through generations Male to Male transmission possible Males and Females equally affected Offspring risk is 1 in 2 for an affected parent Reduced penetrance and variable expression
AR	Usually only members of one sibship affected Males and females equally affected Positive association with parental consanguinity Offspring risk is 1 in 4 for carrier parents, very low for affected parent
XD	Transmitted by females to sons and daughters Transmitted by males only to daughters No male to male transmission Both sexes affected, girls more often than boys Some conditions lethal in males
XR	Generally only males affected Offspring risks for a carrier female are 1 in 2 for an affected son and for a carrier daughter Offspring risk for an affected male is that all daughters are carriers No male to male transmission
M	Only transmitted by females with offspring risk of up to 100% dependent on homoplasm/heteroplasm Both sexes usually equally affected
Chr	Carriers of balanced rearrangements can have affected children due to unbalanced chromosome complements, so pedigree can contain apparently randomly affected individuals of either sex

Observation	Implication
Affected members in different generations Affected members in only 1 sibship Parental consanguinity Only males affected More females affected than males Male to male transmission Males have no affected descendants	Unlikely to be autosomal recessive Likely to be autosomal recessive Likely to be autosomal recessive Consider X-linked inheritance Consider X-linked dominant inheritance Probable autosomal dominant inheritance Cannot be X-linked Consider Mitochondrial inheritance



Barriers and solutions

- Inequities
- Funding
- Geography
- Population size
- Perceptions and beliefs
- Adapting to Lockdown
- Inclusivity
- Consultation
- Collaboration
- Self sufficiency
- Realism
- Online learning

