

Early Detection, Lifetime Protection: Familial Hypercholesterolemia Genetic Testing

Tavintharan S^{1, 2}, Pek S.L.T²

¹General Medicine, ²Clinical Research Unit
Khoo Teck Puat Hospital, Admiralty Medical Centre

Aim

The **FHCARE** study aims to reduce the risk of premature cardiovascular (CV) events within the Familial Hypercholesterolemia (FH) population.



Facilitate genetic testing and cascade screening in clinics for efficient and early detection of FH.



Assist physicians to provide optimized treatment plans for FH patients faster.



Empower patients and healthcare professionals (HCPs) with the knowledge to improve clinical care and treatment adherence.

We plan to achieve this through these sub-aims:

Background

FHCARE is a program established in 2015 to screen patients with very high cholesterol and family history of high cholesterol and premature cardiac diseases for FH. With the commencement of the Clinical Implementation Program (CIP) in 2023, we established a more organized FH research genetic test workflow for patients and cascade screening of family members (FMs). This was done through organizational collaborations and genetic counselling training.

Team Members

Name	Designation	Department
Sanjaya Dissayanake	Senior Consultant	General Medicine
Jeremy Hoe Kian Ming	Consultant	General Medicine
Justin Tang I-Shing	Consultant	Cardiology
Atiqa Binte Zulkifli	Research Coordinator	Clinical Research Unit
Madhumetaa D/O Selvakumar	Research Coordinator	Clinical Research Unit
Nurhusna Alifia Binte Suhaimi	Research Coordinator	Clinical Research Unit
Sharmaine Low	Research Coordinator	Clinical Research Unit
Chen Hoe Meng	Research Officer	Clinical Research Unit
Nike Pui You Qi	Research Assistant	Clinical Research Unit

Interventions / Implementation

1. Standardized clinical workflow to streamline patient's journey for FH genetic testing



2. Partnering with CAP accredited labs: Lifestrands & Ambry



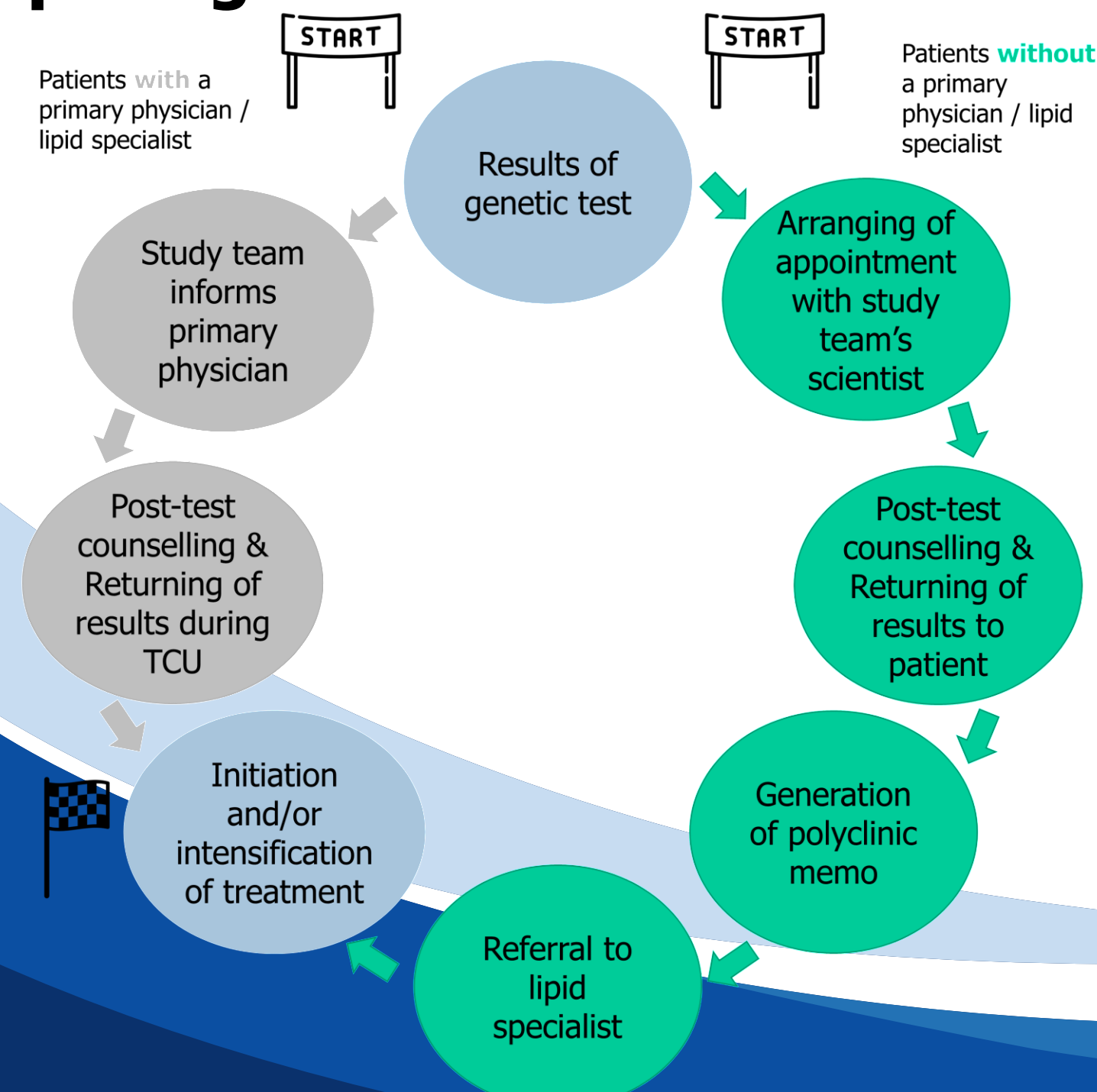
- Faster turn-around time for genetic test results.
- Generation of standardized clinical reports.

3. Training of study team members for genetic counselling

Pre-test counselling	Post-test counselling
Physicians	Physicians
RCs	Scientist

- Relevant checklists in place.
- Improve patient communication.

4. Providing continuity of care post-genetic test



5. Engagement with various stakeholders and members of public.

- Review case studies, research data & its implications on clinical care with patient's primary care team.
- Raise awareness through collaborations.



Onward 2026

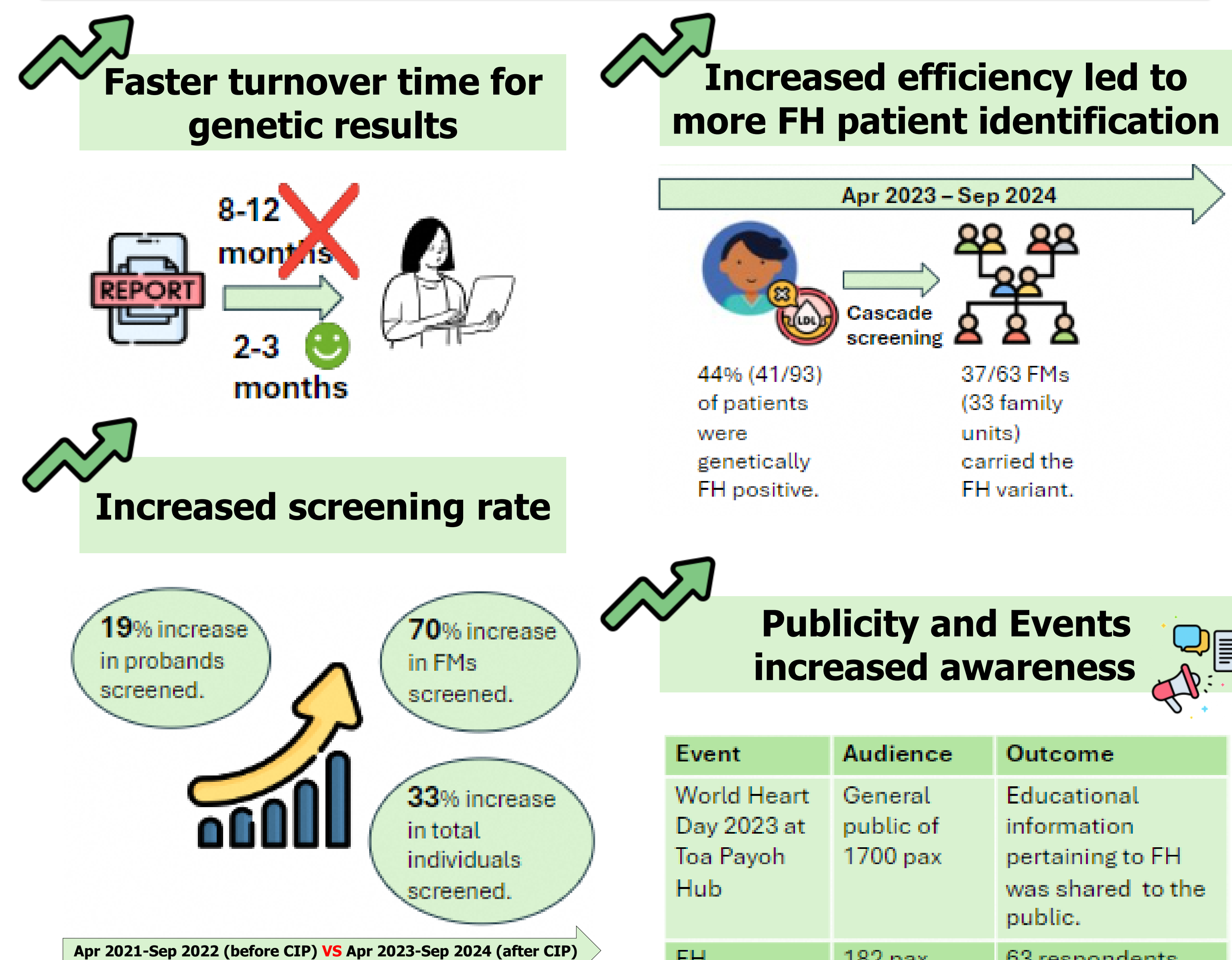
FHCARE works with doctors to identify & diagnose FH patients & **assign them to care streams** for timely & optimal treatment.

Our centralised team of trained RCs handling referrals for FH genetic testing ensures **operational resilience**.

- FH genetic testing enables accurate diagnosis.
- Aggressive treatment initiated in this high-CV risk group.**
- Cascade screening program identifies new FH patients. **We direct them to receive specialized lipid care.**
- FHCARE are trained in genetic counselling, allowing Drs to **focus on FH clinical care.**
- Research insights highlighted to Drs.
- Promotes **enhanced patient care.**

- Prevents double/unnecessary testing and appointments for FH patients.
- Reduced consultation time.
- More patients can be seen.
- Prevention of CVD in early stages = Reduced future A&E admissions.
- Reduced future bed crunch and waiting time for other emergencies.

Results & Outcomes



Increased training & understanding among healthcare professionals leading to better informed patients.

Conclusion

As a centralized referral hub, with a standardized workflow and adequate training, we improved the efficiency of providing FH genetic testing to patients and their family members. The study will continually focus to improve long-term FH-management and health-outcomes through FH-educational initiatives and constant streamlining of workflows when needed. Through this, we can achieve our goal of early detection, lifetime protection of FH patients.