

AI/ML DI ERA PENGOBATAN BERBASIS GENOMIK



ENTREPRENEUR FIRST



Reid Hoffman

Founder and Executive Chairman,
LinkedIn

Investor and board member at
Entrepreneur First, previously PayPal,
Apple and Fujitsu



SINCE 2011

\$10B+

Combined portfolio value

600+

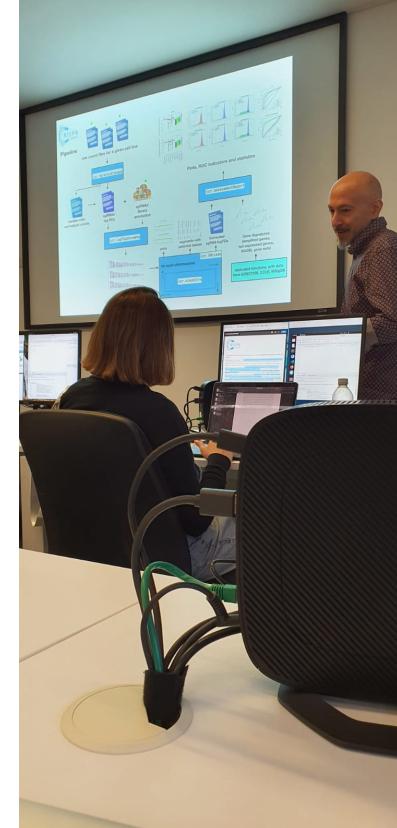
Companies founded

Riset, training, dan aktivitas saya yang didanai EF:

- EEID research group (University of Oxford, UK)
- European Bioinformatics Institute (EMBL-EBI, UK)
- Milner Therapeutic Institute (University of Cambridge, UK)
- Babraham Institute (University of Cambridge, UK)
- Kolaborasi riset dengan Siena Clinical & University Medical Center Utrecht (Netherlands)
- UK-Indonesia Genomics Meeting 2023

Pengalaman kerja saya lainnya:

- Founding Member, Head of Bioinformatics & Data Analytics, Advisor (Nusantics - Indonesia)
 - Bagian dari Task Force COVID-19 BPPT di 2020
 - Kolaborasi dengan Biofarma: kit-kit diagnostics ini telah dipakai jutaan orang di seluruh Indonesia
- Research Consultant (KAUST Pathogen Genomics Lab – Saudi Arabia)
- Visiting Researcher (Universita La Sapienza - Italy)
- Memiliki paper-paper publikasi internasional di bidang genomics, bioinformatics, dan AI/ML



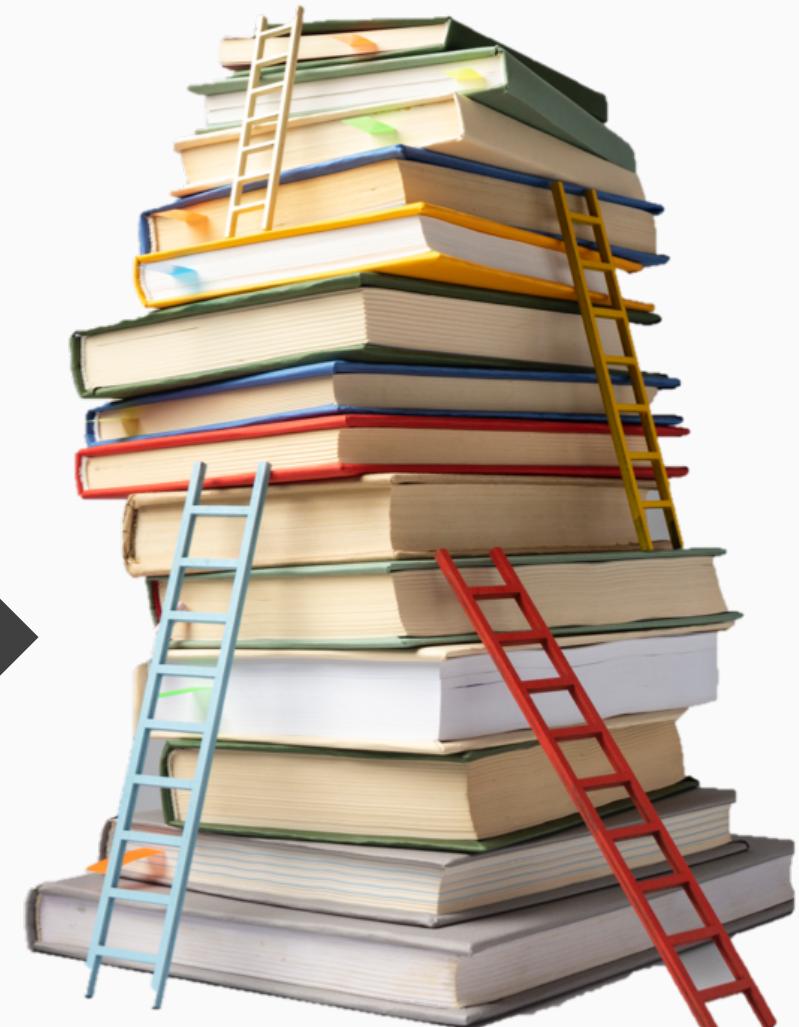
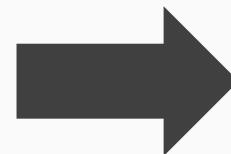


Apa Itu Genomics?

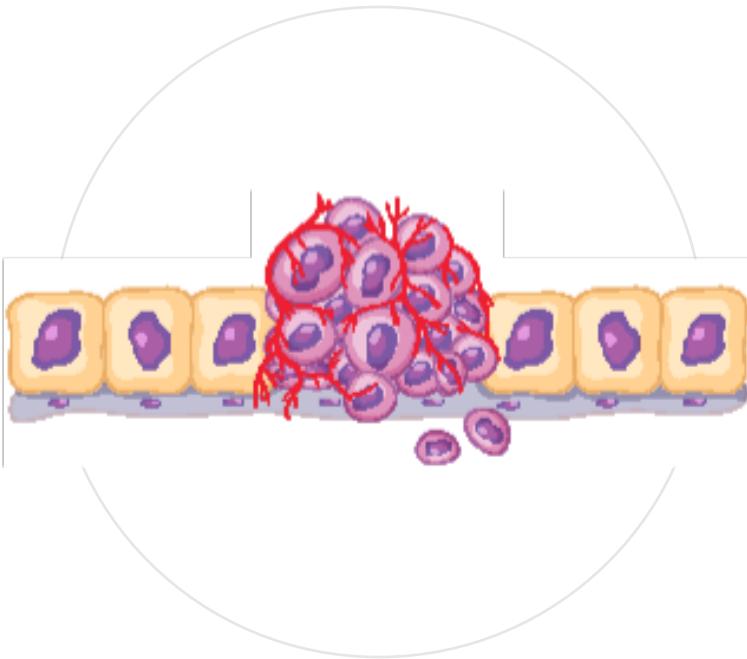
Studi mengenai materi genetik (*genome*), yang terdiri dari DNA atau RNA.

Contoh:

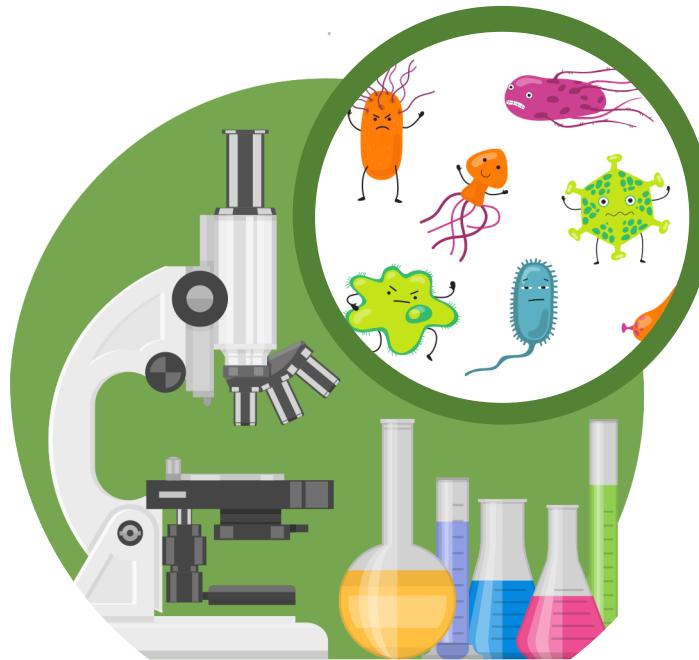
- Setiap manusia memiliki 3,2 milyar karakter kode DNA.
- Kalau dicetak, setara dengan tumpukan buku setinggi 61 meter!*



Genomics tidak hanya digunakan untuk meneliti sel manusia



Cancer genomics

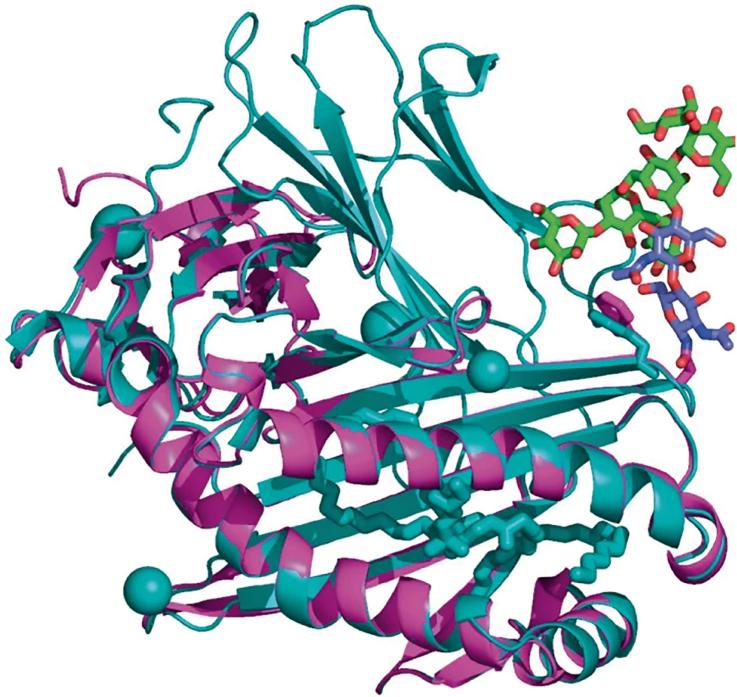


Microbiome genomics



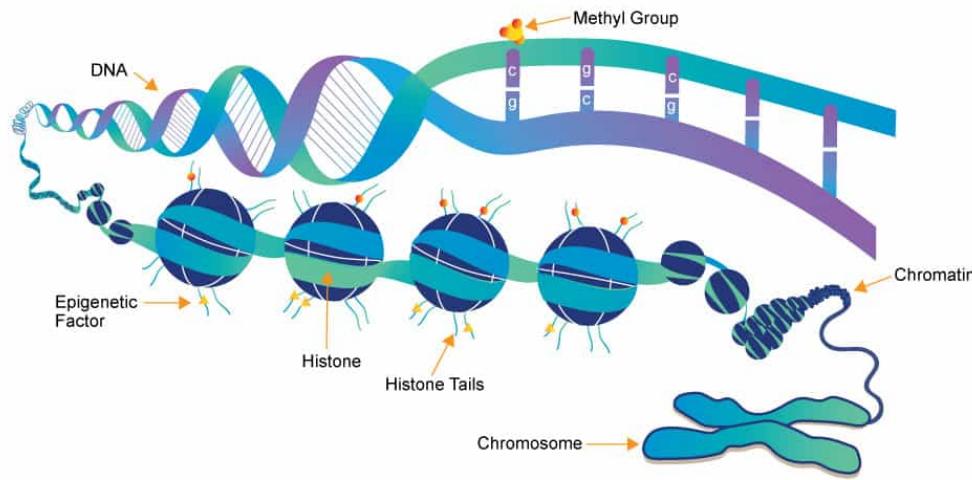
Pathogen genomics

Lahirnya genomics diikuti dengan -omics lainnya



Proteomics

(Bagdonas et al., 2021)



Epigenomics

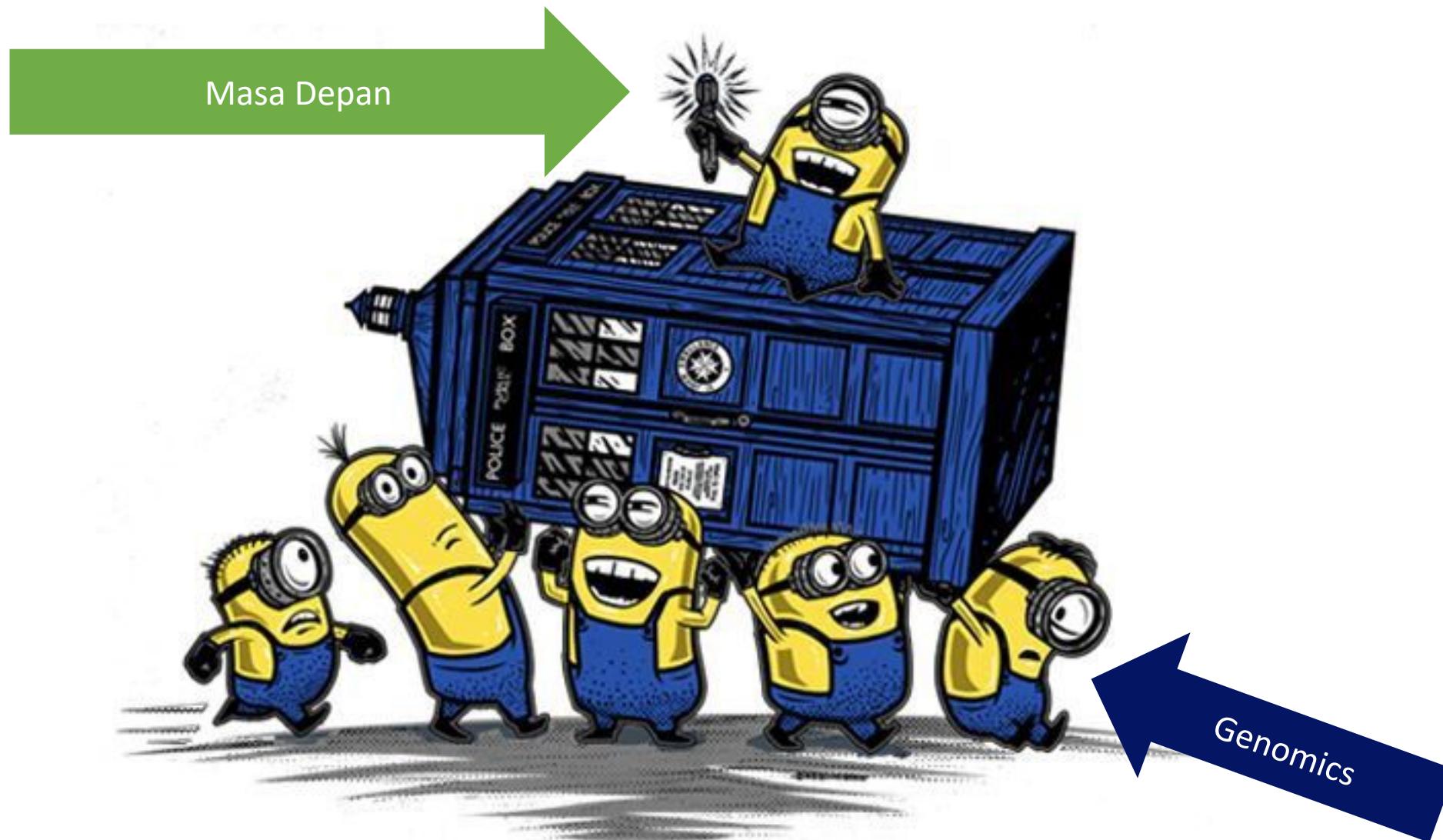
(genetics.org.uk)

Metabolomics

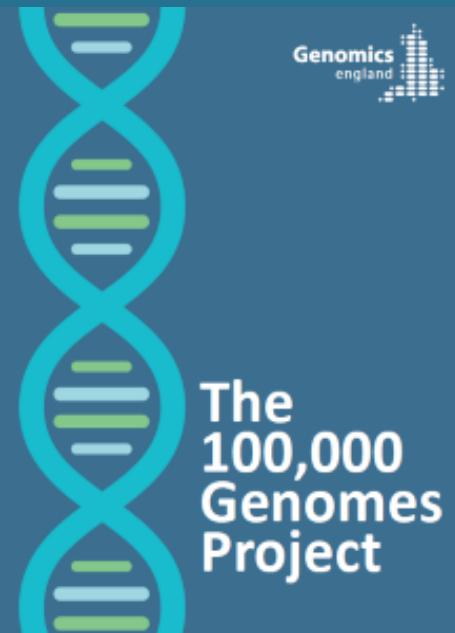
Transcriptomics

Dan lainnya

Kenapa Genomics?

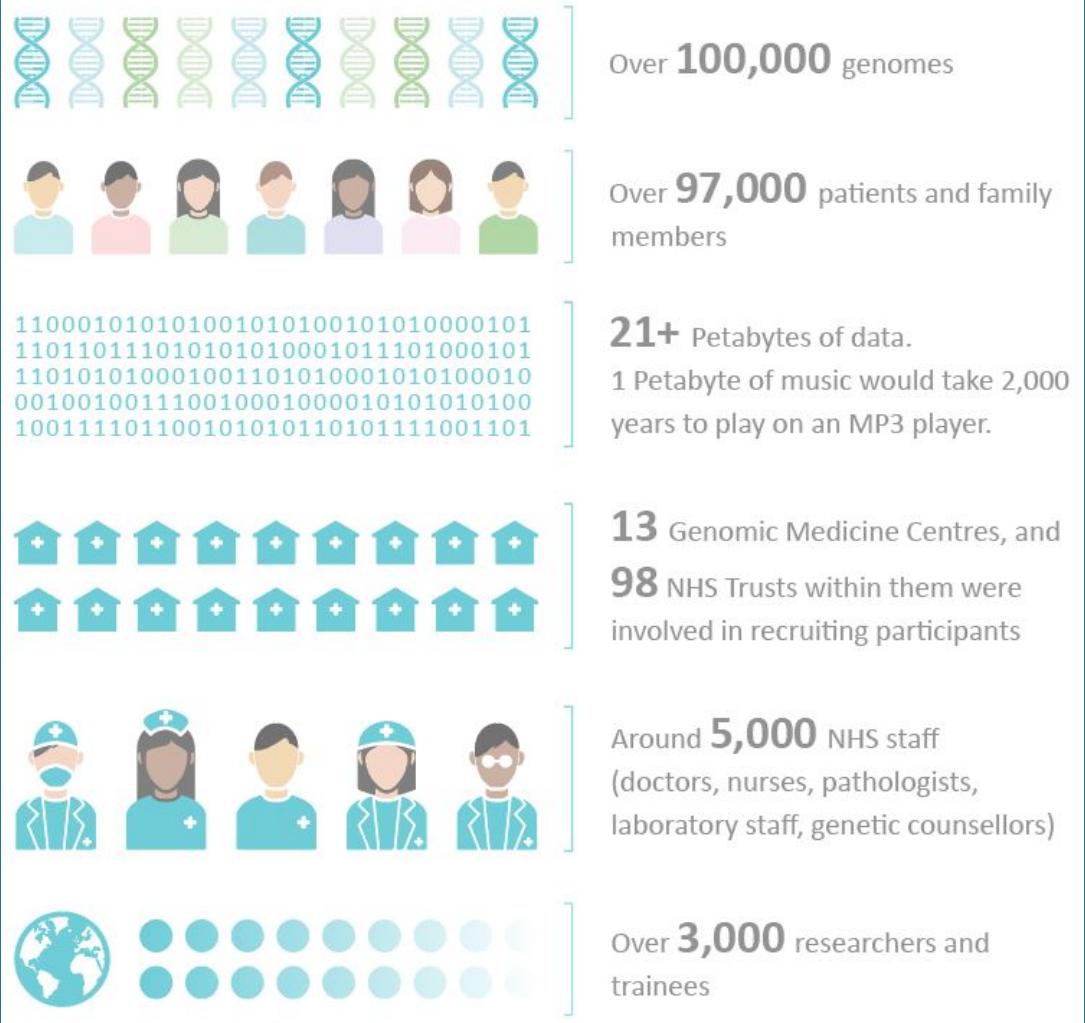


Dunia memasuki era baru:
pengobatan berbasis genomik



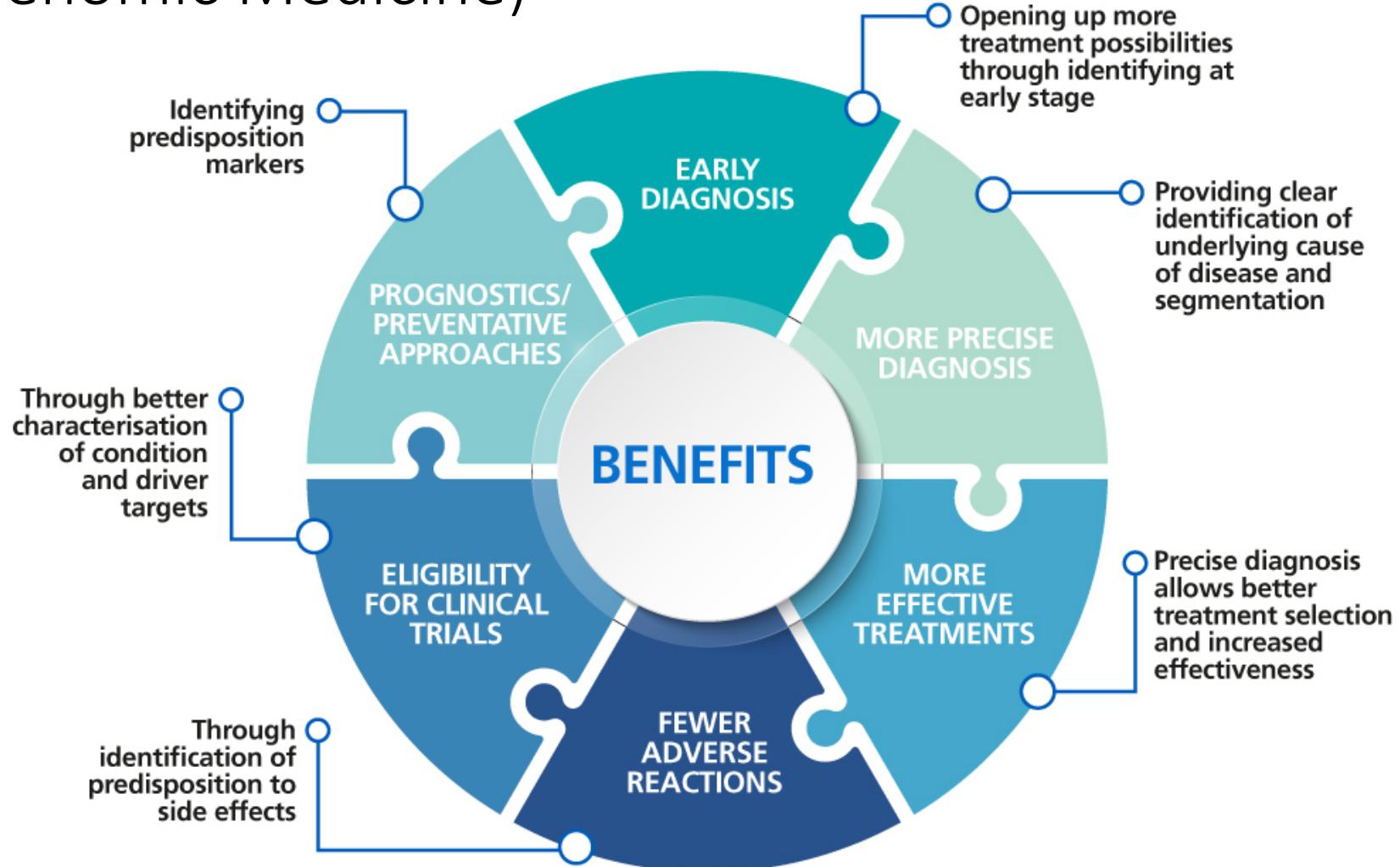
UK telah melakukan *whole genome sequencing* dari 100,000 pasien di 2018

The 100,000 Genomes Project in numbers



Fokus: kanker, penyakit menular, dan *rare diseases*.

Manfaat Pengobatan Berbasis Genomik (Genomic Medicine)



Contoh Manfaat Cancer Genomics

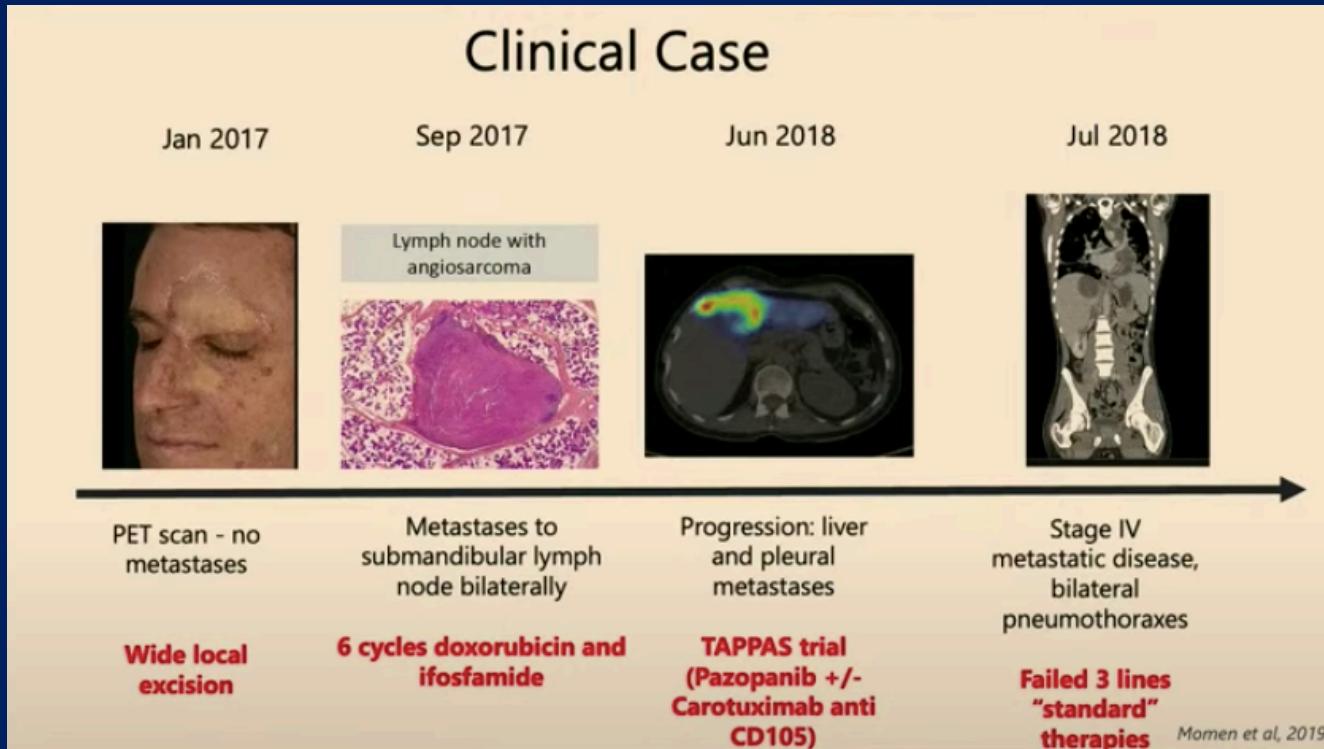


Image Credit: Prof. Serena Nik-Zainal (University of Cambridge)

Paper: Dramatic response of metastatic cutaneous angiosarcoma to an immune checkpoint inhibitor in a patient with xeroderma pigmentosum: whole-genome sequencing aids treatment decision in end-stage disease.

- Analisa *Mutational Signatures* untuk menemukan jenis pengobatan yang tepat untuk pasien kanker.
- Metode ini telah berhasil membantu memulihkan pasien kanker stage 4 yang sudah bermetastasis.
- Empat tahun setelah paper ini dipublikasikan, pasien tsb masih hidup dan sehat (update dari Prof. Nik-Zainal di Cambridge, 5 Juli 2023).
- Paper "Signatures of mutational processes in human cancer"* telah dikutip sebanyak 9000+ kali.

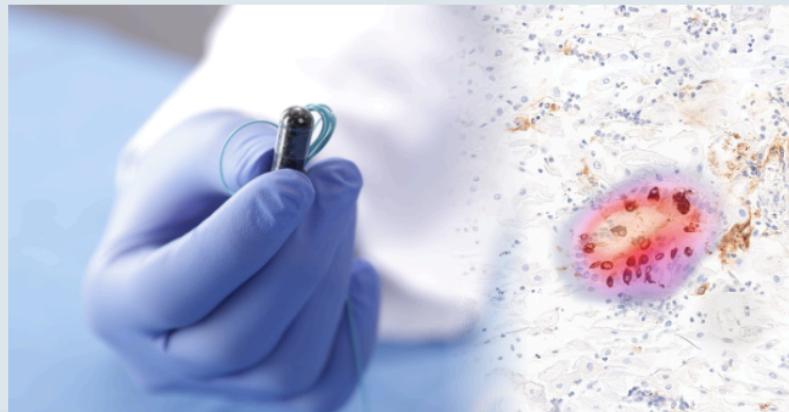
Alexandrov et al, 2013.
Nik-Zainal et al, 2012.

Contoh Aplikasi AI/ML Untuk *Cancer Genomics*

Artificial Intelligence for early cancer detection in
Nature Medicine

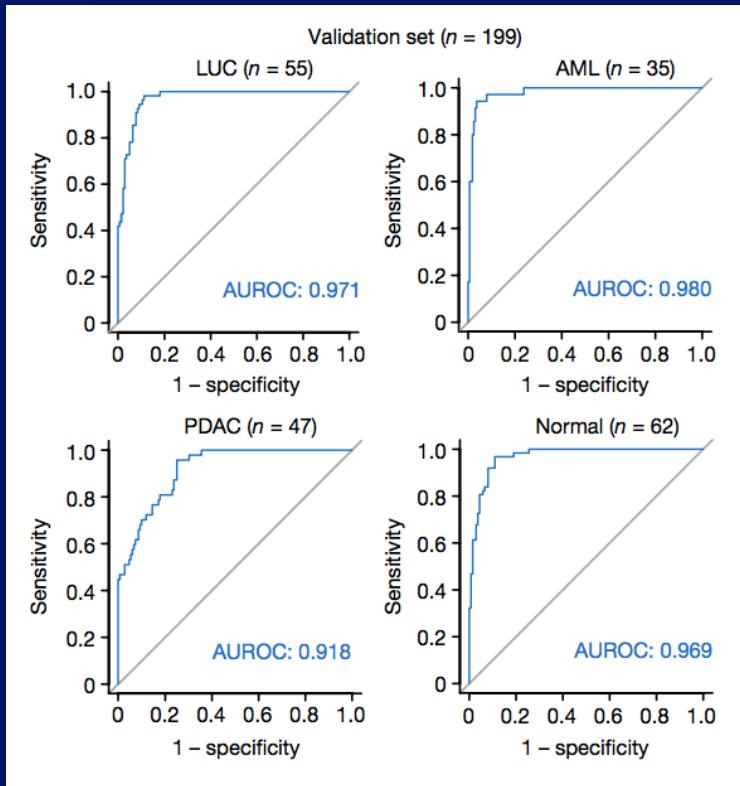
Our CEO, Dr Marcel Gehrung, published in Nature Medicine on the use of
Cytosponge and Artificial Intelligence to diagnose Barrett's oesophagus

15 April 2021

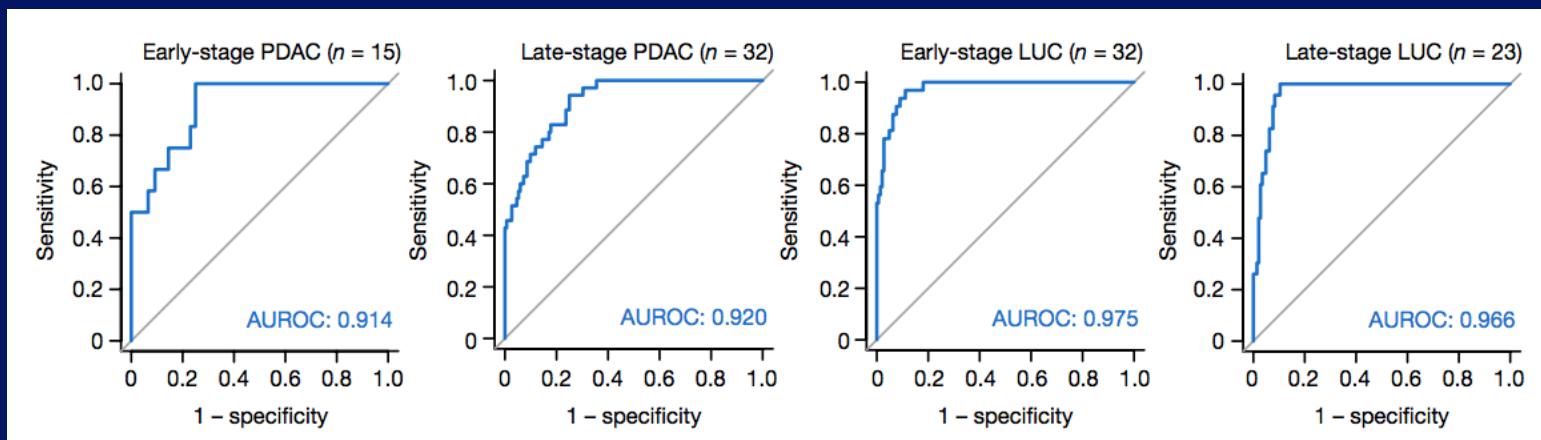


UNIVERSITY OF
CAMBRIDGE

Sensitive tumour detection and classification using plasma cell-free DNA methylomes

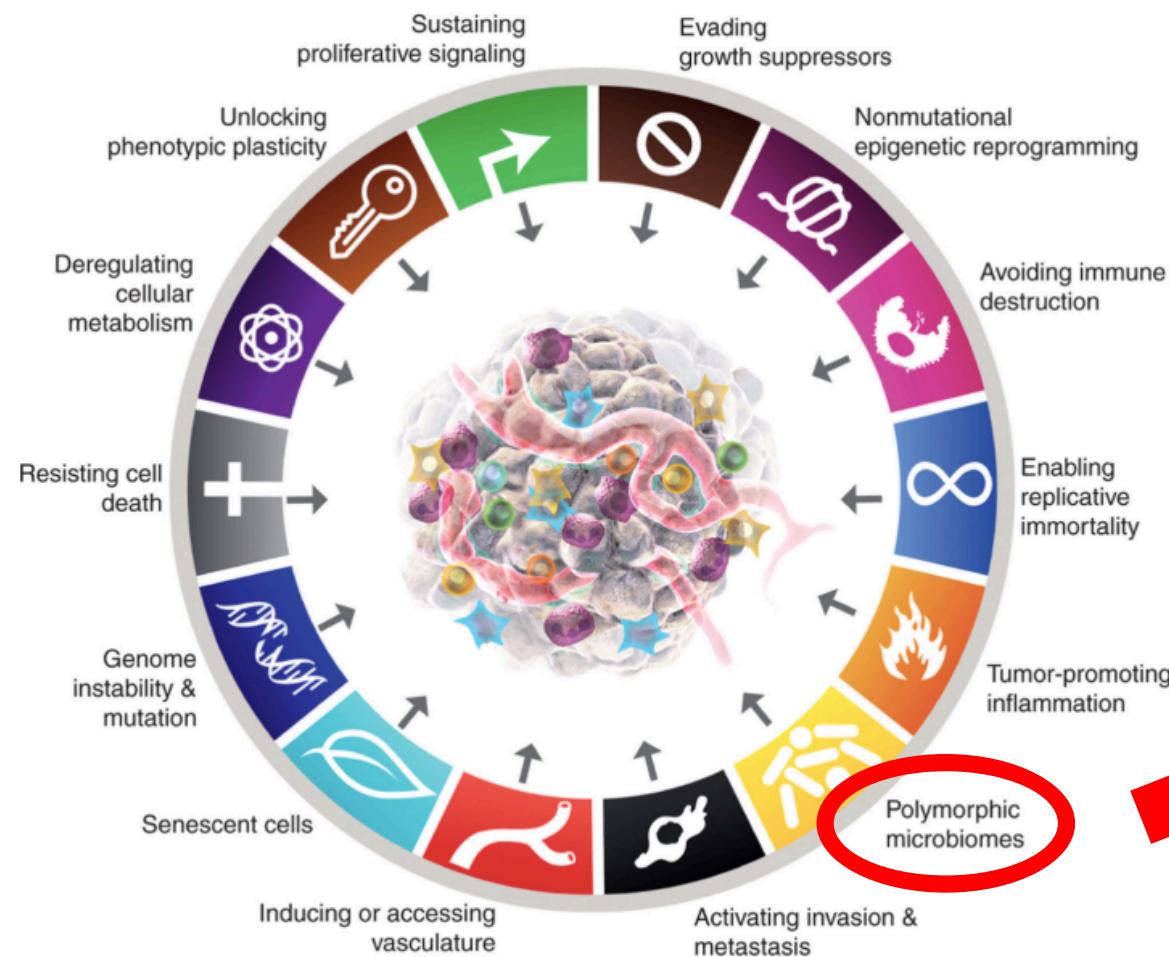


Excellent sensitivity and specificity
in validation set ($n=199$)



Comparable performance
in early versus late stage

Hallmarks of Cancer



Hanahan, 2022.
Lythgoe et al., 2022

Cancer – Microbiome Genomics

nature

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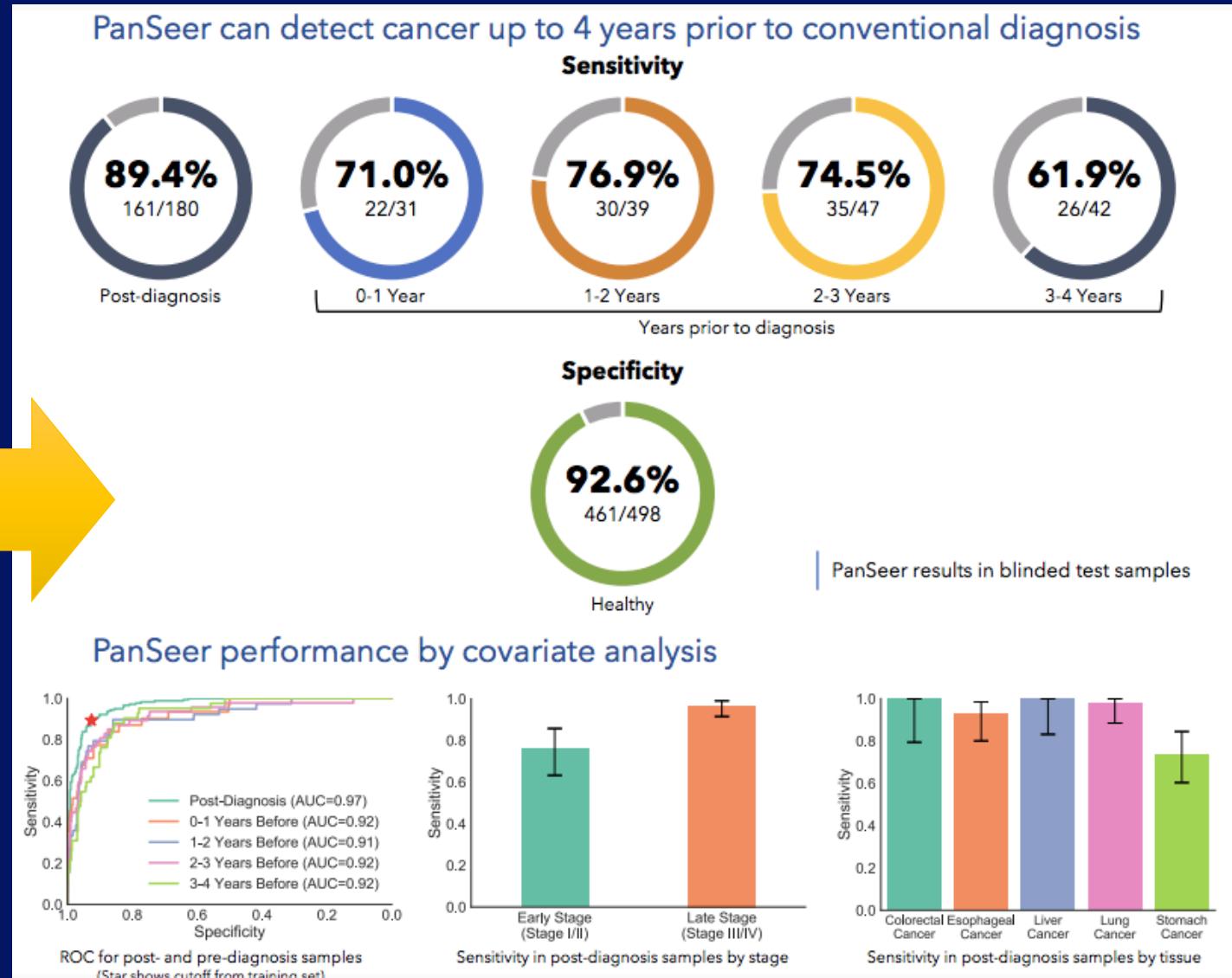
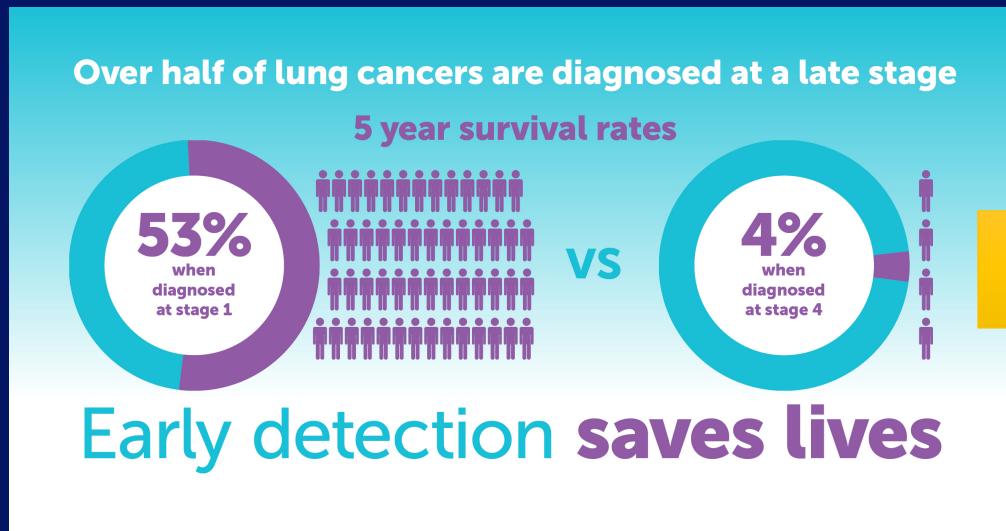
Article | Published: 11 March 2020

Microbiome analyses of blood and tissues suggest cancer diagnostic approach

Gregory D. Poore, Evguenia Kopylova, Qiyun Zhu, Carolina Carpenter, Serena Fraraccio, Stephen Wandro, Tomasz Kosciolek, Stefan Janssen, Jessica Metcalf, Se Jin Song, Jad Kanbar, Sandrine Miller-Montgomery, Robert Heaton, Rana Mckay, Sandip Pravin Patel, Austin D. Swafford & Rob Knight ↗

Nature 579, 567–574 (2020) | Cite this article

68k Accesses | 451 Citations | 892 Altmetric | Metrics



singleraoncology.com

Chen, X., Gole, J., Gore, A. et al.
Nat Commun 11, 3475 (2020).

Peluang besar untuk aplikasi AI/ML di bidang Genomics



Problem dengan
kompleksitas tinggi



Data yang besar



Bidang yang
berkembang pesat



Indonesia is at the very start of its Genomics journey and needs to catch up fast to its peers in SEA

Genome Sequencing Maturity and Age
Illustrative

1990

2010

2015

2017

2019

2022

Stages

China and Korea currently have developed limited clinical applications of genomics. Private sector participation also remains on the lower side. Korea restricts areas and scope of genomics for the private sector in the country



Due to high private participation, Vietnam is ahead of other SEA countries. Indonesia should follow a similar model along with public genomics efforts



US and UK lead in terms of genomics research and national studies. The US is further ahead due to wider private-sector participation

Source(s): Expert inputs, Desk research, Redseer Analysis

Stage 1: Nascent

Stage 2: Growth

Stage 3: Maturity

UK – Indonesia Genomics Meeting 2023

Goal: mendorong kolaborasi dan *knowledge transfer* antar kedua negara.
Akan dihadiri oleh ilmuwan dan peneliti dari:



UMC Utrecht



Jika Bapak/Ibu berminat, dipersilakan untuk
register interest di:
genepica.org/genomics2023

General inquiry:
genepica.org/contacts

Funded by

