Precision Health

Unlocking the power of genomics and AI to improve patient care

James Oughton – Chief Advisor Precision Health (Genomics and AI)

Te Kāwanatanga o Aotearoa
New Zealand Government

MINISTRY OF HEALTH
MANATŪ HAUORA

Background


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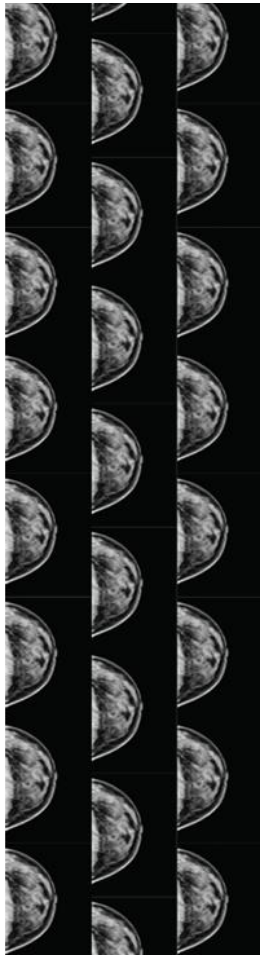
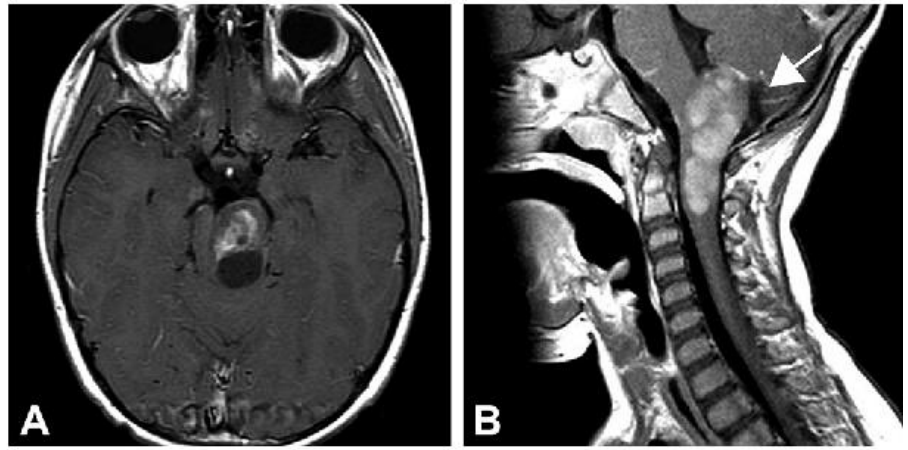
Te Kāwanatanga o Aotearoa
New Zealand Government
MANATŪ HAUORA
MINISTRY OF HEALTH
Te pae tika:
e tūhura ana i ngā ara hou me ngā ārai ki te āta matapaetanga, te kauparenga atu, te kitenga me te rongoātanga o ngā take hauora ki Aotearoa
Precision health:
exploring opportunities and challenges to predict, prevent, diagnose, and treat health needs more precisely in Aotearoa New Zealand
Long-term insights briefing
August 2023

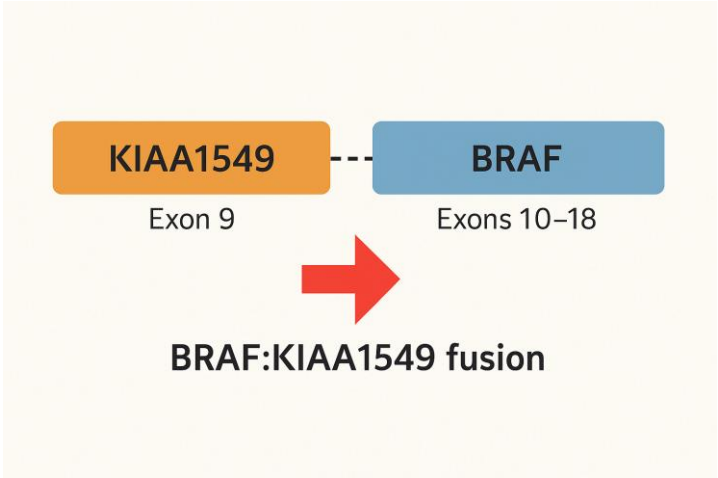

Capturing the benefits of AI in healthcare for Aotearoa New Zealand
A rapid report from the
Prime Minister's Chief Science Advisor
Kaitohutohu Mātanga Pūtaiao Matua ki te Pirimia
Full report
October 2023



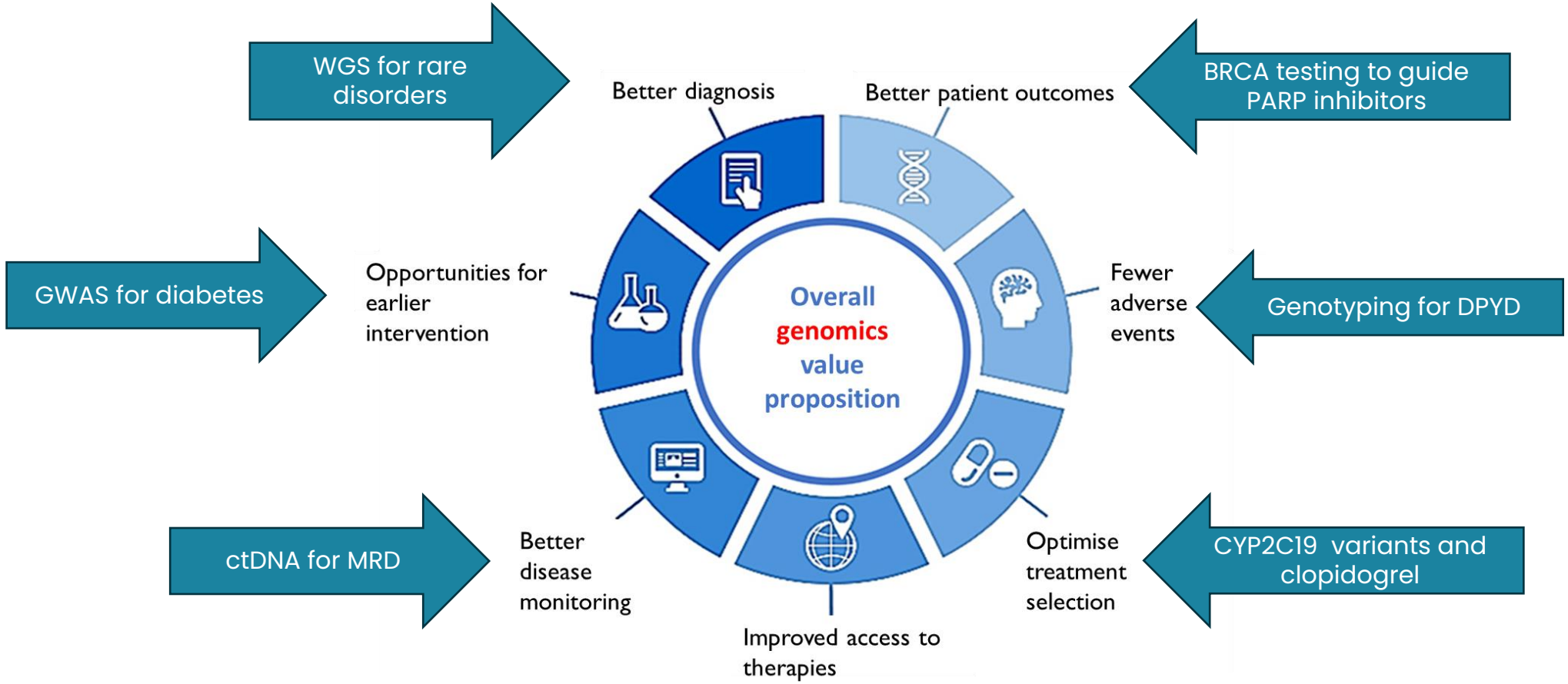
Peter's Story

- In 2020 Peter was a happy and healthy 3-year-old……
- Progressive left-sided weakness
- Drooling – urgent MRI
- Unresectable pilocytic astrocytoma
- Specimen obtained –DNA/RNA sequencing (400 genes/250 genes)





Integrating genomics into healthcare has huge potential



Genomics in healthcare – what's holding us back?

We are lacking a consistent way to systematically evaluate and safely deploy genomics in a wide range of health settings

Issue or Challenge	Example
Genomics services in New Zealand not yet equipped to enable precision health	National Genetic Health Service currently focused and resourced to only deliver a core set of basic genetic tests
Adopting international standards or guideline-driven care	*DPYD genotyping*: international standard of care genetic testing not consistently available, 'post-code lottery' for testing
Return on investment hard to quantify	Unclear where government wishes to invest, lack of robust HTA for genomic testing leading to little or no investment
Insufficient / outdated legislative and regulatory frameworks	Medicines Act 1981 – 40 years old, never envisaged regulating widespread genetic testing, let alone possibly gene editing therapies
Genomic data availability, collection, storage, usage and sharing	Concerns re: privacy, genetic discrimination, fit with Te Tiriti – e.g. tissue and data as a taonga, lack of representative genomic databases that reflect NZ's diverse population, no infrastructure for sharing genomic data
Workforce constraints	Lack of clinical geneticists , genetic counsellors, genetic pathologists, genetic scientists and bioinformaticians (and options for training and education pathways)

Precision Health (Genomics and AI) Key Pillars

Leadership and stewardship

- Establish governance and principles to guide our work
- Leverage international connections = 'fast follower'

Patient rights and safeguards

- Explore policy options to protect patients
- Ensure nationally consistent, equitable access

Enablers

- Understand gaps (people, parts and processes)
- Work towards building what's missing (i.e. Health Technology Assessment)

Case studies

- Highlight ongoing progress in genomics and AI
- Explore adoption of these case studies (alongside investment)

Engagement and Social License

- Build trust via comms + engagement
- Engage with stakeholders across the sector

*Illustrative examples only - not an exhaustive list

2025 Genomics Work Programme:

Improving the health of all New Zealanders by enabling more efficient, effective and precise healthcare

Leadership and Stewardship

- Establish aims, principles + agency roles
- Leverage international connections - Australian Genomics, GA4GH

Patient rights + safeguards

- Genetic discrimination
- Data privacy, storage and interoperability
- Advocate for nationally consistent DPD testing
- Establish IMER

Enablers

- Establish HTA assessment process (alongside Pharmac and Health NZ)
- Align with HNZ genomic strategy
- Complete genomics maturity assessment
- Develop 'Whole-of-system' genomic implementation roadmap

Case studies*

Examples:

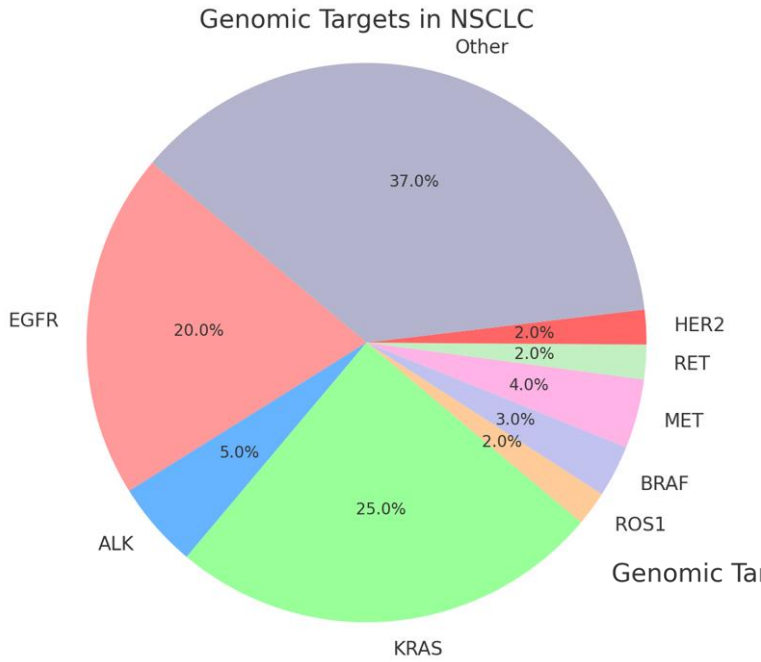
- Nationally consistent NGS for late-stage NSCLC and mCRC (biomarker funding)
- He Kakano - mapping the NZ Variome to personalise patient care
- OMICO: explore potential precision oncology partnership

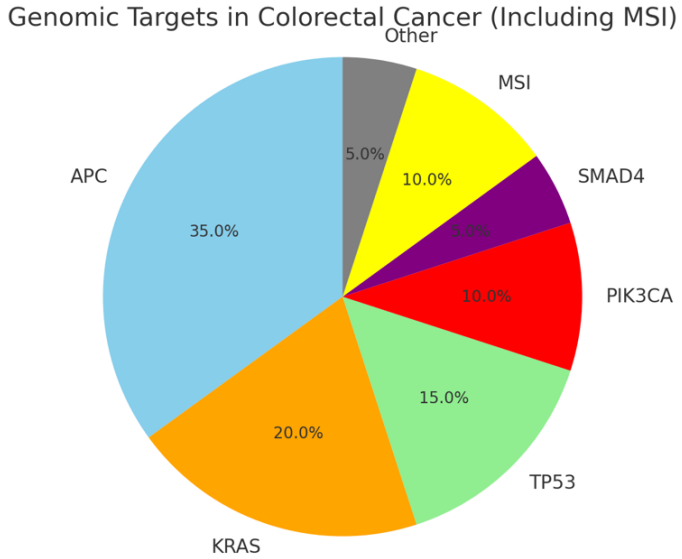
Social License and Engagement

- Build trust via comms + engagement strategy (e.g. Ministry website, press releases, consultation)

Illustrative examples only - not an exhaustive list

Unlocking Access to Standard of Care Testing That NZ Cancer Patients Expect





- Cancer is a disease of the genome - genetic mutations drive cancer
- Many cancers have targeted therapies that work on specific genetic mutations
- Diagnostic tools are critical to find these mutations and enable precision oncology
- Rather than use 1 test to detect 1 gene, NGS uses a broad panel to test many genes at once
- For NSCLC: gene testing must cover ALK, ROS1 and EGFR (with more to come)
- For mCRC: gene testing for must cover RAS, BRAF and MSI (with more to come)
- Multi-gene panels may be cost effective vs. sequential, single-gene testing in NSCLC and approaching cost-neutral in mCRC
- Getting patients on the right therapies (or avoiding them) early in their journey improves outcomes for cancer and is cost-effective

But it's not that simple……

- Significant work needed to:
  - Address workforce and infrastructure needs
  - Create a national test directories with clinical pathways
  - Consolidate testing in a small number of labs
  - Other work underway laying groundwork for nationally-consistent testing

Global Alliance for Genomics & Health
Collaborate. Innovate. Accelerate.

Omico.

Equitable access to new tech is challenging

Financial Sustainability

Technology responsible for 25-50% of cost growth annually

Current Processes ad hoc

Unplanned, haphazard & unsustainable approach to adopting new technologies

Unwarranted Variation & Inequity

Where patients have access in some areas but not others

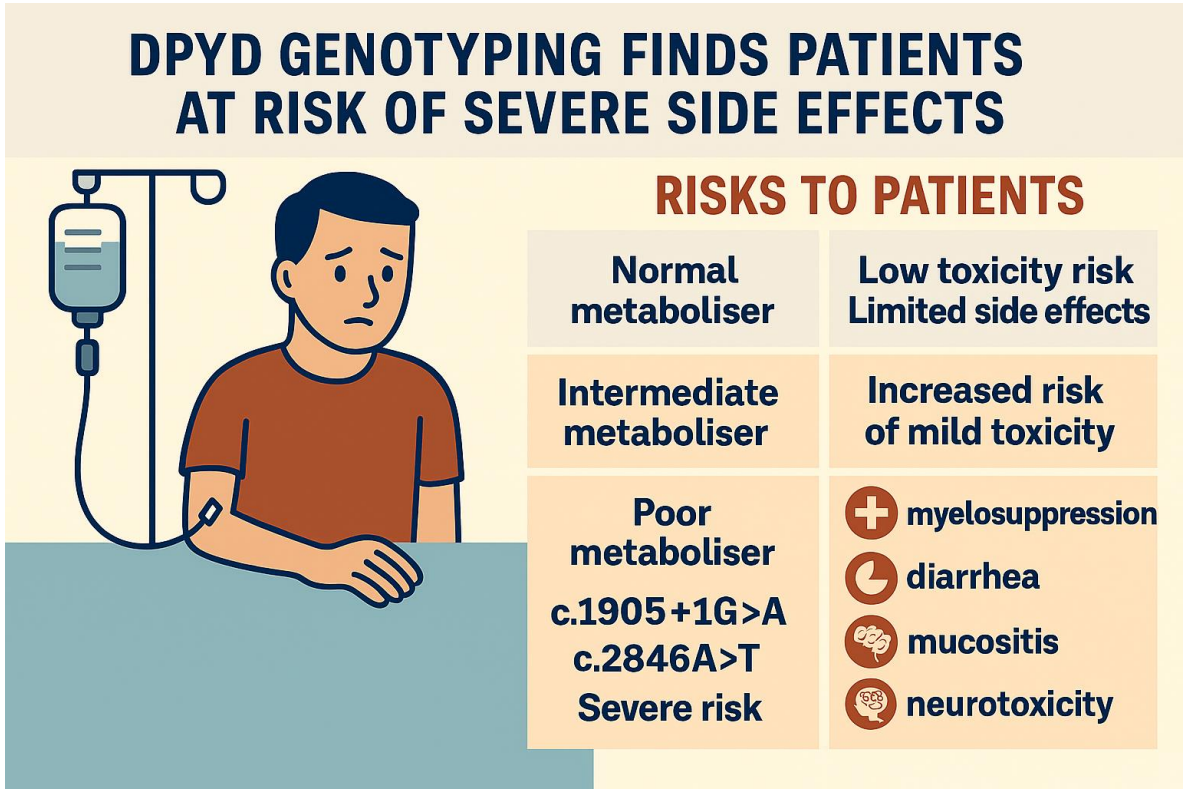
Health Technology Evaluation Pathway (HTEP)

Tests, devices, technologies. Not services

*"A single, nationally-consolidated, systematic process for triaging, **assessing**, and appraising **health technologies** to inform their adoption and use within the New Zealand **public** health system."*

Safety, efficacy, effectiveness, cost, cost-effectiveness, equity, sustainability

Case Study: DPYD Genotyping



- DPYD genotyping saves lives and prevents hospital admissions
  - *Could it be a cost effective intervention?*

- Application to HTEP
- Collaboration with Te Aho o Te Kahu, Health NZ
- Health economic assessment
- Positive outcome – technology is cost-effective and may approach cost-saving
- Currently with Health NZ for national implementation

Genetic Discrimination – Insurance

Health champions claim victory in 'genetic discrimination' fight

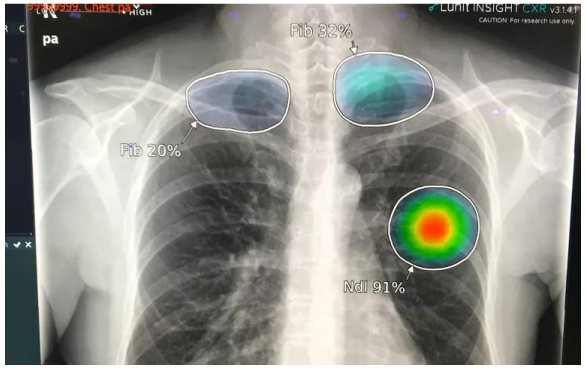
Health workers urge regulation of genetic discrimination

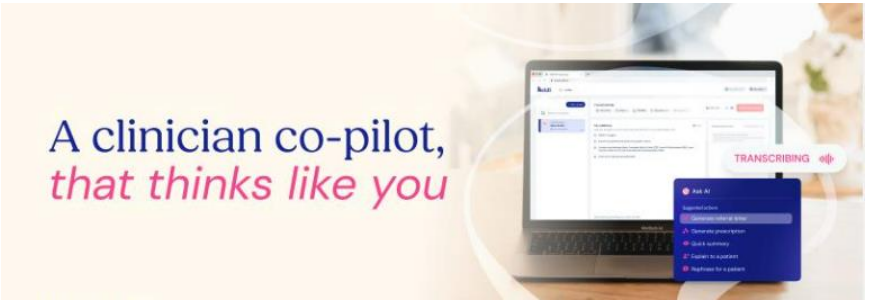
Insurers want to self-regulate their access to genetic test results. Advocates want a total ban

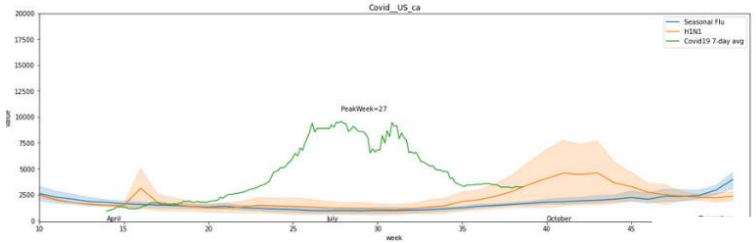
Artificial Intelligence is EVERYWHERE

Opportunities for AI in Healthcare

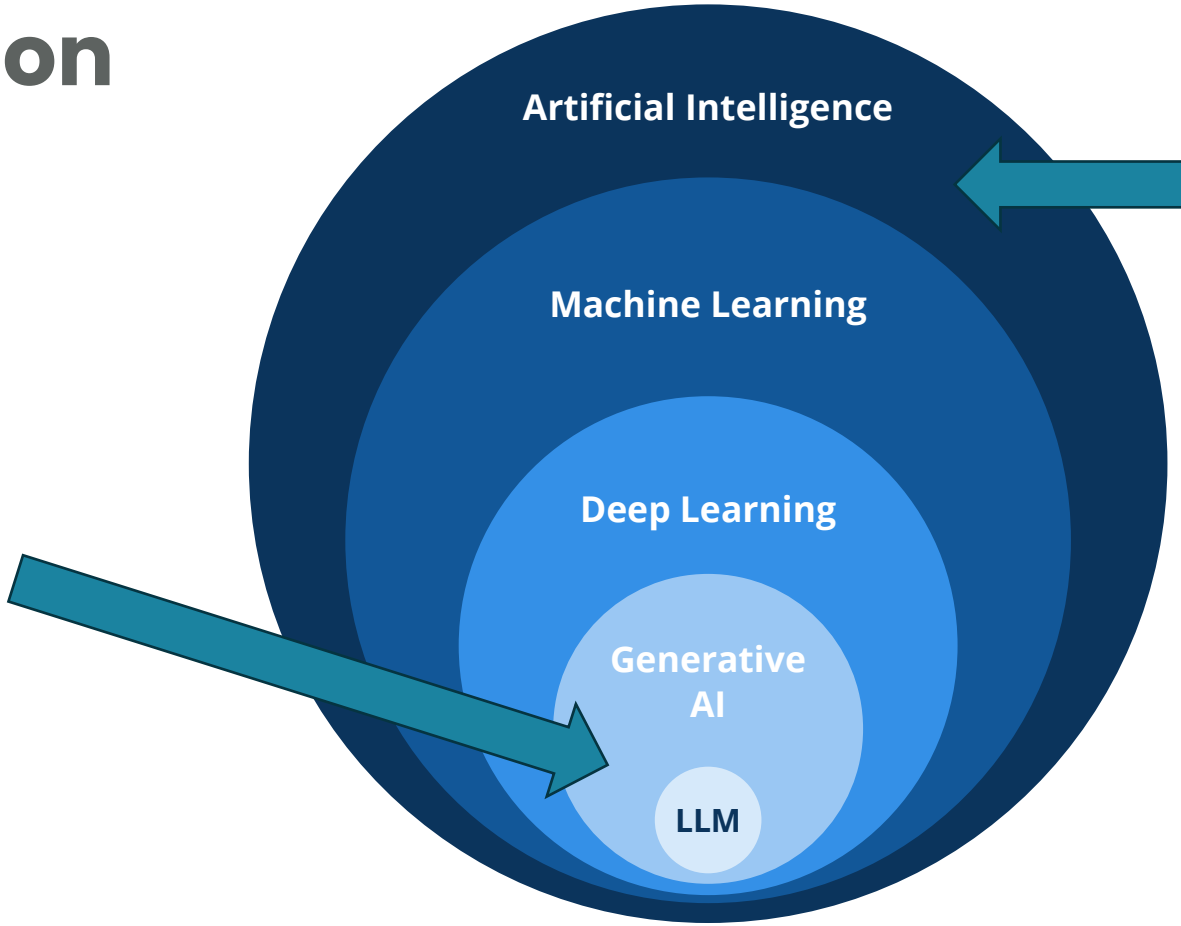
- Enhanced Diagnostics and Early Detection ★
- Predictive Analytics for Proactive Care
- Operational Efficiency and Productivity ★
- Personalised Medicine and Treatment
- Health Monitoring and Remote Patient Care
- Clinical Decision Support
- Population Health and Epidemiology







AI Definition



Simple predictive algorithms have been used in healthcare for >40 years....

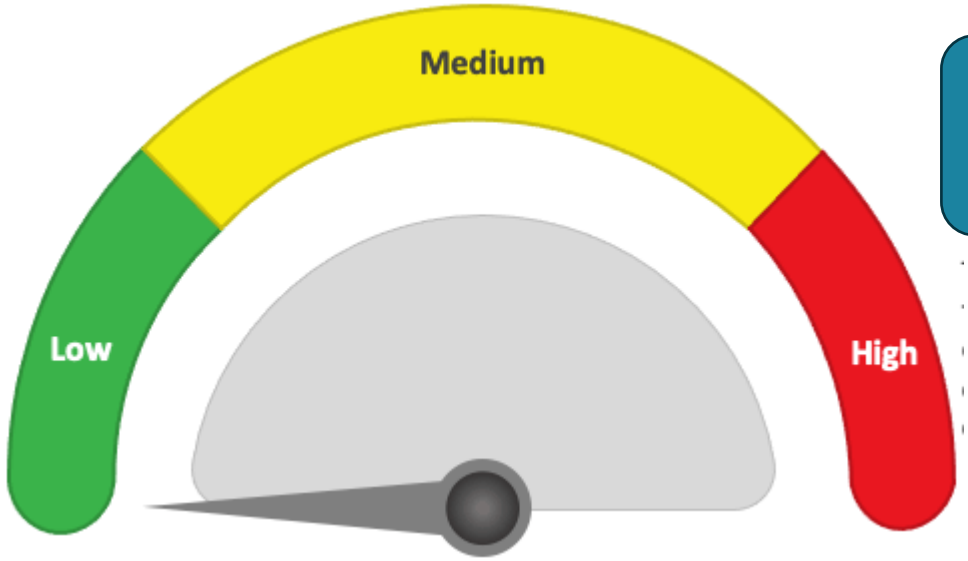
Most of the hype today.....

An AI system is a machine-based system that, for explicit or implicit objectives, infers, from the input it receives, how to generate outputs such as **predictions**, **content**, **recommendations**, or **decisions** that can influence physical or virtual environments. Different AI systems vary in their **levels of autonomy** and adaptiveness after deployment *(Public Service AI Framework, 2025)*

Health AI Solutions: Risk Spectrum

- Patient remote monitoring (wearables)
- Clinical prioritisation/triage tools based on patient symptoms
- Diagnostic assistance tools such as radiology algorithms

- Administrative tools: scheduling and workflow management
- Clinical note-taking scribes using speech recognition

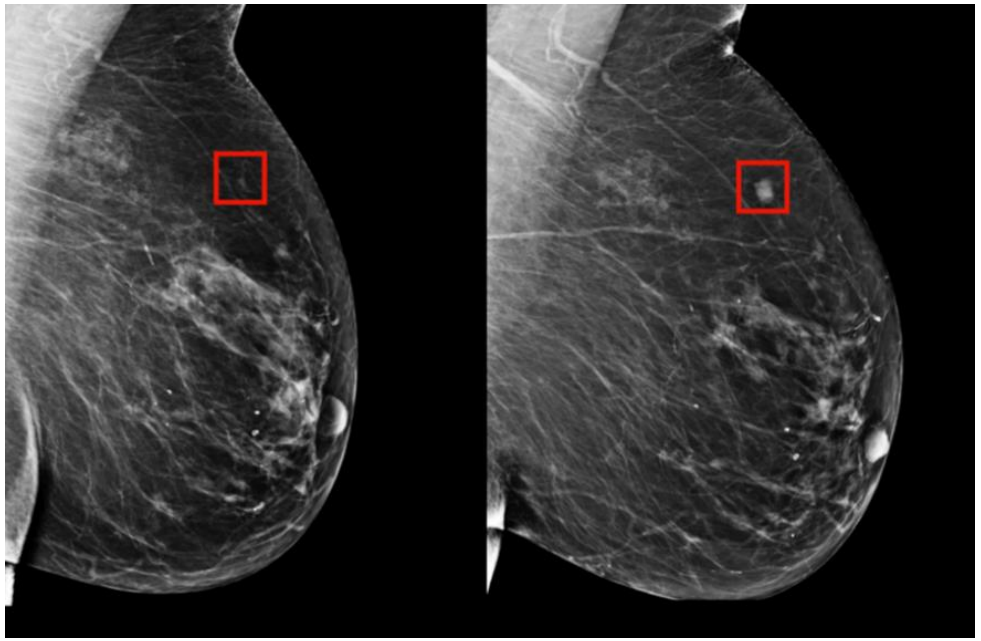


To be regulated as "Software as a Medical Device – SaMD" in the upcoming Medical Products Bill

- Clinical Decision-Support Systems providing diagnostic and treatment recommendations
- Fully autonomous diagnostic systems and robotic surgery

AI can have a transformational impact……..

Case Study: Enhanced Diagnostics and Early Detection



Article | Open access | Published: 06 March 2025

Artificial intelligence for breast cancer screening in mammography (AI-STREAM): preliminary analysis of a prospective multicenter cohort study

Yun-Woo Chang ✉, Jung Kyu Ryu, Jin Kyung An, Nami Choi, Young Mi Park, Kyung Hee Ko & Kyunghwa Han

Nature Communications **16**, Article number: 2248 (2025) | Cite this article

19k Accesses | **8** Citations | **120** Altmetric | Metrics

Abstract

Artificial intelligence (AI) improves the accuracy of mammography screening, but prospective evidence, particularly in a single-read setting, remains limited. This study compares the diagnostic accuracy of breast radiologists with and without AI-based computer-aided detection (AI-CAD) for screening mammograms in a real-world, single-read setting. A prospective multicenter cohort study is conducted within South Korea's national breast cancer screening program for women. The primary outcomes are screen-detected breast cancer within one year, with a focus on cancer detection rates (CDRs) and recall rates (RRs) of radiologists. A total of 24,543 women are included in the final cohort, with 140

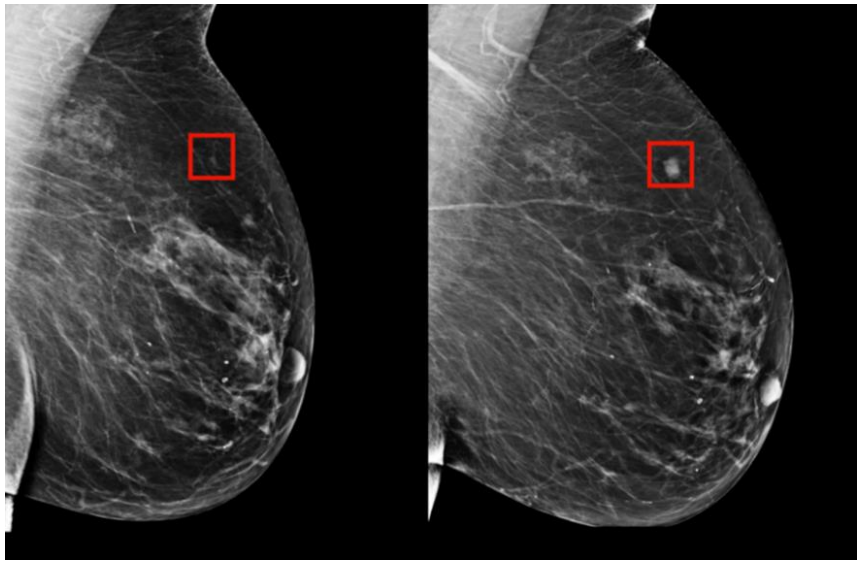
Effort Needed for Safe Adoption

Case Study: Enhanced Diagnostics and Early Detection

Algorithm trained on NZ data

Patient consent

Workforce trained, workflow updated



Trust and social licence established

Data stored and processed safely

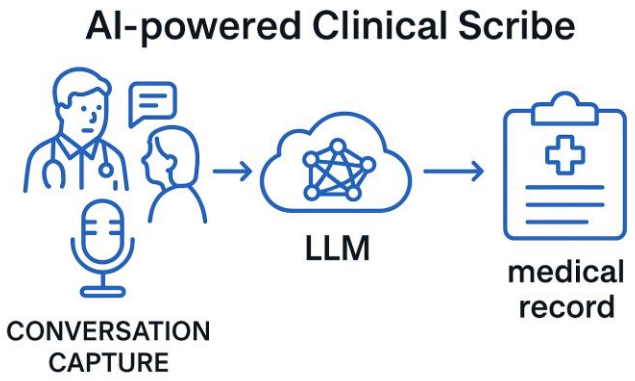
BSA policy updated

Monitoring plan in place

Case Study: Operational Efficiency and Productivity

- Large Language Models (LLMs) can summarise conversations, integrate with medical record
- Rapidly being adopted in private and primary care
- Two scribes recently endorsed for use by Health NZ

"I had my first ED shift using a clinical scribe last week. I really loved it – doesn't create anything new, just captures and organises what I've said in the room. Now I spend more time explaining the plan to the patient. I can get through the ED waiting room much faster. Big win in my book".



AI Governance in New Zealand

- Currently limited regulation for AI within New Zealand
- Health NZ has established an AI governing body – National AI and Algorithm Expert Advisory Group (NAIAEAG)
- Provide advice during research & development lifecycle
- Endorse AI tools for use within Health NZ
- Wide representation/membership - reflective of the assessment framework
- Framework developed based on internal Health NZ and international research – appropriateness key central theme

npj | digital medicine www.nature.com/npjdigitalmed

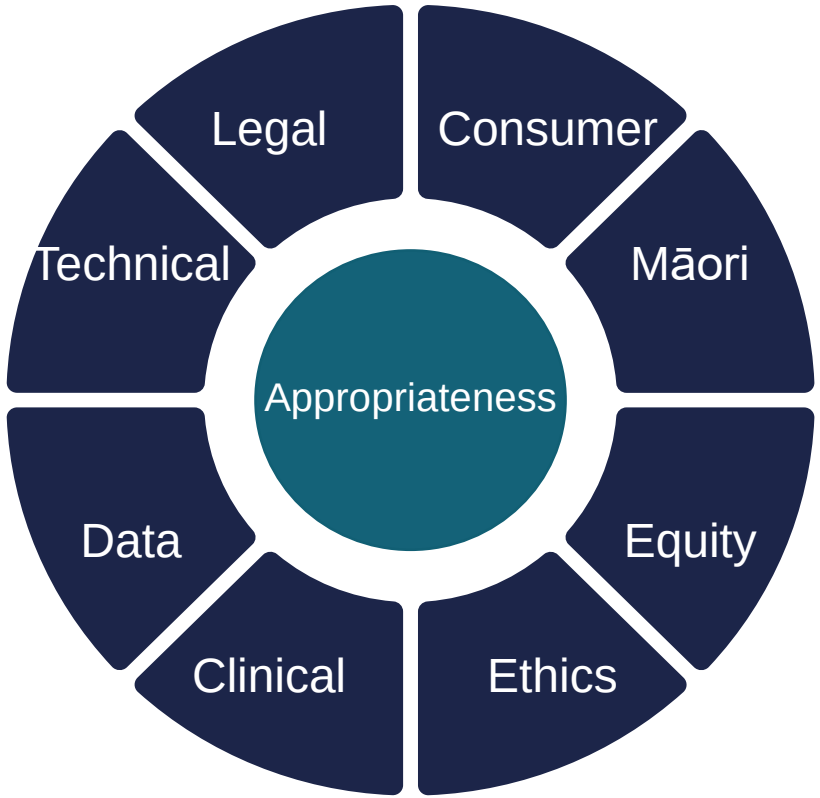
ARTICLE OPEN

An example of governance for AI in health services from Aotearoa New Zealand

R. Whittaker[1,2✉], R. Dobson[1,2], C. K. Jin[1], R. Style[1], P. Jayathissa[1], K. Hiini[1], K. Ross[3], K. Kawamura[1], P. Muir[1] and the Waitematā AI Governance Group*

Artificial Intelligence (AI) is undergoing rapid development, meaning that potential risks in application are not able to be fully understood. Multiple international principles and guidance documents have been published to guide the implementation of AI tools in various industries, including healthcare practice. In Aotearoa New Zealand (NZ) we recognised that the challenge went beyond simply adapting existing risk frameworks and governance guidance to our specific health service context and population. We also deemed prioritising the voice of Māori (the indigenous people of Aotearoa NZ) a necessary aspect of honouring Te Tiriti (the Treaty of Waitangi), as well as prioritising the needs of healthcare service users and their families. Here we report on the development and establishment of comprehensive and effective governance over the development and implementation of AI tools within a health service in Aotearoa NZ. The implementation of the framework in practice includes testing with real-world proposals and ongoing iteration and refinement of our processes.

npj Digital Medicine (2023)6:164; https://doi.org/10.1038/s41746-023-00882-z



2025 AI work programme:

improving the health of all New Zealanders by enabling more efficient, effective and precise healthcare

Leadership + Stewardship

- Establish aims, principles + agency roles
- Leverage international connections (HealthAI, Catalyst research funding)
- Explore governance model for Health AI (data, regulatory, strategy and operations)

Patient rights + safeguards

- Regulation of SaMD via Medical Products Bill
- Policy options for data governance, access and sovereignty
- HPCA/HIPC review

Enablers

- Robust HTA (alongside Pharmac and Health NZ)
- Understanding data and digital reset at Health NZ
- Internal MOH Pilots of GenAI
- Workforce: guidance to RAs and prof. groups

Case studies*

Examples:

- Clinical scribes
- Diabetic retinopathy screening
- Predictive algorithms for diabetes

Social License and Engagement

- Build trust via comms + engagement strategy (e.g. Ministry website, press releases, consultation)
- Establish Social License Work Programme

*Illustrative examples only - not an exhaustive l

Overarching Aim and Guiding Principles

We have adopted the following aim to guide our collective work in genomics and AI:

To improve the health of all New Zealanders by enabling more personalised and precise health care

Principles advocate for a wise (or "A SAGE") adoption of precision health technologies such as genomics and AI:

- Accessible – all New Zealanders should enjoy timely access to the benefits of precision health
- Safe and effective – tools and technologies should only be employed if they are both safe and effective
- Accountable - tools and technologies should be implemented fully transparently, be well-supervised and controlled
- Good value - all precision health tools and technologies must deliver value
- Equitable – ensures all New Zealanders can enjoy the benefits while being protected from the downsides. Also ensures services can be tailored to the needs of individuals and groups

Case Study: Enhanced Diagnostics and Early Detection

- June 2023: 36% Māori and 32% Pasifika eligible patients received diabetic retinopathy screening in South Auckland
- AI can categorise retinal photographs taken during screening and flag patients requiring ophthalmologist intervention
- Trained and validated on over 250,000 retinal images from retinal screening programs, including New Zealand
- A clinical trial demonstrated high sensitivity for detecting severe disease (100% of cases of ‘sight-threatening’ disease)
- Potential to improve access to screening, reduce wait times, ease workforce challenges
- Currently being piloted in 7 cameras in communities in South Auckland