Establishing a Clinical Genomics Framework at SingHealth
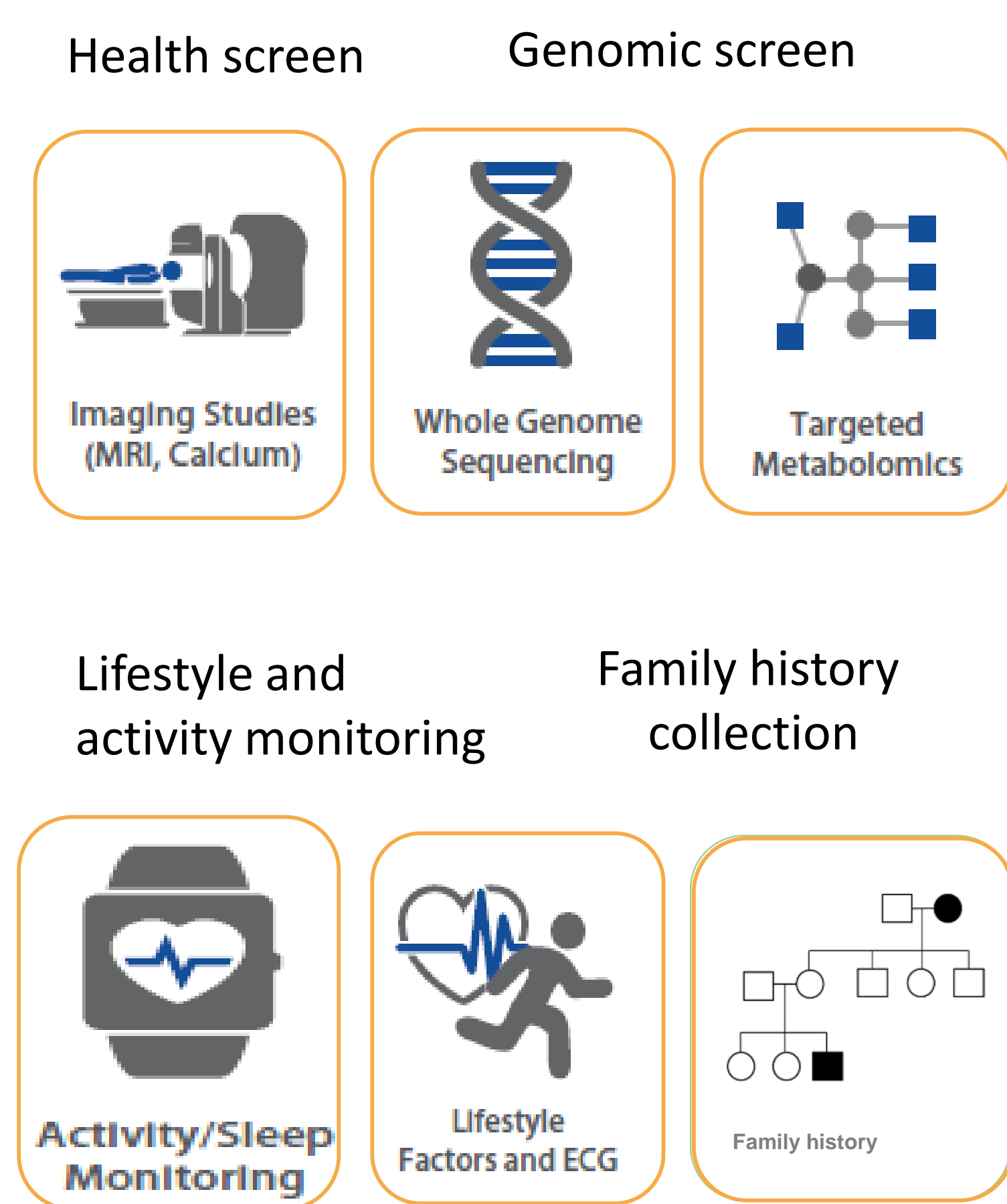
Nellie Chai, Yasmin Bylstra, Sonia Davila

BACKGROUND

Personalised healthcare is now the focus of future medicine. SingHealth Duke-NUS Institute of Precision Medicine (PRISM) aims to promote precision health by providing a comprehensive Clinical Genomics Service from screening to clinical management to improve the healthcare service for Singaporeans.

METHOD

Individuals with no known pre-existing or family history of health conditions are consented to take part in:

Health screen   Genomic screen

Imaging Studies (MRI, Calcium)   Whole Genome Sequencing   Targeted Metabolomics

Lifestyle and activity monitoring   Family history collection

Activity/Sleep Monitoring   Lifestyle Factors and ECG   Family history

PRISM PARTICIPANT PATHWAY

Health assessment
- Genomic screen
- Clinical screen
- Family history

Data stored ← No ← Consent to return results

Yes

Patient data   MDT review
Information resources

Results interpretation

Results disclosure
For probands:
- Issue research report
- Consent for confirmatory testing

Predictive testing for family members

Pathogenic/ Likely pathogenic
- Results appointment arranged

Increased family history
VUS/ likely benign/ benign variants
- Appointment offered or letter sent

Clinical management
- Referral to specialist clinic
- Issue of clinical report

Yearly review at PRISM clinic
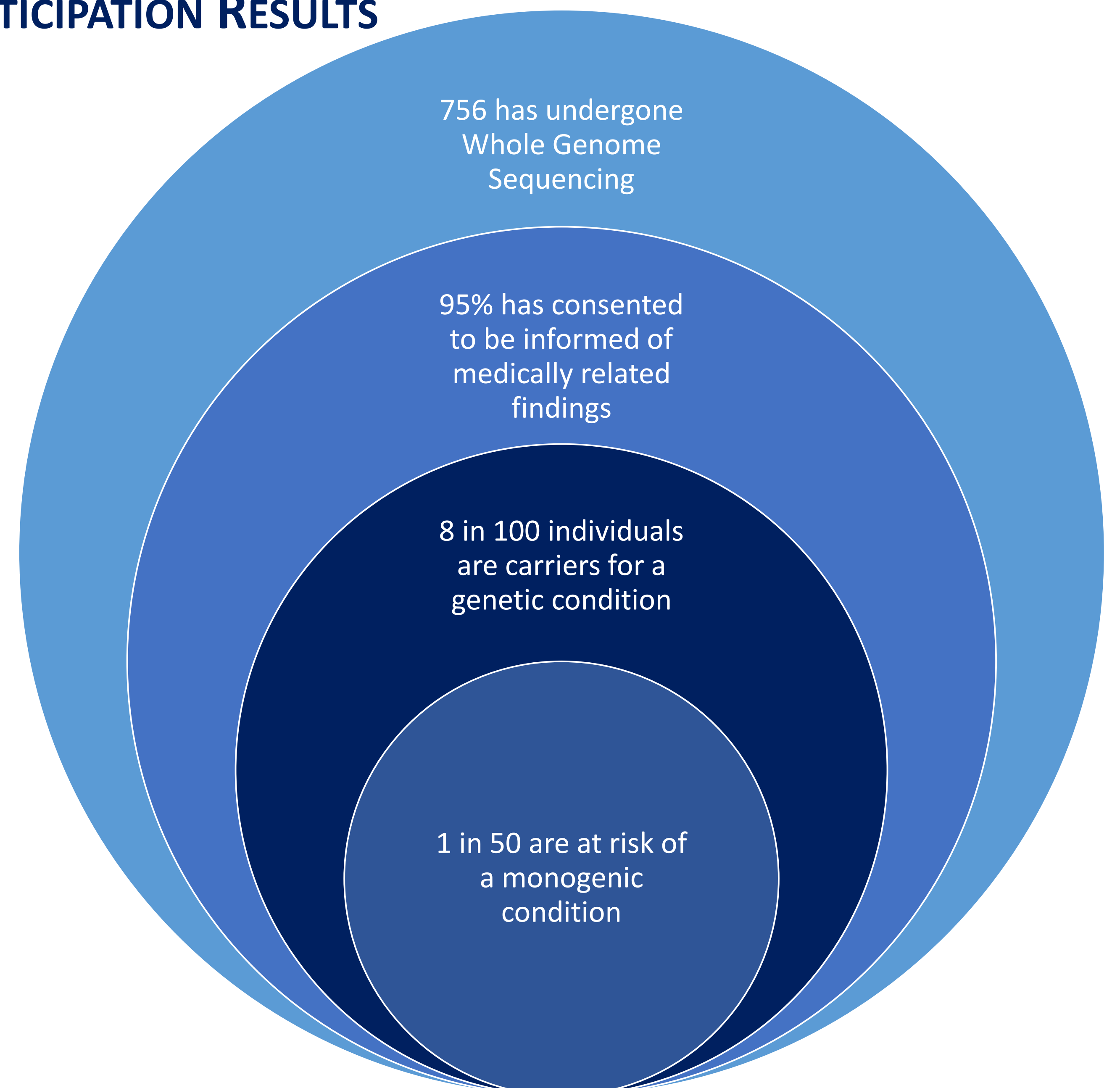
RESULTS INTERPRETATION

The participant data is reviewed in our multidisciplinary team (MDT) meeting comprising of clinical genetics and medical specialists, consultants, bioinformaticians and scientists.

Clinical, Clinical, Clinical, Bioinformatics, Bioinformatics, Genetics, Genetics, Genetics, Genetics
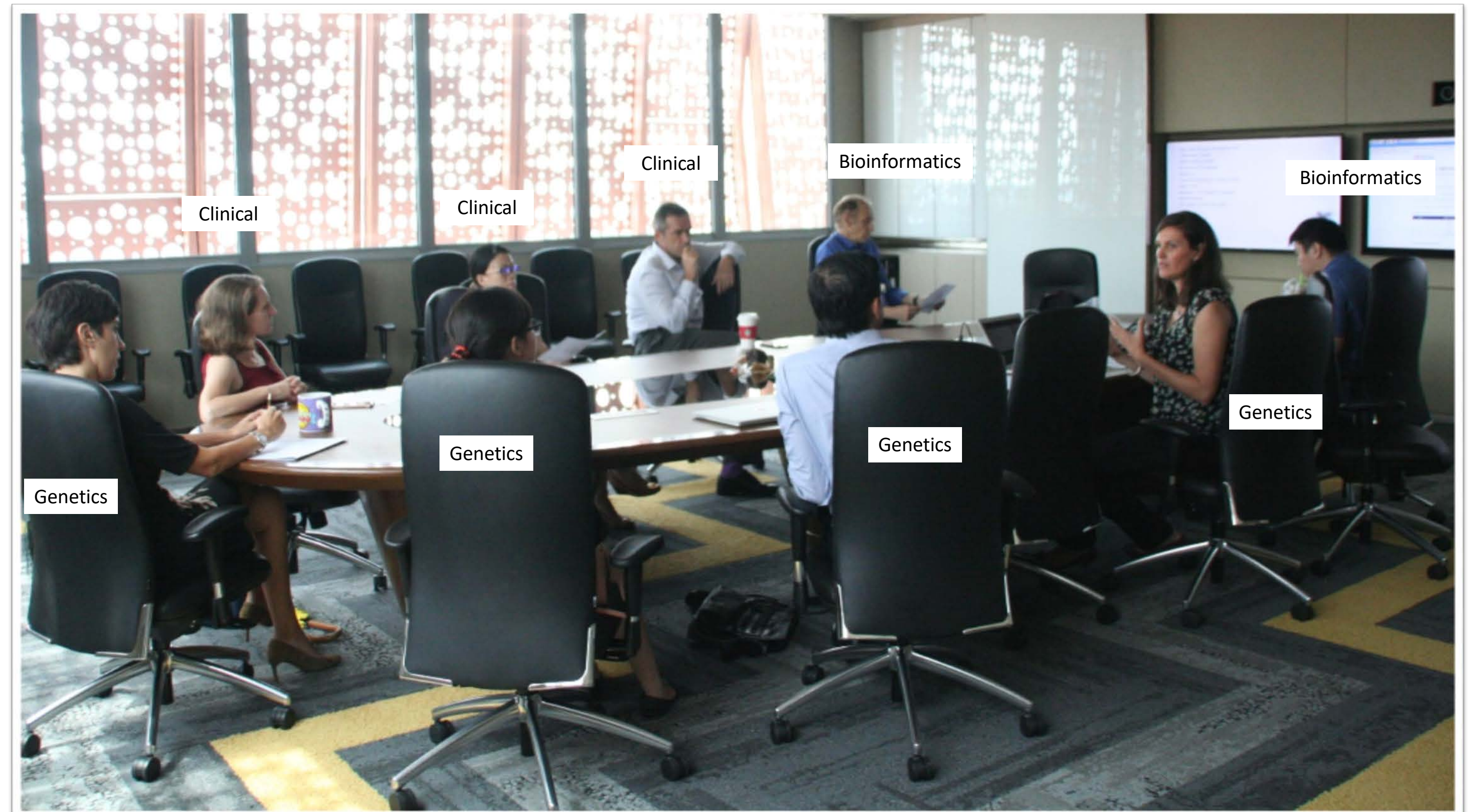
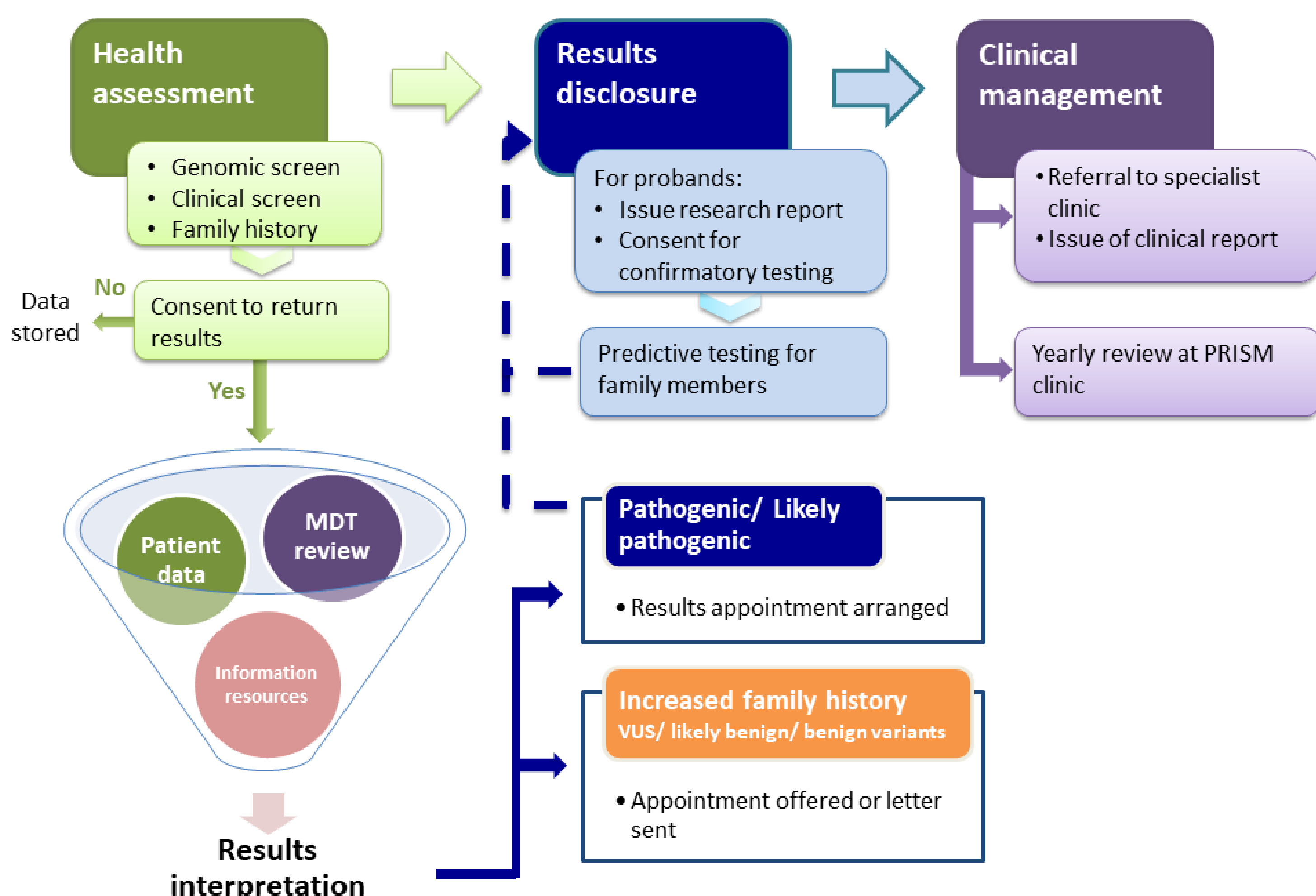## PARTICIPANT ENGAGEMENT AND REFERRAL

For those with clinically actionable results the participants are invited to meet with our genetic counsellors for results explanation and the opportunity to consent for clinical validation. Upon confirmatory testing, participants are referred to a specialist within SingHealth for ongoing health management and are also reviewed yearly with PRISM.

PARTICIPATION RESULTS

756 has undergone Whole Genome Sequencing

95% has consented to be informed of medically related findings

8 in 100 individuals are carriers for a genetic condition

1 in 50 are at risk of a monogenic condition

- Genetic counselling appointment has been offered to 38 participants and of these, 5 have declined.
- 27 participants attended PRISM to receive their genomic results and 13 have consented to clinical validation.

CONCLUSION

We are able to determine the range of Asian normality through detailed phenotyping and genotyping of healthy Singaporeans and have established a systematic clinical framework to identify and return medically significant results. Through our Clinical Genomics Service, we are increasing the efficiency of healthcare management for participants and collecting data to direct health policies.