ตอบโจทย์การวิจัยและธุรกิจจีโนมด้วย “computational genomics Platform”
Research and business enabler as a national computational genomics Platform

ดร.ศิษเฎศ ทองสิมา

ผู้อำนวยการธนาคารทรัพยากรชีวภาพแห่งชาติ

สำนักงานพัฒนาวิทยาศาสตร์และเทคโนโลยีแห่งชาติ

ประวัติการทำงาน:

2561-ปัจจุบัน	ผู้อำนวยการธนาคารทรัพยากรชีวภาพแห่งชาติ สำนักงานพัฒนาวิทยาศาสตร์และเทคโนโลยีแห่งชาติ
2550-2561	Head of Biostatistics and Informatics Laboratory, Genome Institute, BIOTEC, Thailand

Computational Genomics Platform

Research and business enabler as a national computational genomics platform

Sissades Tongsima, PhD

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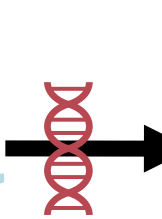
Genetic factors play “roles” in disorders

129.36
Meter

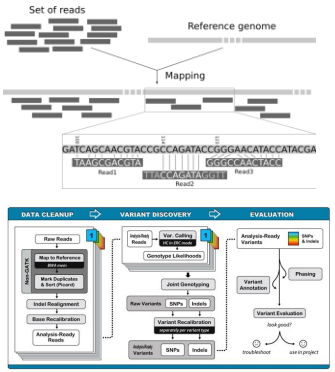
Paper stack of Human genome

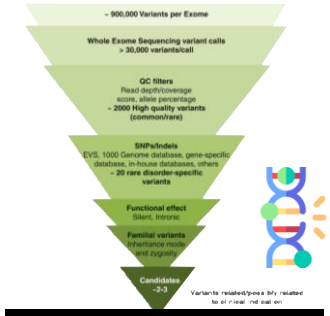
> 3 billion nucleotides
or DNA “letters."

Population specific requirements



100GB
raw data





2-3 candidate variants!!

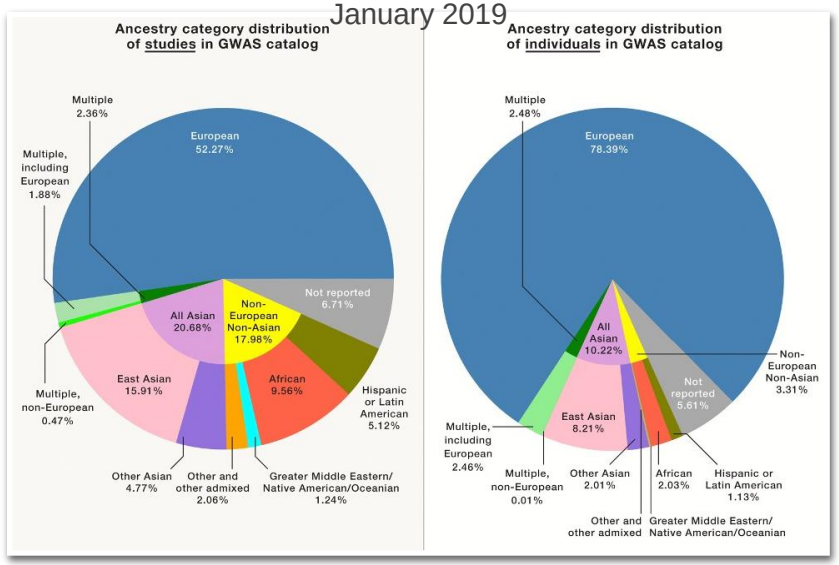
Thai reference exome database (T-REx)

https://trex.nbt.or.th

- **Caucasian bias reference genome**
- **Require specialized High Performance Computer (HPC)**
  - Time ***AND*** Storage ⇒ **critical demands**

    Normal HPC ⇒ 30 hours VS customized HPC ⇒ ~1 hour
  - NBT HPC for genomic-related processing
- **Require advanced bioinformatic tools and services**
- **Require skillful bioinformaticians/programmers/system admin**

Summary of GWAS Studies in the GWAS Catalog through January 2019



DOI:https://doi.org/10.1016/j.cell.2019.02.048

GeTH 50K

1 HPC & Storage

- 2x Accelerated AI machines (DGX-A100)
- 3x FAT nodes (192 cores / 3TB)
- 8x Web & Database servers
- 3000 TB storage & 10000 TB buffered storage

2 Software Management & QC

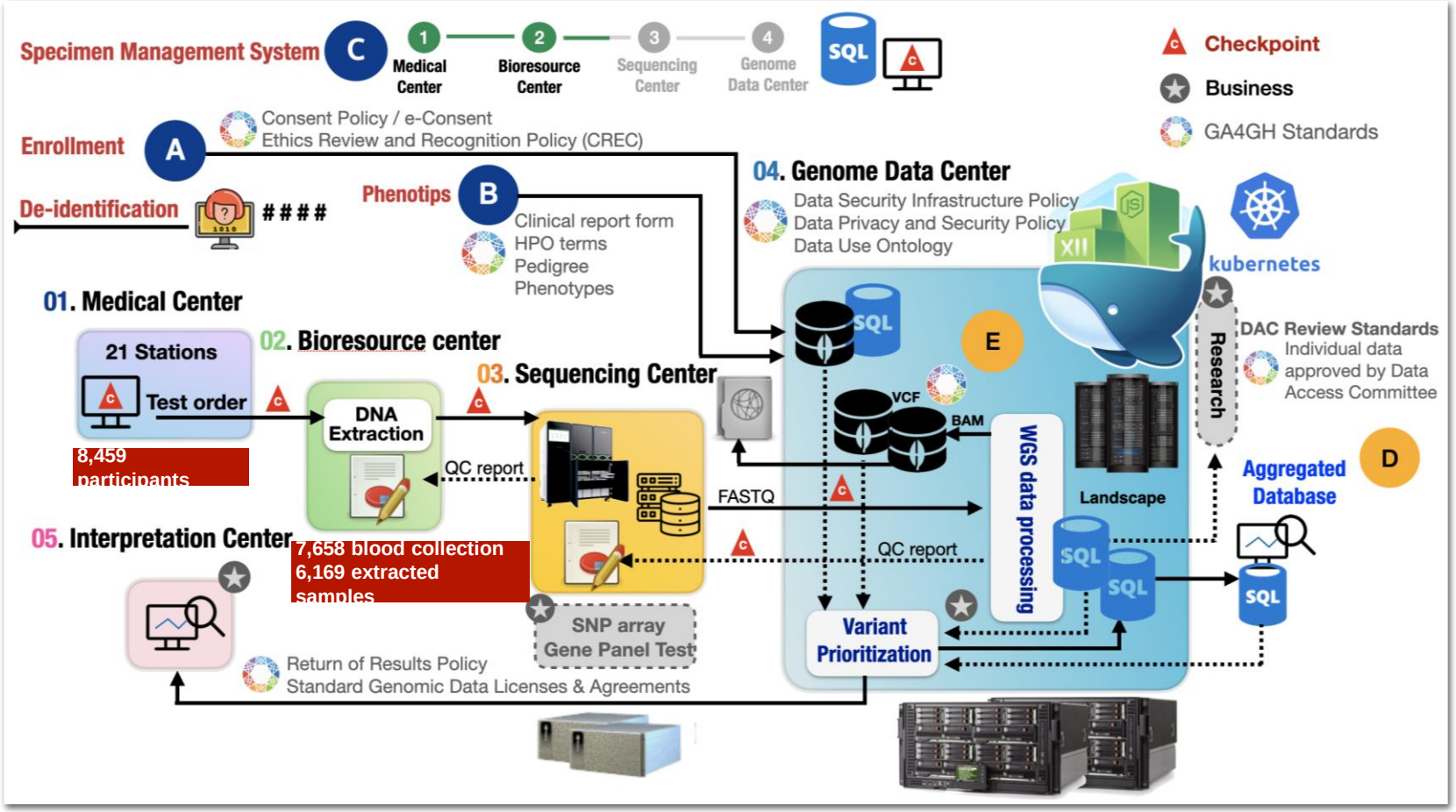
- Enrollment & e-consent
- PHENOTIPS
- Specimen Mangement System (SMS)
- Whole Genome Sequence Data QC
- Accelerated WGS processing

3 Genomics dB & Prioritization platform

- Population genomics platform
- T-REx Database
- Thai Reference Genome dB
- Variant Annotation and Prioritization (VAPP)

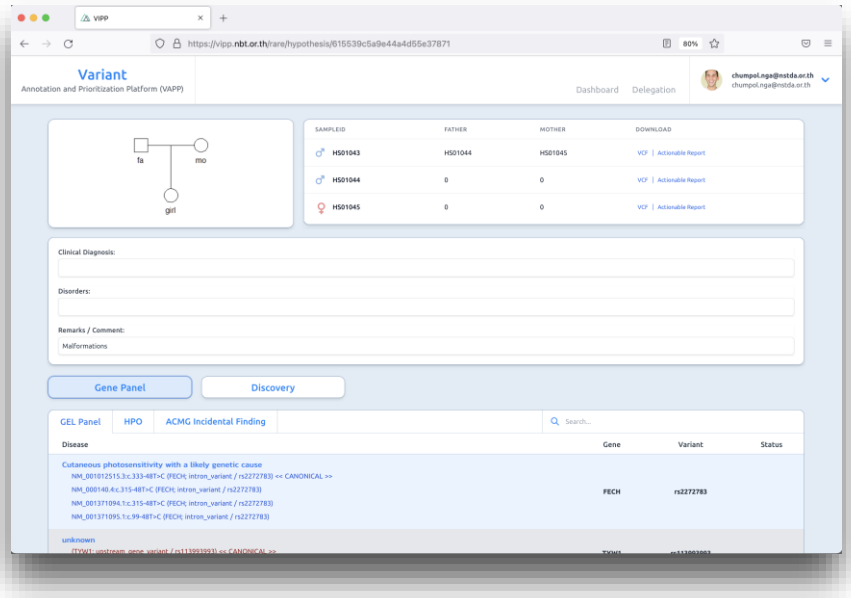
4 Data governance & Policy

- GeTH Data Governance policy
- Data Use Access policy
- Data sharing policy
- Intellectual Property policy

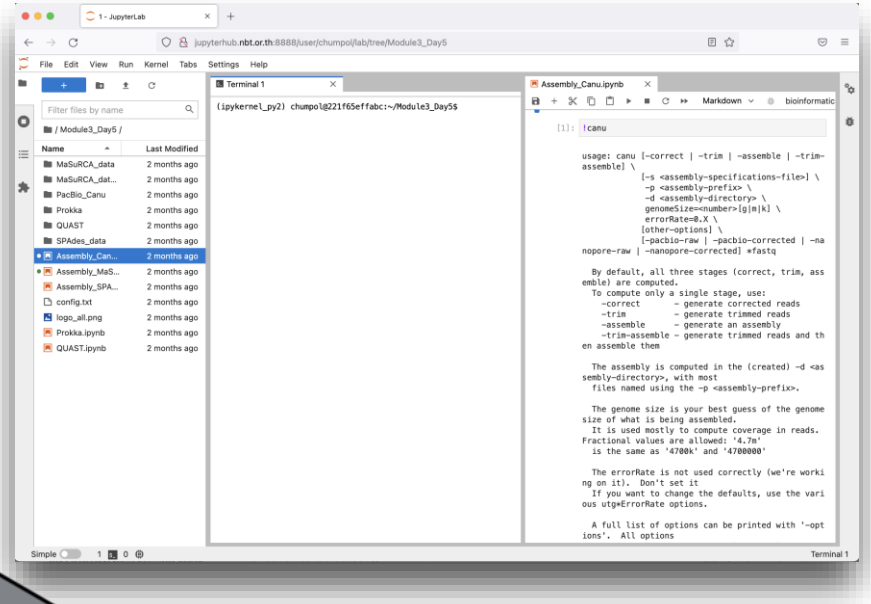


Computation services and Data access

Web Application with preannotated results



Jupyterhub / Terminal / Virtual Env. for Adv. proc on control data



Doctor - Researcher - Private comp.

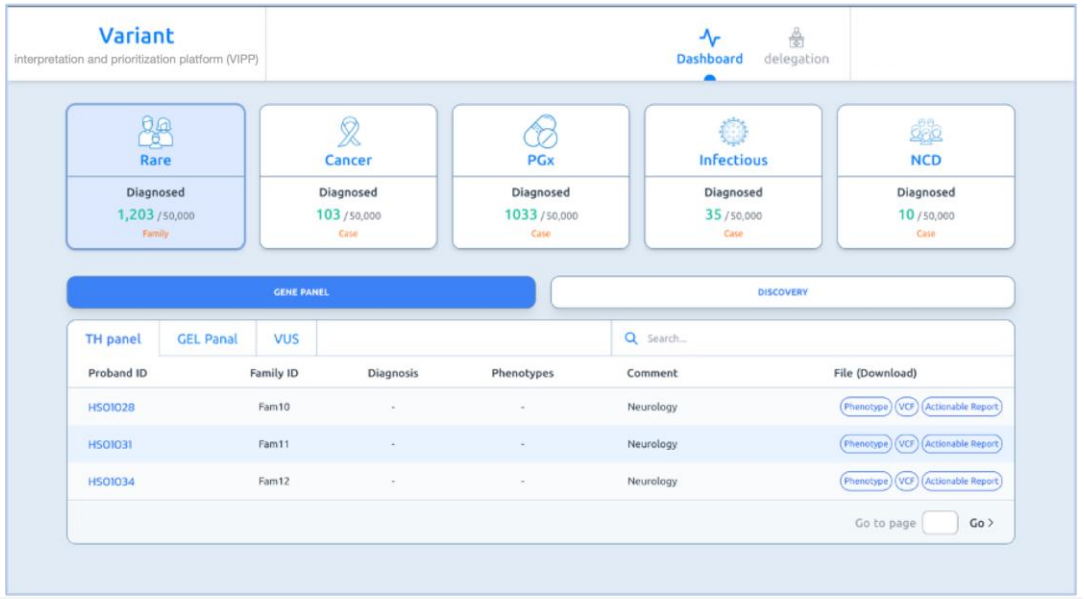
HPC & Storage

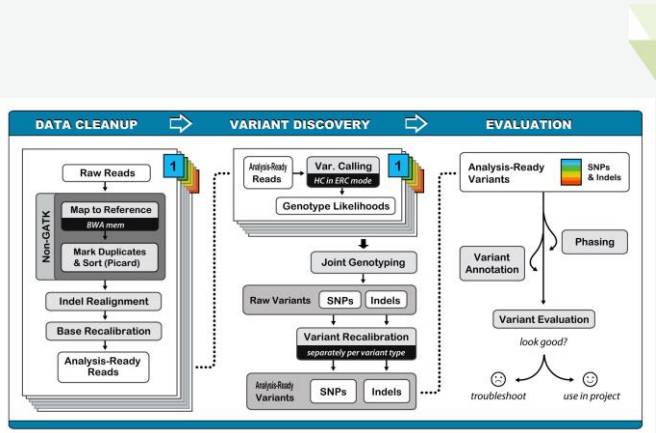
To transfer genome data is Big problem !

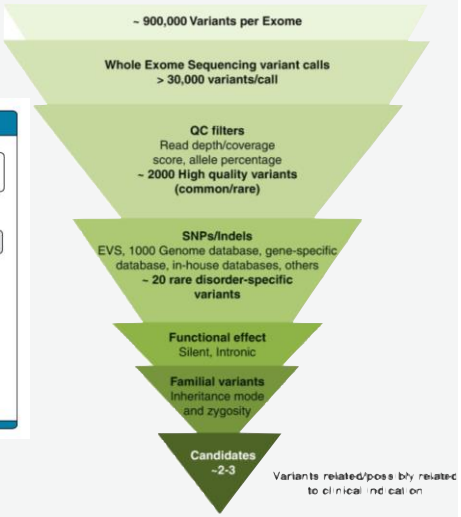
Internet bandwidth
Computing resources
Data leak !

Variant Annotation and Prioritization: VAPP

- Annotation & Prioritization (ANALYSIS)
  - Rare
  - Cancer
  - NCD
  - PGx
  - Infectious
- GEL panel + HPO terms
- Large polygenic risk score (PGS) database
- Genomics sequence annotate for “supporting” clinical interpretation







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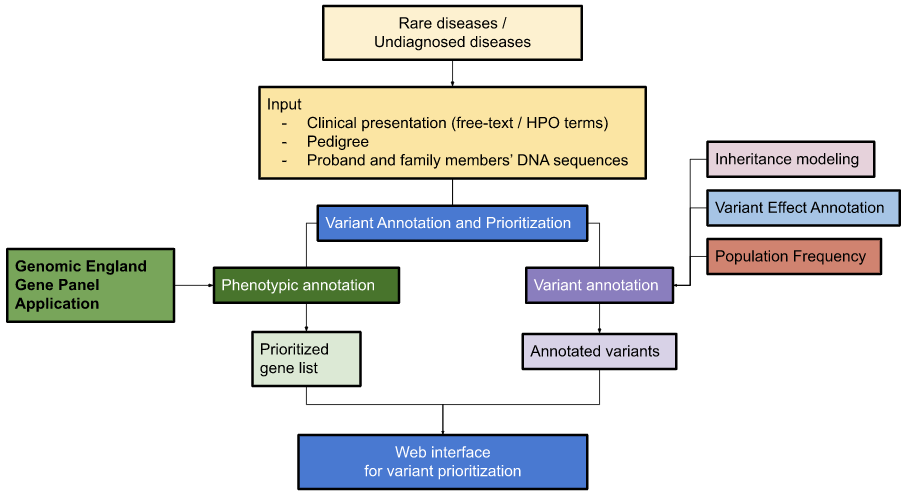
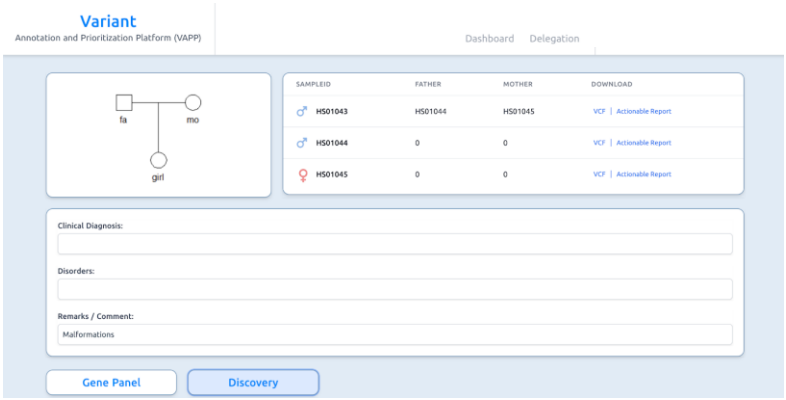
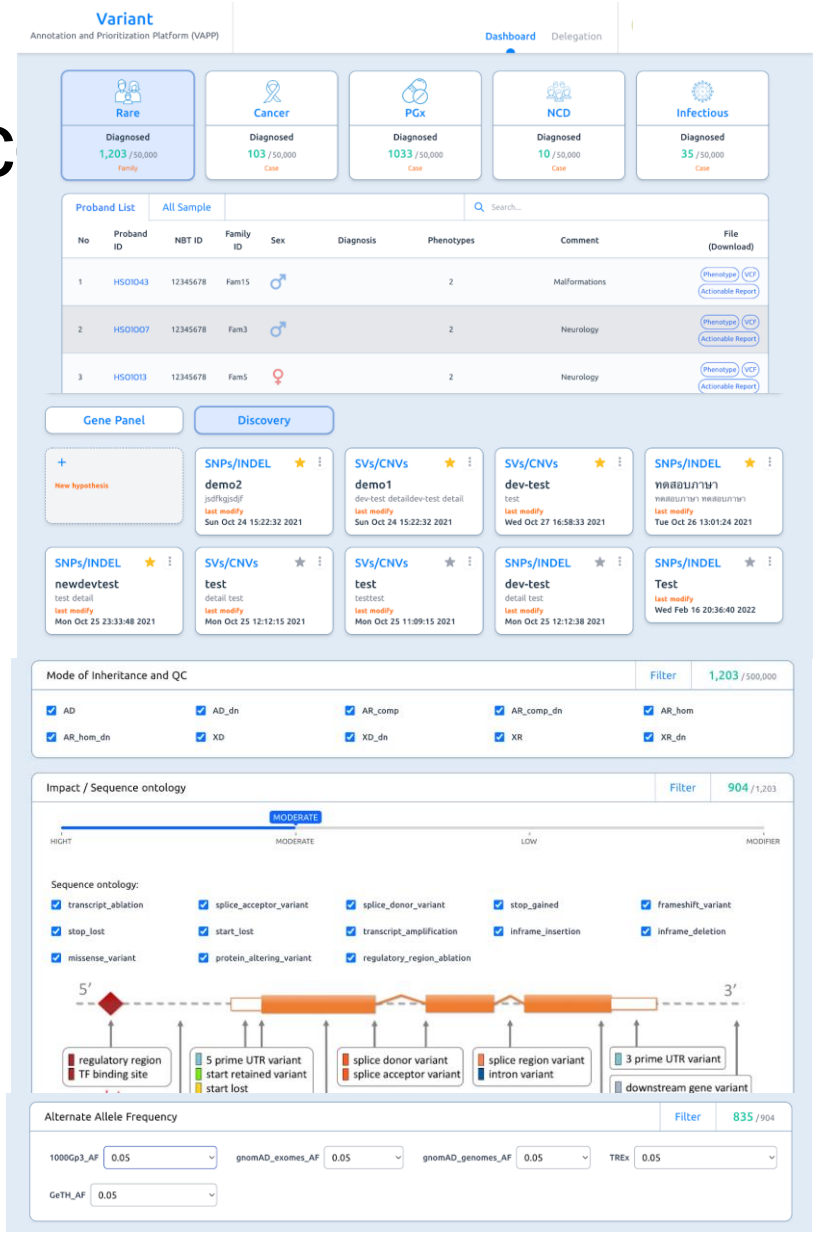
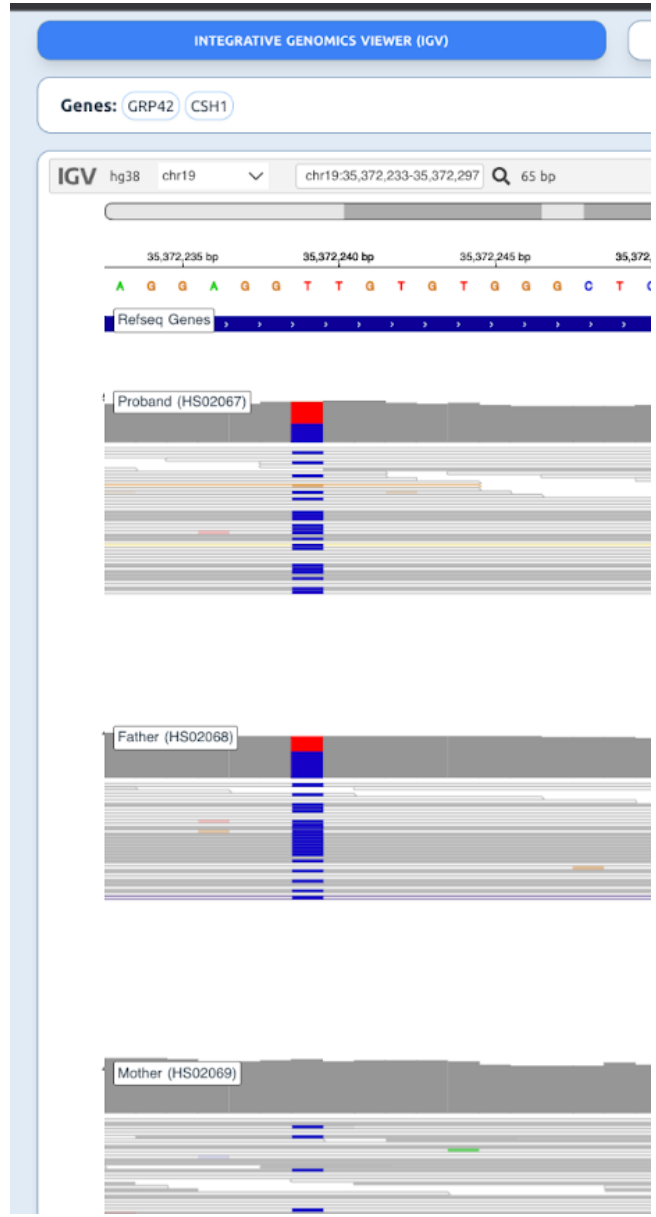
VAPP

Rare disease + Canc









VAPP

NCDs

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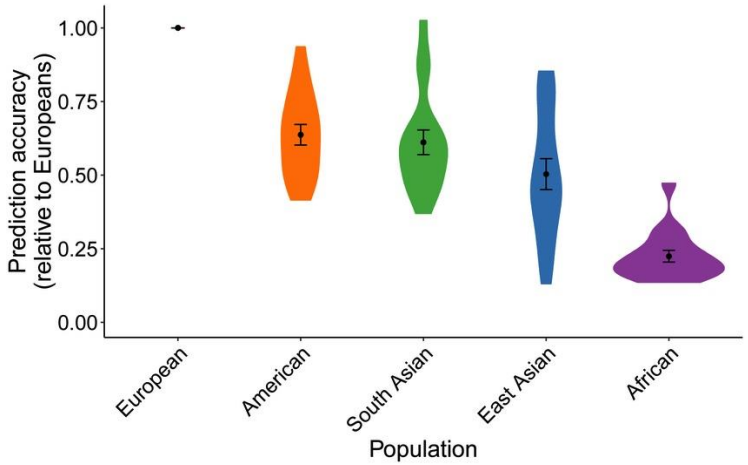
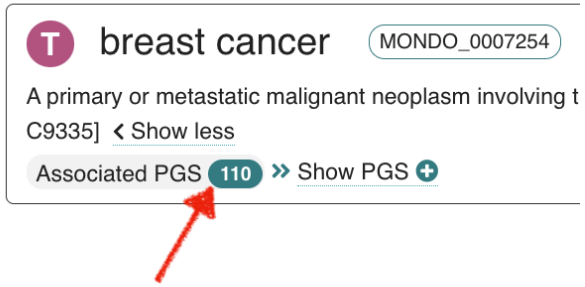
PGS Catalog

The development of the PGS Catalog is supported by:
UNIVERSITY OF CAMBRIDGE Department of Public Health and Primary Care · EMBL-EBI · HDRUK Health Data Research UK · Baker Heart & Diabetes Institute

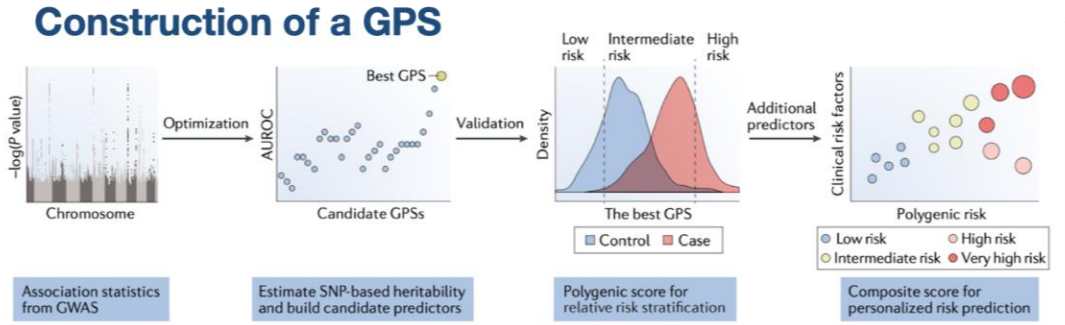
Polygenic Scores	Traits	Publications
2,174	531	288

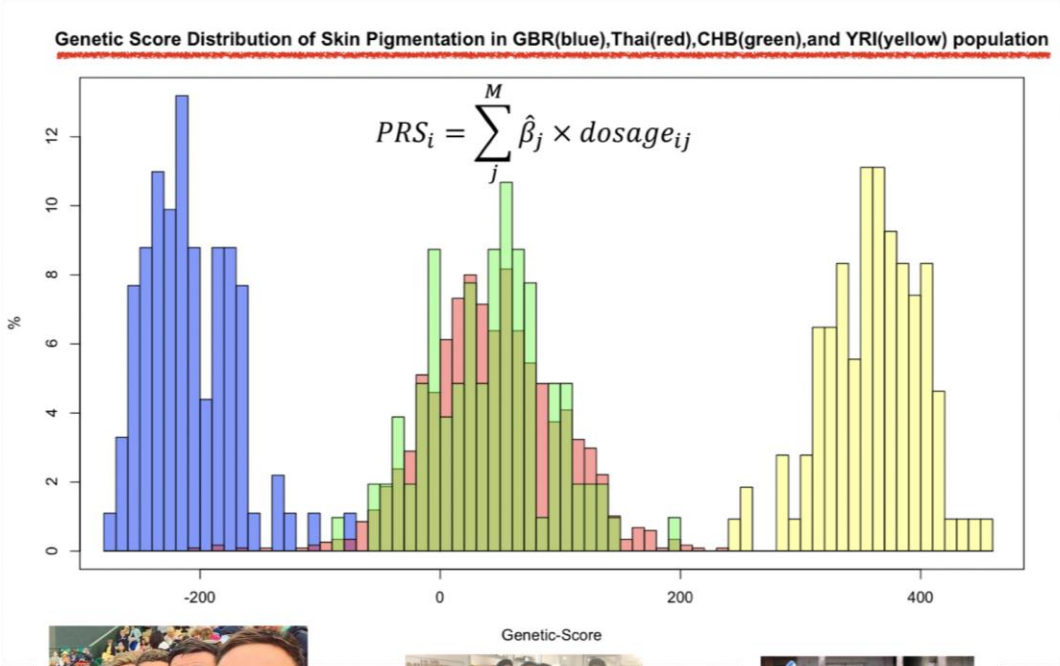




Which one should we use for Thai PGS?

Martin et. al., 2019. Clinical use of current polygenic risk scores may exacerbate health disparities. Nature Genetics, 51(4), pp.584-591.






GBR


Thai


Yoruba in Ibadan, Nigeria


Chinese

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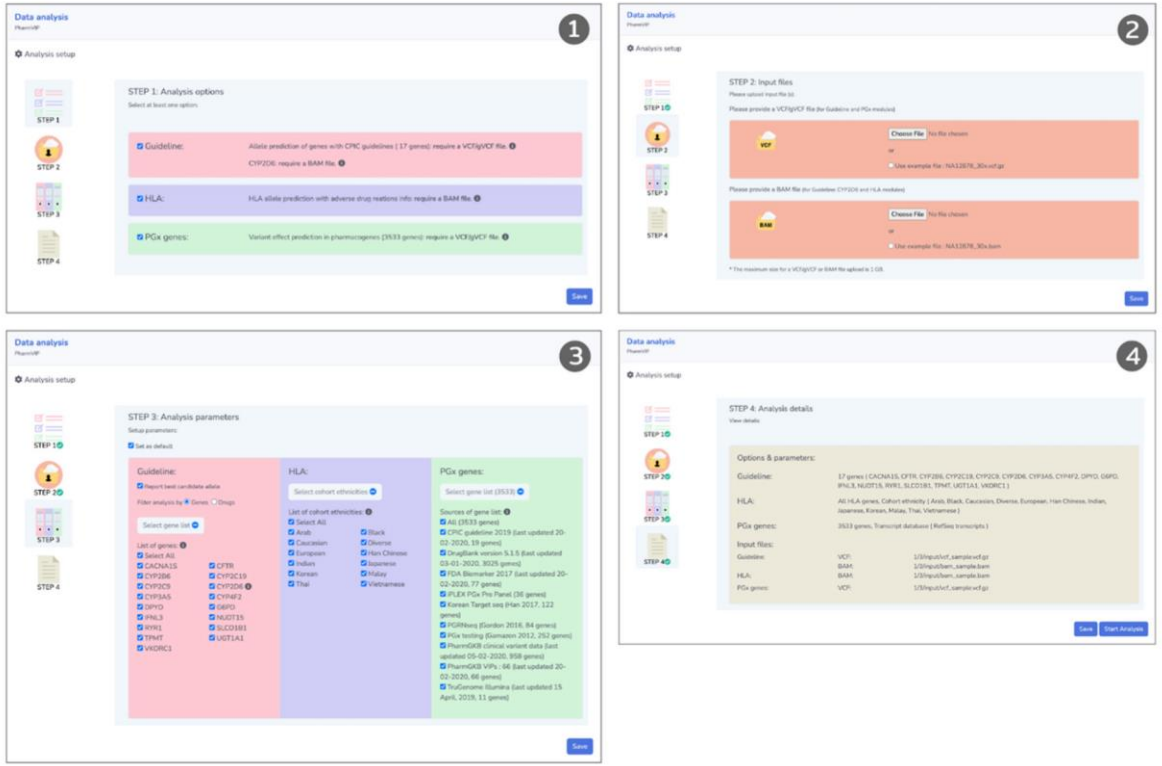
VAPP

Pharmacogenomics: PGx

PharmVIP https://pharmvip.nbt.or.th

- Pharmacokinetics (Cytochrome P450)
- Pharmacodynamics
- Immune-mediated adverse drug reaction; IM-ADR

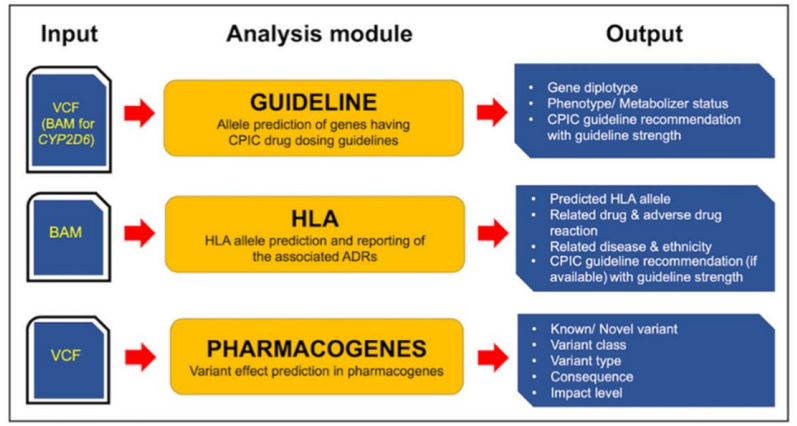
Video demo

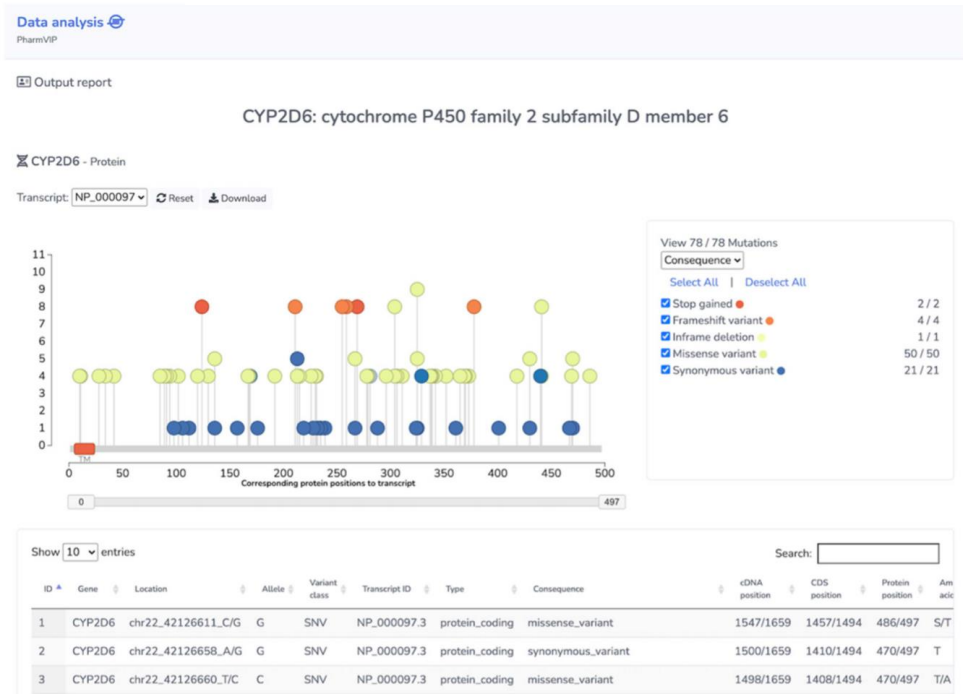


Journal of Personalized Medicine
MDPI
Article

PharmVIP: A Web-Based Tool for Pharmacogenomic Variant Analysis and Interpretation

Jittima Piriyapongsa [1,*], Chanathip Sukritha [1], Pavita Kaewprommal [1], Chalermpong Intarat [1], Kwankom Triparn [1], Krittin Phornsiricharoenphant [1], Chadapohn Chaosrikul [1], Philip J. Shaw [2], Wasun Chantratita [3], Surakameth Mahasirimongkol [4] and Sissades Tongsima [1]



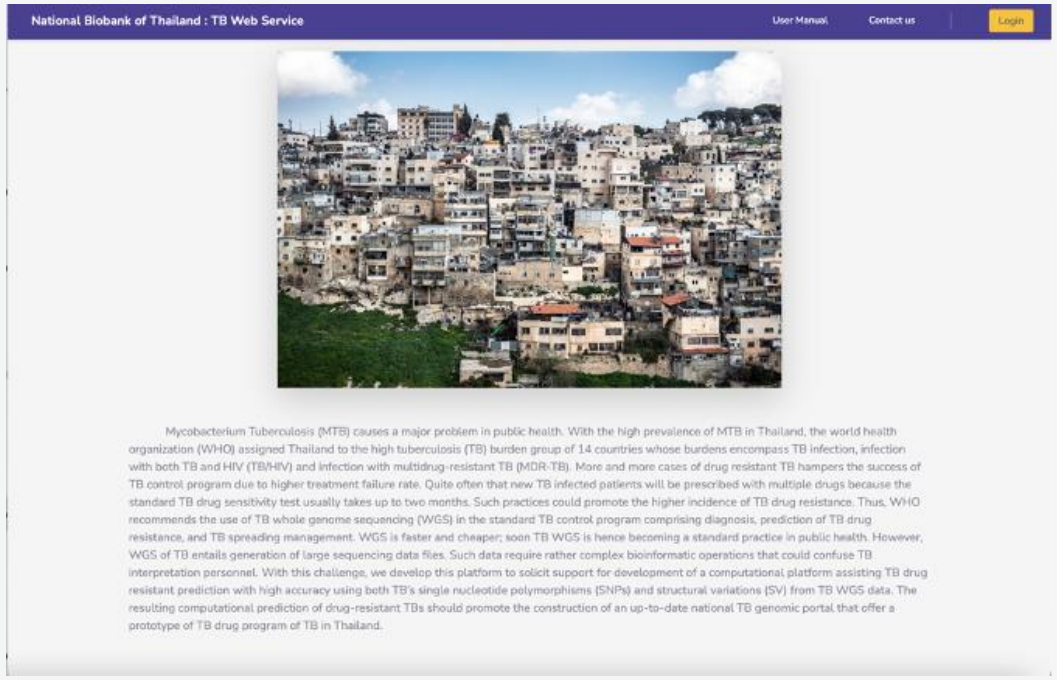


Other platform services

Infectious disease platform

https://mtb.nbt.or.th

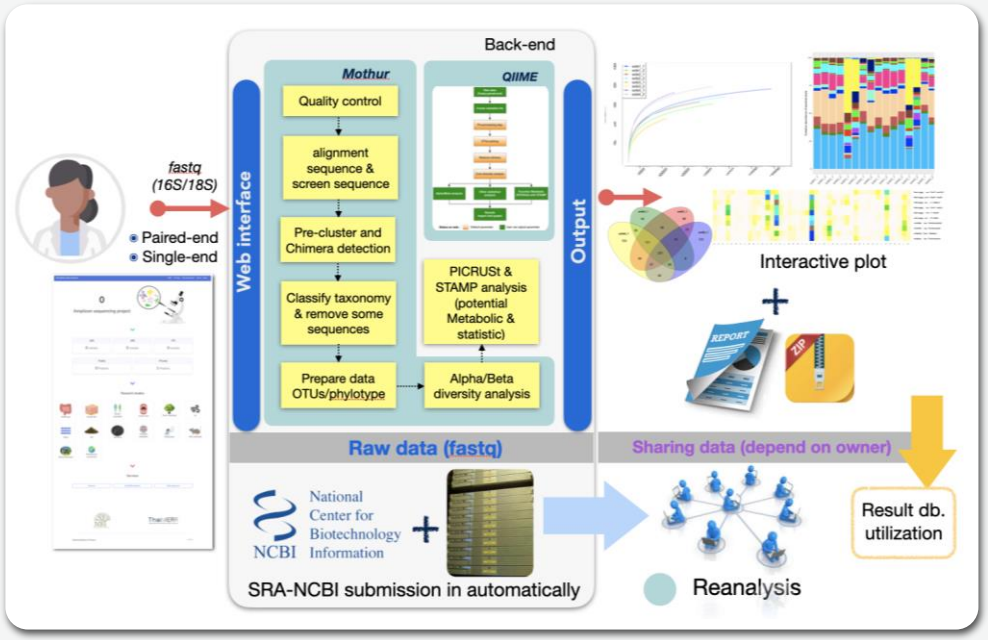
MDR-Tuberculosis



Metagenomics

https://ampmet.nbt.or.th

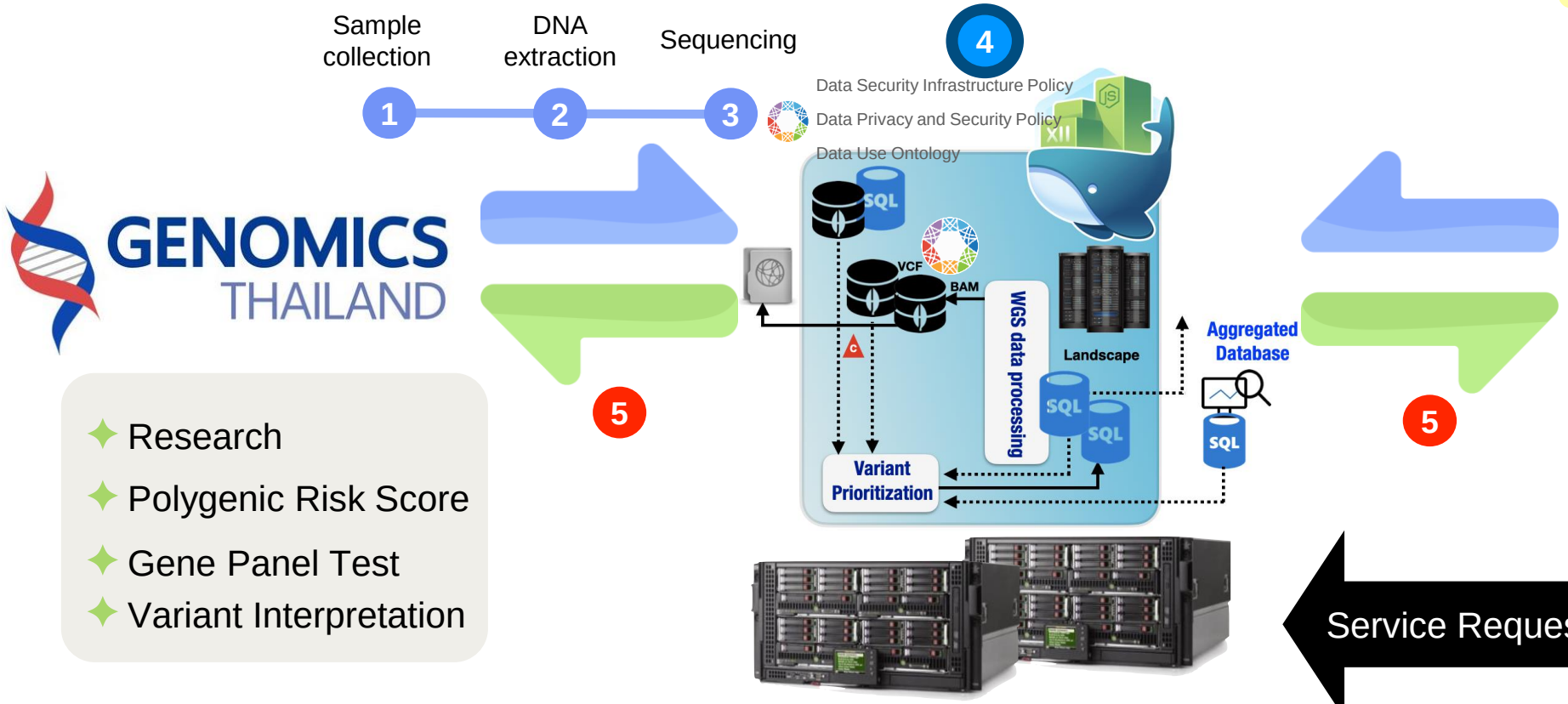
Amplicon Metagenomics Web Analysis (AmpMet)



Computational Genomic Platform

NBT/NSTDA as a ***service enabler***

Genomic-related Businesses



- Research
- Polygenic Risk Score
- Gene Panel Test
- Variant Interpretation

- Wellness
- Nutrigenomics
- Pharmaco
- Medicine

Service Requests/API

GeTH Policy

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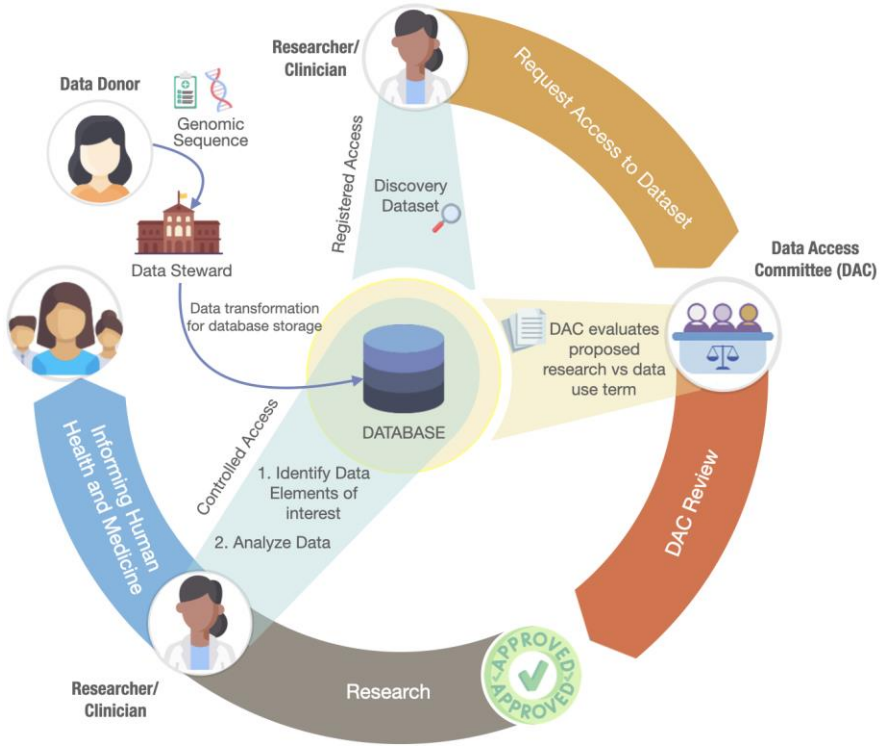
IP Policy

- Access Use data and restricted data
  - Web-based interface
  - Jupyter Notebook
- Data sharing
  - Data Access Committee (DAC)
  - Data stay local policy:
    algorithms move to data analysis
  - A reading library not a lending library:
    can not remove the data
  - Equal Data Governance and Securities policy standard
- Intellectual Property

Data Management policy

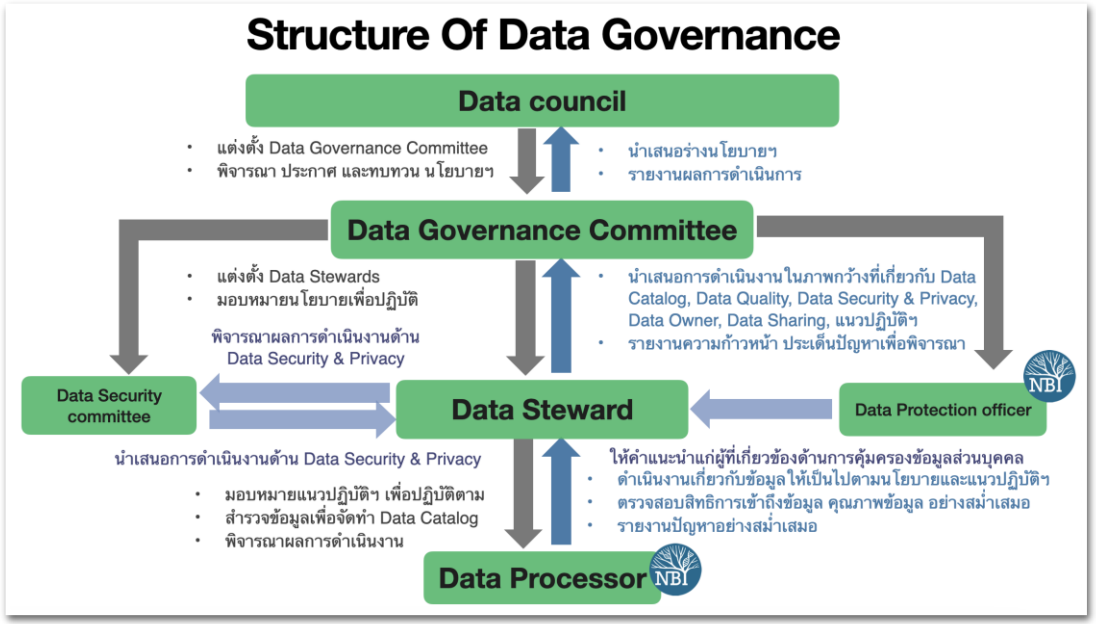
- GeTH Data Governance policy
- Data Security Infrastructure
- Data Privacy and Security
- Return of Results Policy



Genomics england

Global Alliance for Genomics & Health

Collaborate. Innovate. Accelerate.



Acknowledgement

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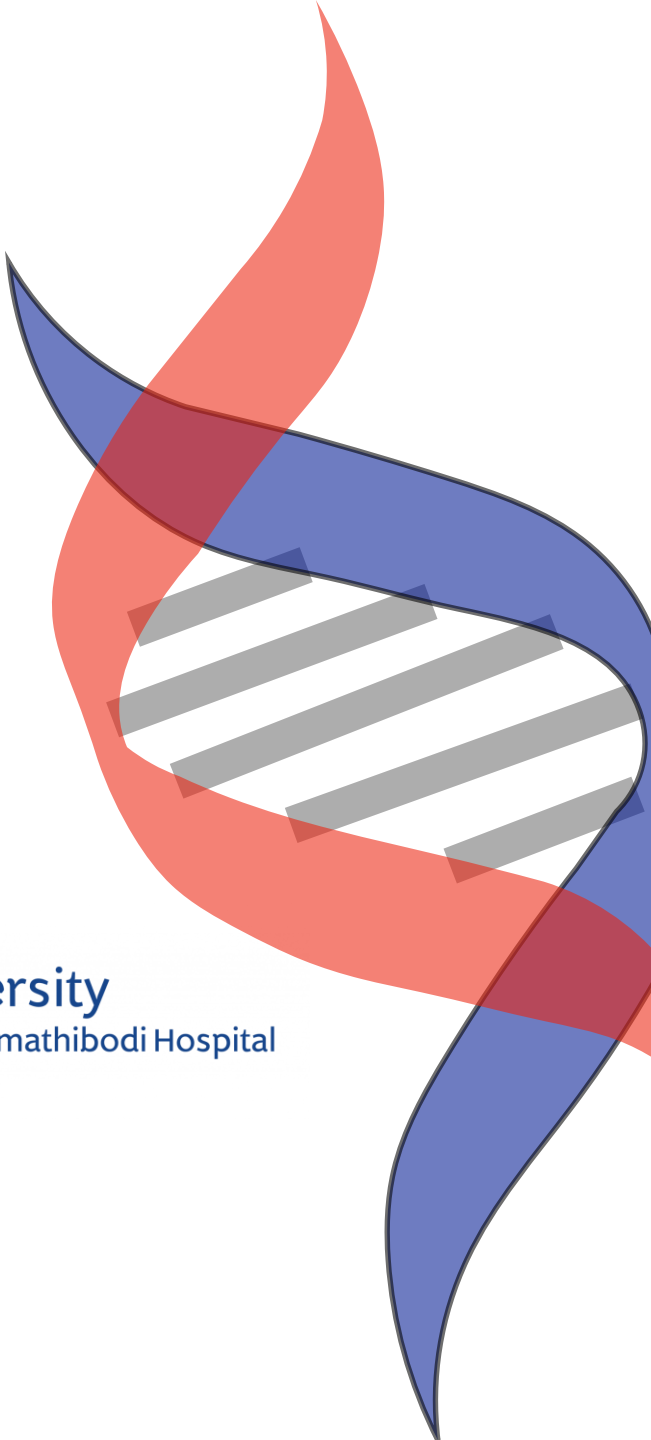

GENOMICS THAILAND

EEC
เขตพัฒนาพิเศษภาคตะวันออก
เชื่อมโลก ให้ไทยแล่น

สวทช NSTDA

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Department of Medical Sciences

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คณะแพทยศาสตร์

ขอบคุณครับ