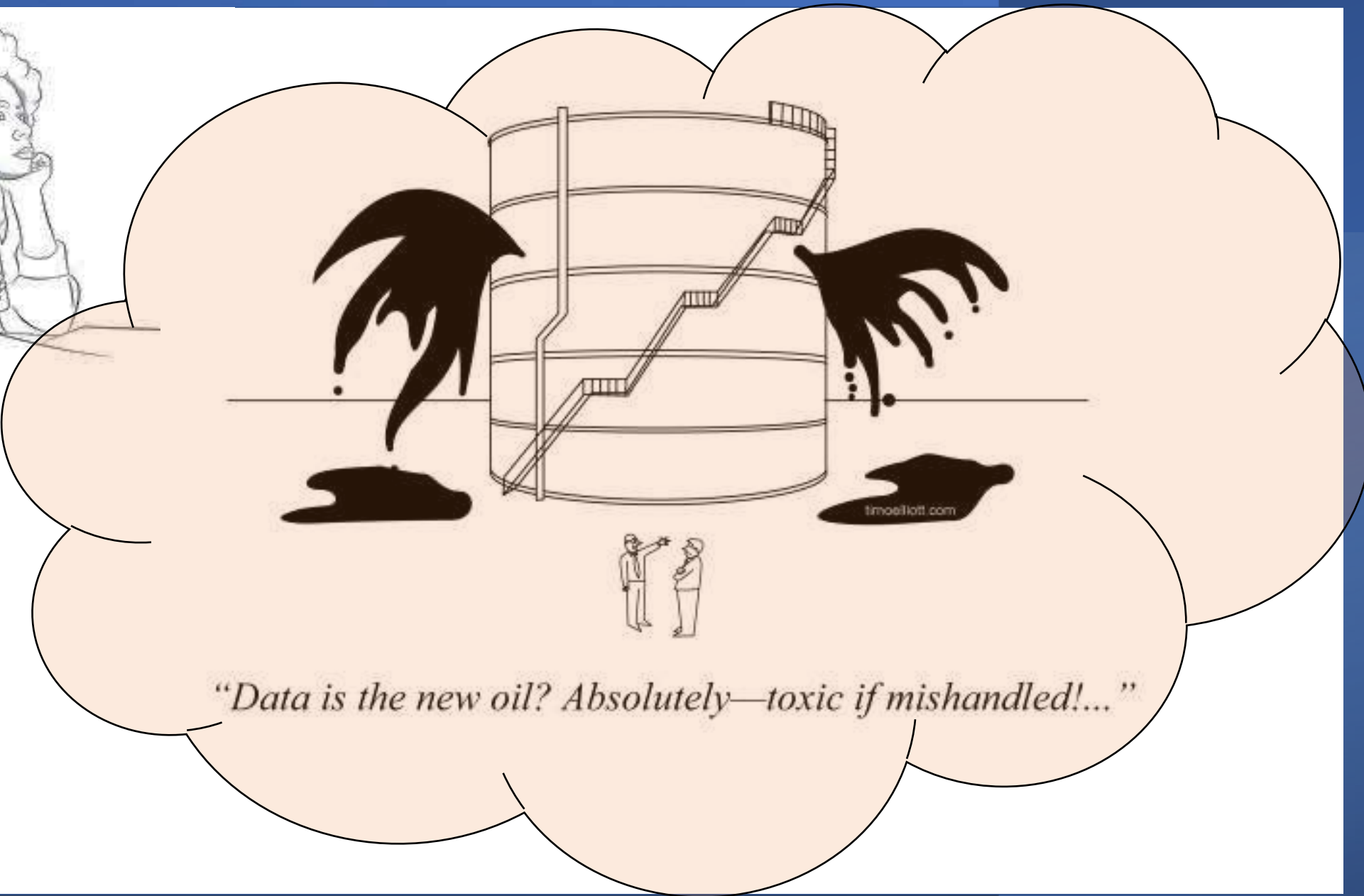


# Data to Real World Impact: A Journey with Purpose

Neerja Karnani  
Deputy Director (Clinical)  
Bioinformatics Institute, A\*STAR  
[neerja\\_karnani@bii.a-star.edu.sg](mailto:neerja_karnani@bii.a-star.edu.sg)





*“Data is the new oil? Absolutely—toxic if mishandled!...”*

# Data to Real World Impact: Our End-to-End Approach

DATA	INTEGRATION	ANALYZE	REAL WORLD IMPACT
<ul style="list-style-type: none"> <li>• High quality</li> <li>• Multi-dimensional</li> <li>• Adequate Sample Size</li> <li>• Secure interphase for storage and analysis</li> </ul>	<ul style="list-style-type: none"> <li>• Discovery database and hypothesis playground</li> <li>• Field/Disease specific use cases</li> <li>• Cross-Population/cohort</li> </ul>	<ul style="list-style-type: none"> <li>• Develop problem statements</li> <li>• Use deep domain knowledge to derive meaningful insights</li> </ul>	<ul style="list-style-type: none"> <li>• Disease Risk Prediction and Surveillance</li> <li>• Clinical application</li> <li>• Evidence based intervention</li> </ul>

Trusted partner in the ecosystem with multiple collaborations with key stakeholders



Prevention

to

Treatment

Early life development

Function and Performance (Adults)

Healthy longevity



GUSTO, S-PRESTO, NiPPeR



GUSTO, S-PRESTO, NiPPeR, iDAD\_SG, TEAMS Study, ATTRaCT, PRISM, HELIOS, SERI, TTSH, MEC, PREPARE



MEMOSA, SG10K Health, SG90, SCHS, DaHA

Clinical Data

Multi-Omics Data

Metabolic Health

Cardiovascular

Disease



Mental Wellness

Immune Health



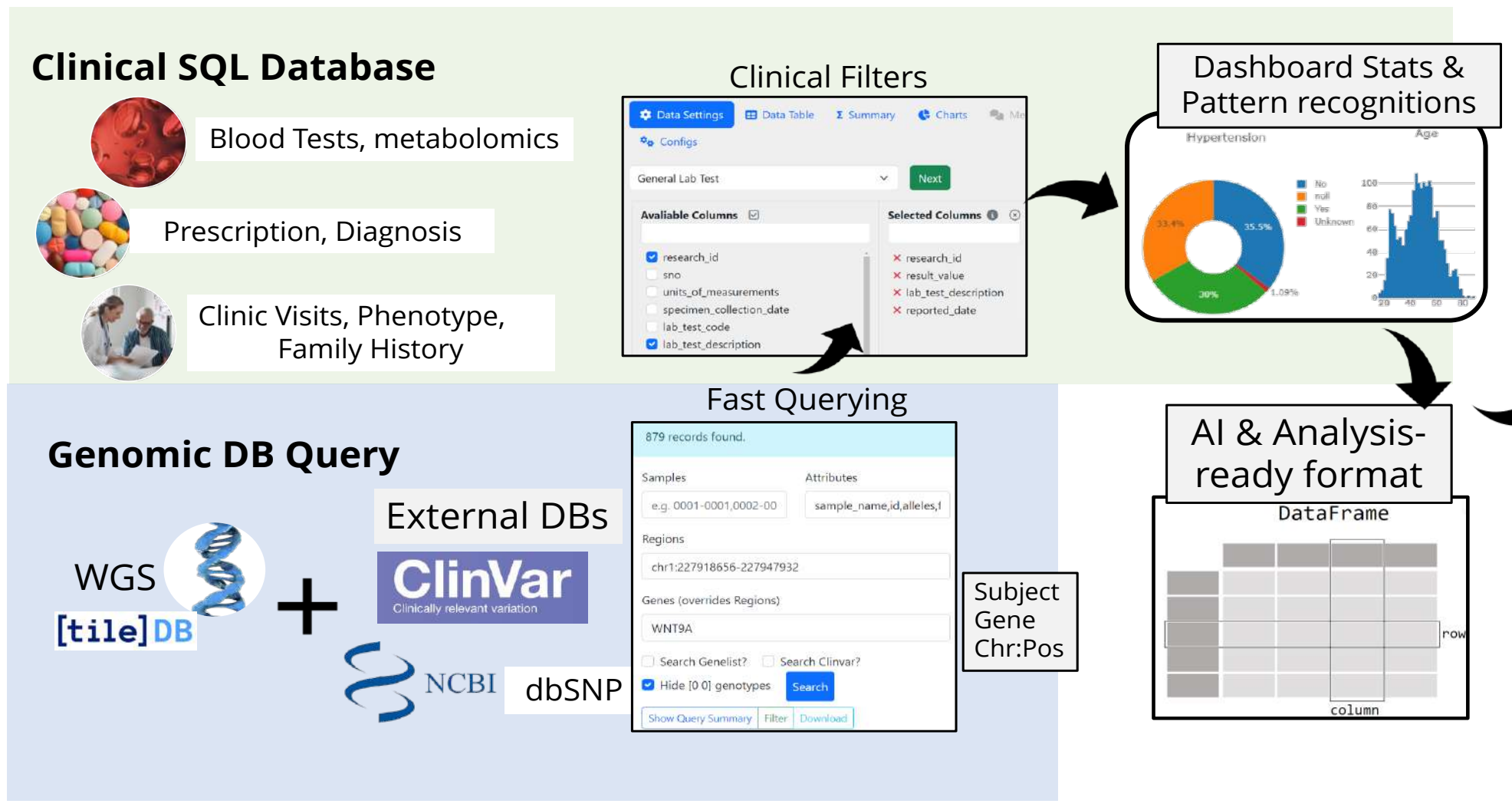
>80 publications

3 patents and Industry licenses

Genetics (GWAS & Polygenic Risk Scores)  
Epigenetics (EWAS & EpiClocks)  
Transcriptomics (Coding & noncoding)  
Lipidomics  
Metabolomics  
Microbiome



## Genetic Counsellor App



## Genomic DB Query

WGS [tile]DB + External DBs

ClinVar: Clinically relevant variation

NCBI dbSNP

# Use case 2 Multi-platform Engagement and Data Integration for a Unified National Goal



**PREPARE**

Map of Singapore showing hospital locations across West, Central, and East regions. Logos include NUH, Tan Tock Seng, Singapore General Hospital, and Singhealth.

### Virus genome surveillance

BII has global (GISAID, WHO, CEPI) and national role (NCID, MOH)

Sebastian Maurer-Stroh

National Centre for Infectious Diseases → Bioinformatics Institute

**REDCap** Research Electronic Data Capture

Demographics Exposures

Editing existing Unique patient code SUBJ0001.

Unique patient code: SUBJ0001

Month and Year of Birth: 25-02-1979

Singaporean; Permanent Resident; Long-term pass-holder; Short-term pass-holder: Singaporean

Residency if Other: Text: [ ]

Living situation (majority of time in last 2 weeks): Others

Other living situation, specify: Text: [ ]

Penny Chan  
Vachiranee Limviphuvad

### Inter-individual variation in Infection Severity

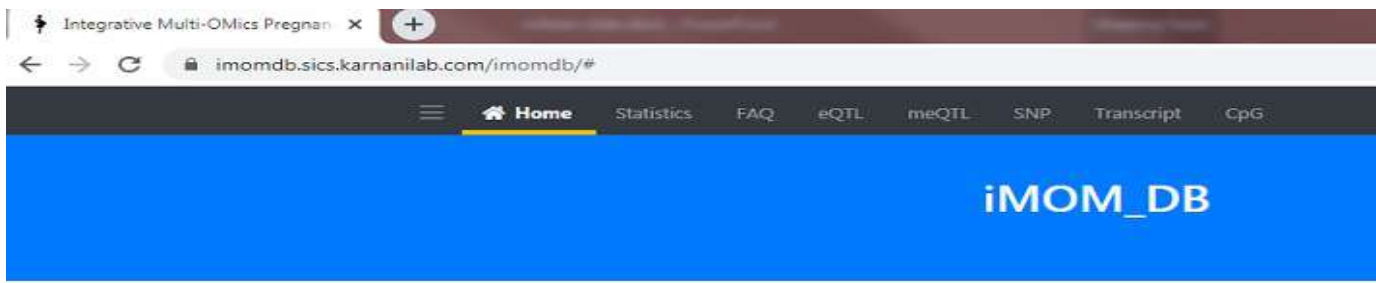
SG10K Health, MINISTRY OF HEALTH SINGAPORE, TRUST

Same format, 9-hospital scale  
Enables cross-hospital longitudinal patient tracking

Automated CSV data import

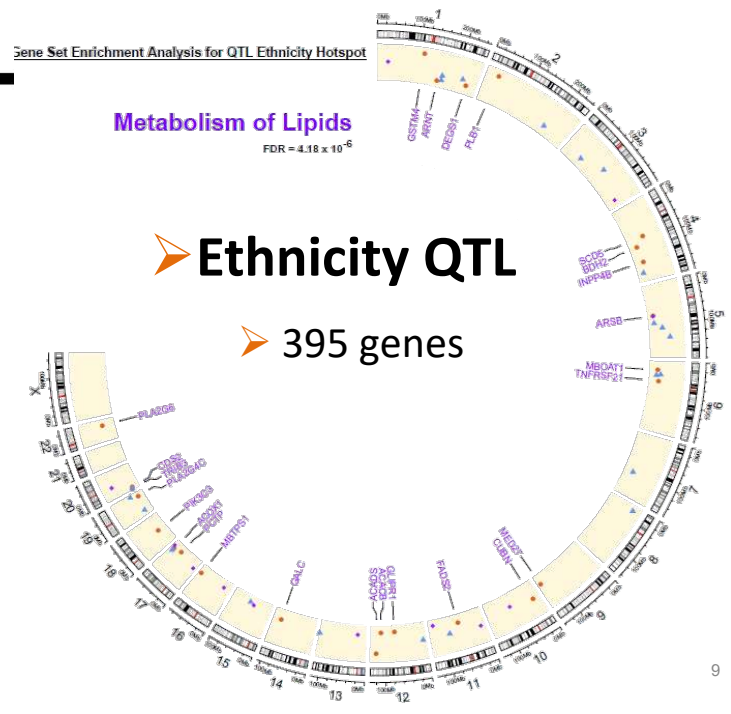
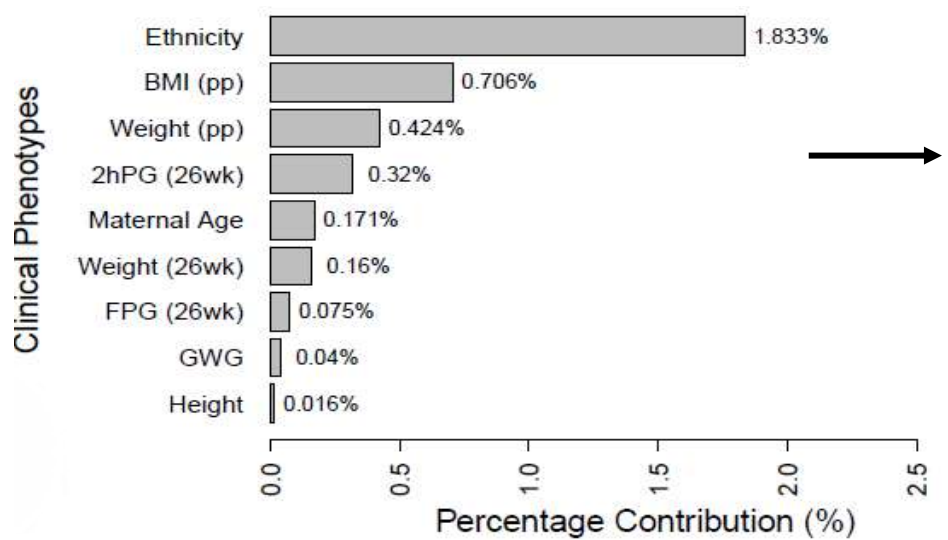
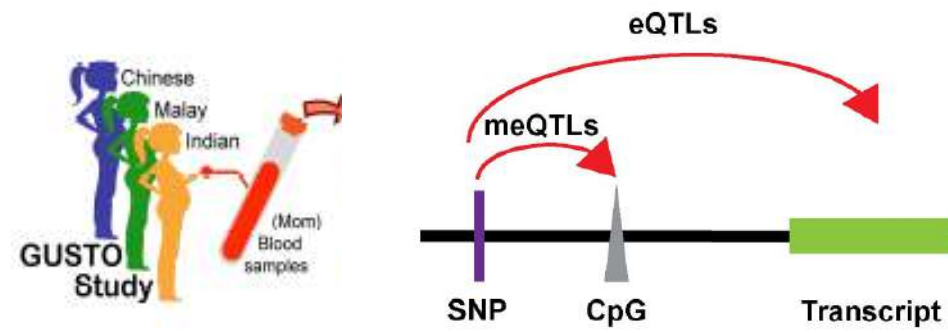


# Use case 4 Integrated Multi-Omics Database (iMOM\_db) of Asian Pregnant Women

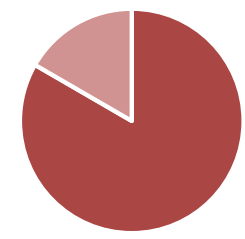


**What is included in iMOMdb?**  
 iMOMdb consists of ethnicity based genome-wide association (GWAS), epigenome-wide association (EWAS) and transcriptome-wide association (TWAS) results. To identify potential molecular interactions between genetic and epigenetic mechanisms, Quantitative Trait Loci (QTL) information and their association with ethnicity were also made available. Most importantly, iMOMdb is open accessed with

Hong, Tan, Lim, Huang and Karnani et al. Human Mol Genet 2022

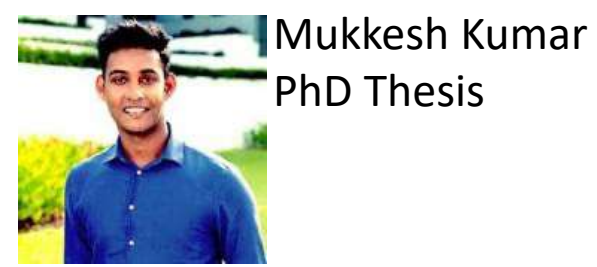


Plasma lipidome (validation)



83.3% of lipids detected sig. assoc. with ethnicity





50% GDM cases routinely missed in SG



> *Diabetes Res Clin Pract.* 2022 Feb 4;185:109237. doi: 10.1016/j.diabres.2022.109237.  
Online ahead of print.

## Population-centric risk prediction modeling for gestational diabetes mellitus: A machine learning approach

Mukkesh Kumar<sup>1</sup>, Li Chen<sup>2</sup>, Karen Tan<sup>2</sup>, Li Ting Ang<sup>3</sup>, Cindy Ho<sup>3</sup>, Gerard Wong<sup>2</sup>,  
Shu E Soh<sup>4</sup>, Kok Hian Tan<sup>5</sup>, Jerry Kok Yen Chan<sup>6</sup>, Keith M Godfrey<sup>7</sup>, Shiao-Yng Chan<sup>8</sup>,  
Mary Foong Fong Chong<sup>9</sup>, John E Connolly<sup>10</sup>, Yap Seng Chong<sup>8</sup>, Johan G Eriksson<sup>11</sup>,  
Mengling Fena<sup>12</sup>, Neeria Karnani<sup>13</sup>

UK NICE guidelines showed poor predictability in Singaporean women [AUC:0.60 (95% CI 0.51, 0.70)]. The non-invasive predictive model comprising of 4 non-invasive factors: mean arterial blood pressure in first trimester, age, ethnicity and previous history of GDM, greatly outperformed [AUC:0.82 (95% CI 0.71, 0.93)] the UK NICE guidelines.

## Preconception Predictive Risk Modelling for GDM

## GDM to T2D Risk analysis



Article

### Automated Machine Learning (AutoML)-Derived Preconception Predictive Risk Model to Guide Early Intervention for Gestational Diabetes Mellitus

Mukkesh Kumar<sup>1,2,3</sup>, Li Ting Ang<sup>1,2</sup>, Hang Png<sup>1,2</sup>, Maisie Ng<sup>1,2</sup>, Karen Tan<sup>1</sup>, See Ling Loy<sup>4,5</sup>,  
Kok Hian Tan<sup>4,6</sup>, Jerry Kok Yen Chan<sup>4,5,7,8</sup>, Keith M. Godfrey<sup>9</sup>, Shiao-yng Chan<sup>1,8</sup>, Yap Seng Chong<sup>1,8</sup>,  
Johan G. Eriksson<sup>1,8,10,11,†</sup>, Mengling Feng<sup>3,12,\*,†</sup> and Neerja Karnani<sup>1,2,13,\*,†</sup>

**Machine Learning Derived Prenatal Predictive Risk Model to Guide Intervention and Prevent the Progression of Gestational Diabetes Mellitus to Type 2 Diabetes.** [Accepted for publication in *JMIR Diabetes* on 22 March 2022]

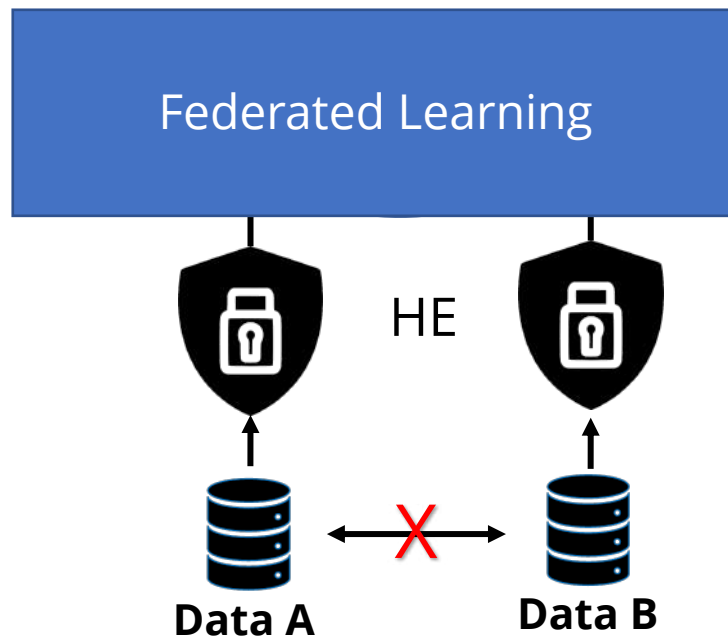
Kumar M, Ang LT, Ho C, Soh SE, Tan KH, Chan JK, Godfrey KM, Chan SY, Chong YS, Eriksson JG, Feng M, Karnani N



# Federated Data Learning - Overcoming Data Sharing Obstacles

## Homomorphic Encryption (HE)

Enables computation of encrypted information without need for decryption



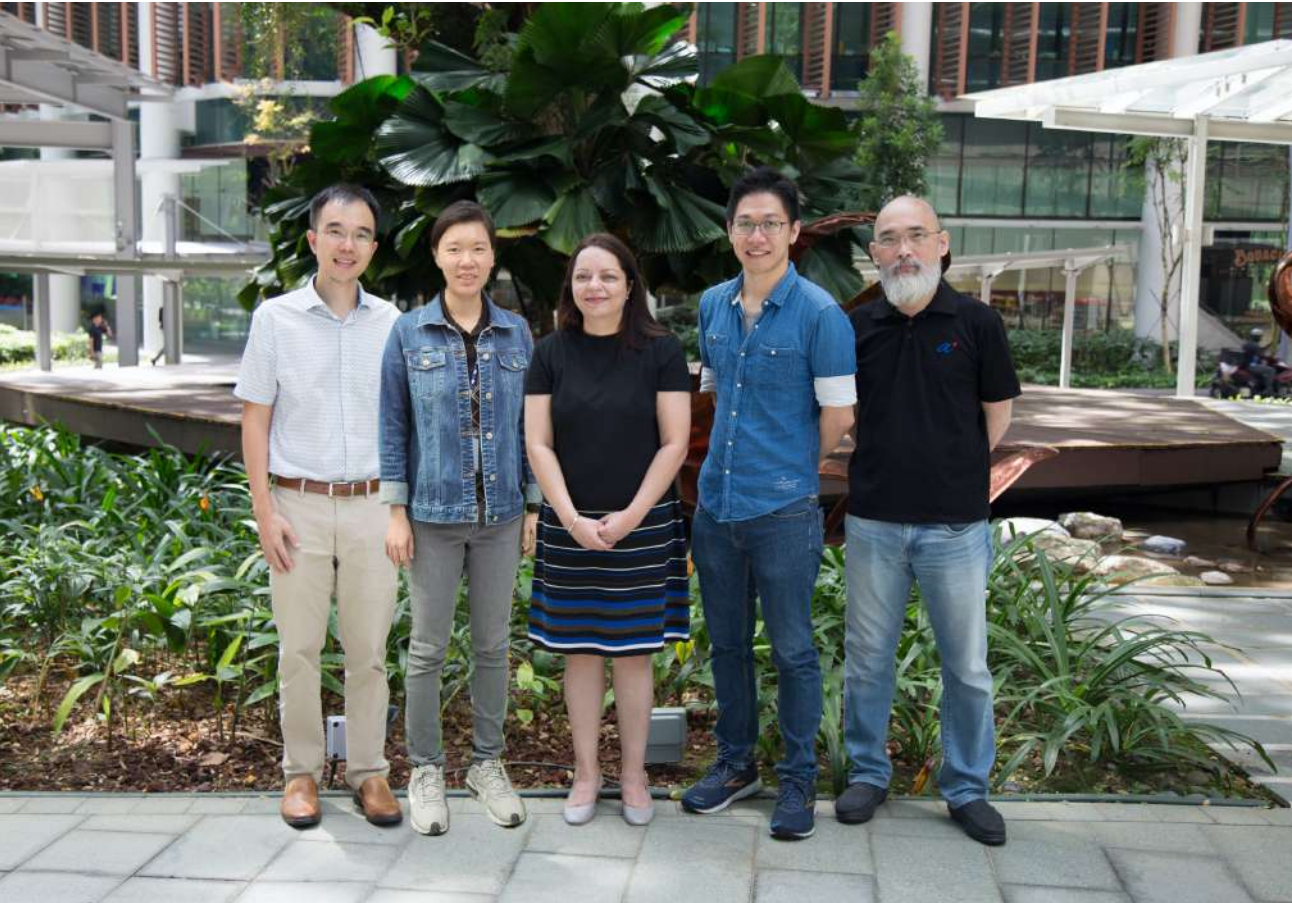
### Benefits:

- ❑ Increased data security owing to HE
- ❑ Reduced regulatory hurdle for local and international cohort data integration

*Example – The Cardiometabolic Health Initiative  
**Polygenic Risk Score calculation**  
Two local cohorts (PRISM & ATTRaCT) and >3000 subjects*



# The Science Mavericks



Kang Qi  
Developing an LSTM Model for Predicting Nutritional Deficiency Diseases with Medication Administration Records

@ SICS



Jia Xu



Kelly Ong



Gong Min



Felicia Tin





CREATING GROWTH, ENHANCING LIVES



# THANK YOU

---

[www.a-star.edu.sg](http://www.a-star.edu.sg)