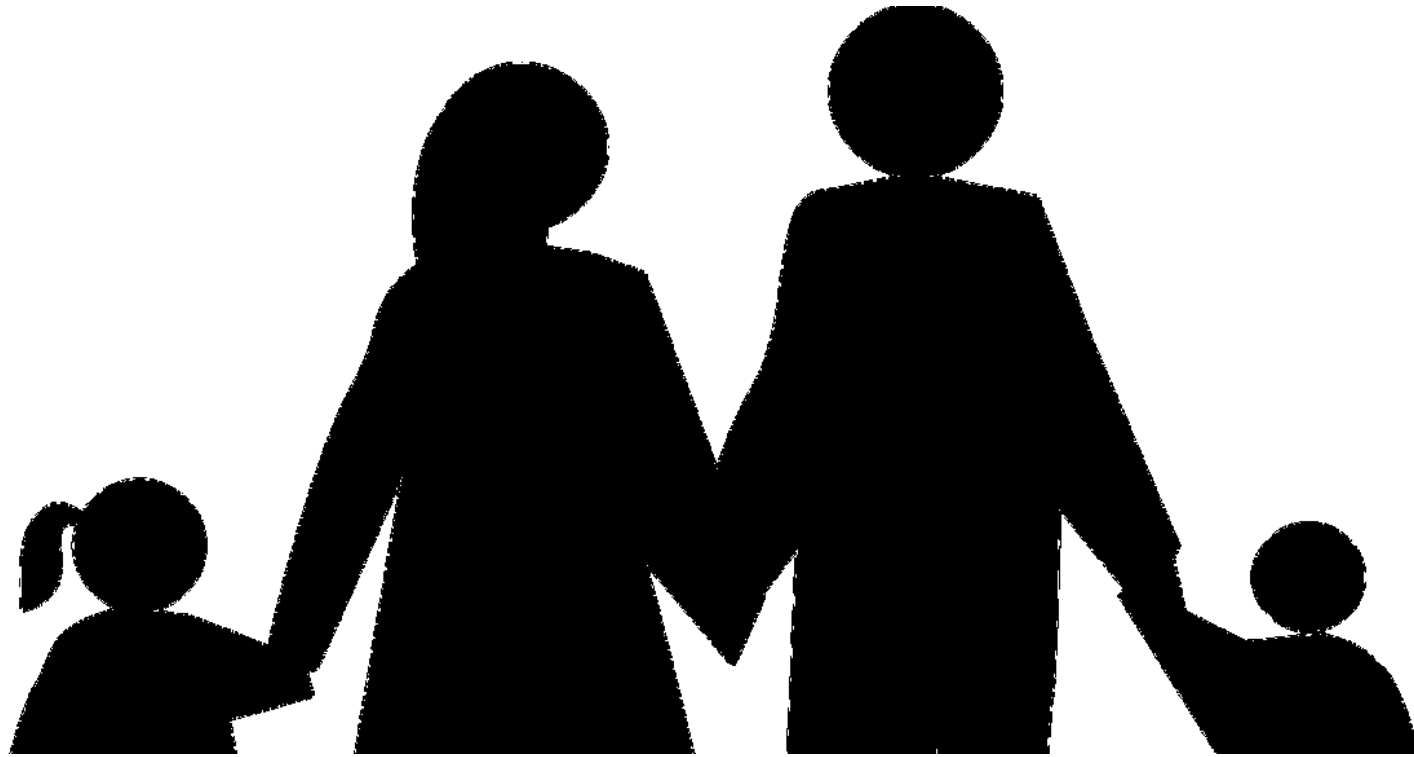# Familial Hypercholesterolemia
Registry and Screening in the Philippines

Cecilia A. Jimeno, MD

Director, Philippine Lipid and Atherosclerosis Society
Professor, UP College of Medicine Department of Pharmacology
Clinical Professor, Division of Endocrinology, Diabetes and Metabolism

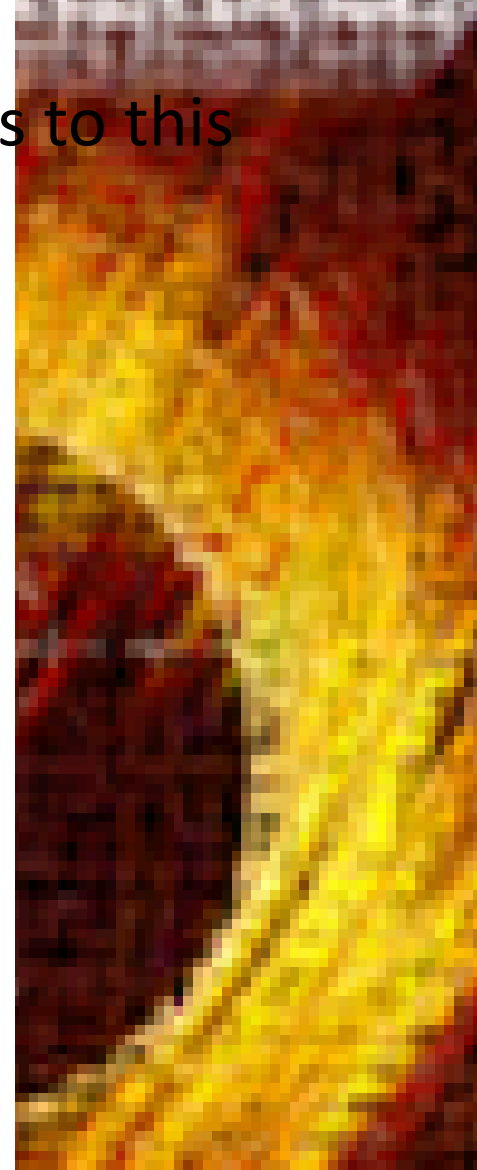# FH²: Filipino Heart for Familial Hypercholesterolemia
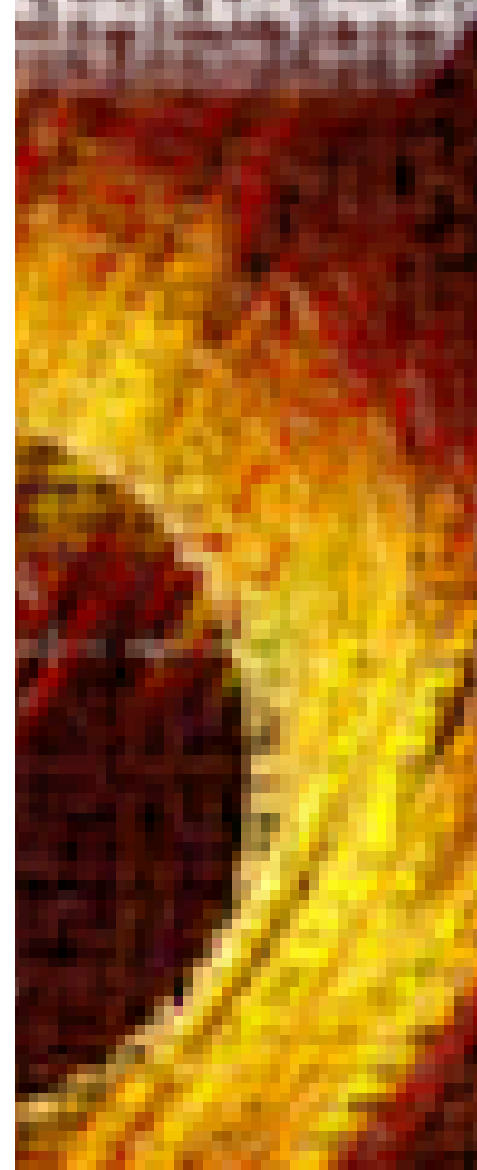

Familial Hypercholesterolemia Research Registry

Disclosures

- The speaker has no conflicts of interest to declare with regards to this presentation.

Outline of the Presentation

- What is the FH Registry Project (FH^2 registry)
- Purposes of Disease or Patient Registries
- Harnessing the Power of the FH Champions
- Networking and Collaboration
- Future Directions

What is the FH Registry Project?

- A patient or disease registry among persons with FH

- Spearheaded by the Philippine Lipid and Atherosclerosis Society (PLAS) in cooperation with various specialty organizations such as the Philippine Heart Association and members of the Philippine Pediatric Society

- Clinical data/patient or disease registries are intended to **evaluate and improve outcomes for a population** defined by a particular condition, disease, or exposure.

Introduction: Why an FH Registry?

FH: Group of genetic disorders of lipid metabolism that leads to **elevated cholesterol levels.**

Preventable cause of early cardiovascular mortality and morbidity.

Have a greater risk of cardiovascular disease because of lifelong exposure to elevated cholesterol levels.

Once thought to be rare, global estimates of prevalence puts it at around 1:200-250.

Introduction: Why an FH Registry?

- FH is considered the **most common life-threatening genetic condition in all races & ethnic groups**.

- No prevalence data in the Philippines.

- Philippines: no existing registry on FH and the vision for this research project is to create one.

- Only 3 researches on FH epidemiology in the Philippines have been previously conducted, and only one has been published.

Introduction: Why an FH Registry?

Cardiovascular disease remain the top cause of mortality & morbidity in the Philippines and FH is probably contributory.

However, despite its disease burden FH remains underdiagnosed & consequently undertreated.

Thus, this research aims to describe the **epidemiologic and clinical profile** of Filipinos with FH.

Introduction: Why an FH Registry?

- By understanding the clinical profile of patients with FH in the Philippines, disease burden may be minimized by **guiding the dev't of guidelines & policies on the screening & detection of FH**, and **implementation of existing guidelines on its management**.
- Data from records of patients who have been previously identified in the hospital or clinic setting will be collected, and
- Prospectively (cascade) screening will be encouraged so that persons with FH can be identified and included in the registry.

Objectives of the Study

- **Primary Objective:** Describe the epidemiologic and clinical profile of Filipino persons with FH

- **Secondary Objectives:**
  - Describe the age of onset or age on diagnosis, sex, family history of early cardiac death or cardiovascular events, socioeconomic level (according to educational level or family income if available), rural or urban, current age, and years from diagnosis.
  - Characterize the clinical presentation (discovered by routine testing or discovered after an acute cardiovascular or cerebrovascular event), comorbid conditions, DLCN score, initial & current treatment, type of doctor treating the patient.

Objectives of the Study

- **Secondary Objectives:**

  – Determine the lipid profile of the Filipino FH patient: baseline Total cholesterol, LDL-cholesterol, Triglycerides, HDL-Chol, and non-HDL cholesterol, and the lipid profile after treatment.

  – Describe the long-term outcomes of persons with FH
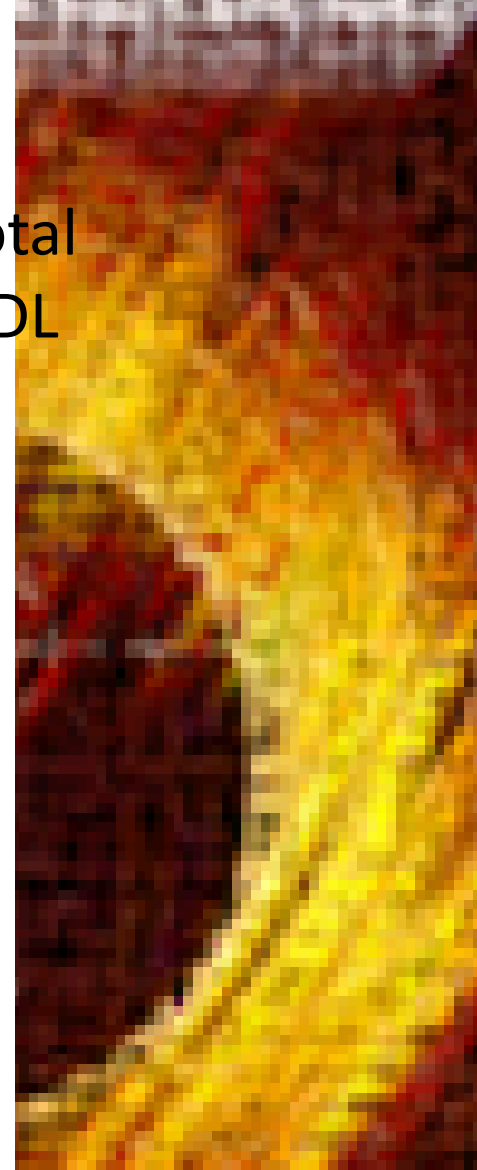
Questions which we hope to answer

- Clinical profile: Demographic characteristics (including numbers of siblings/children both affected and unaffected if known), genotype (if available), treatments, baseline lipids and on-treatment lipid levels.

- Is FH treated by a generalist vs. specialist; how are they being managed and are LDL-c targets being reached?

- FH with pre-existing CVD vs. those without?

- FH with a family history of premature CVD vs. those without?
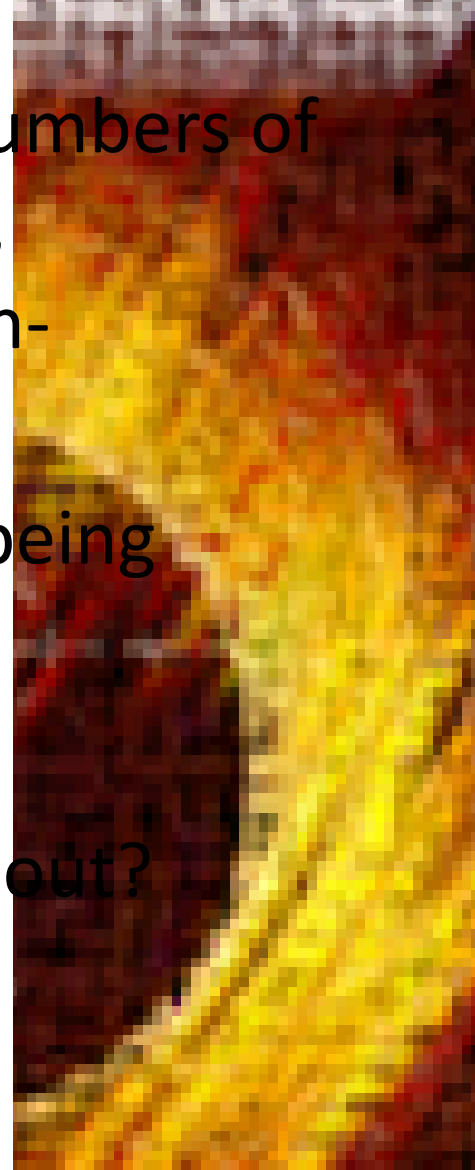
Study Design

Ambispective study design.

Retrospective part : review of records of patients with FH who have been previously identified by the FH Champions clinically through the Dutch Lipid Clinic Network Criteria (DLCNC) Score.

Prospective data collection: from patients who are newly identified to have FH either through the outpatient clinic from for example, cascade screening or from the in-patient hospital records during admission for a cardiovascular event.
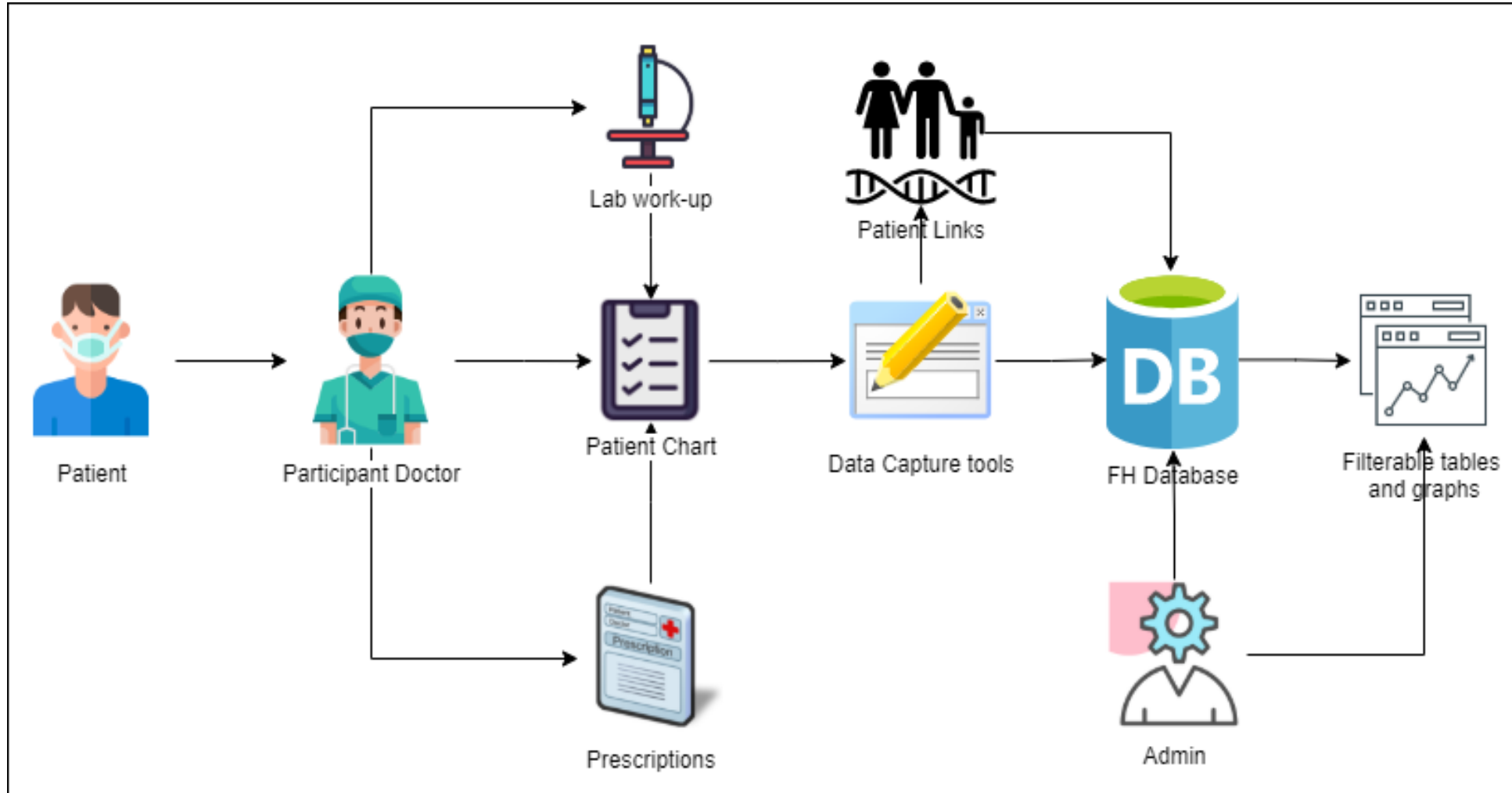
Those participants included in the retrospective part will also be followed up for outcomes.
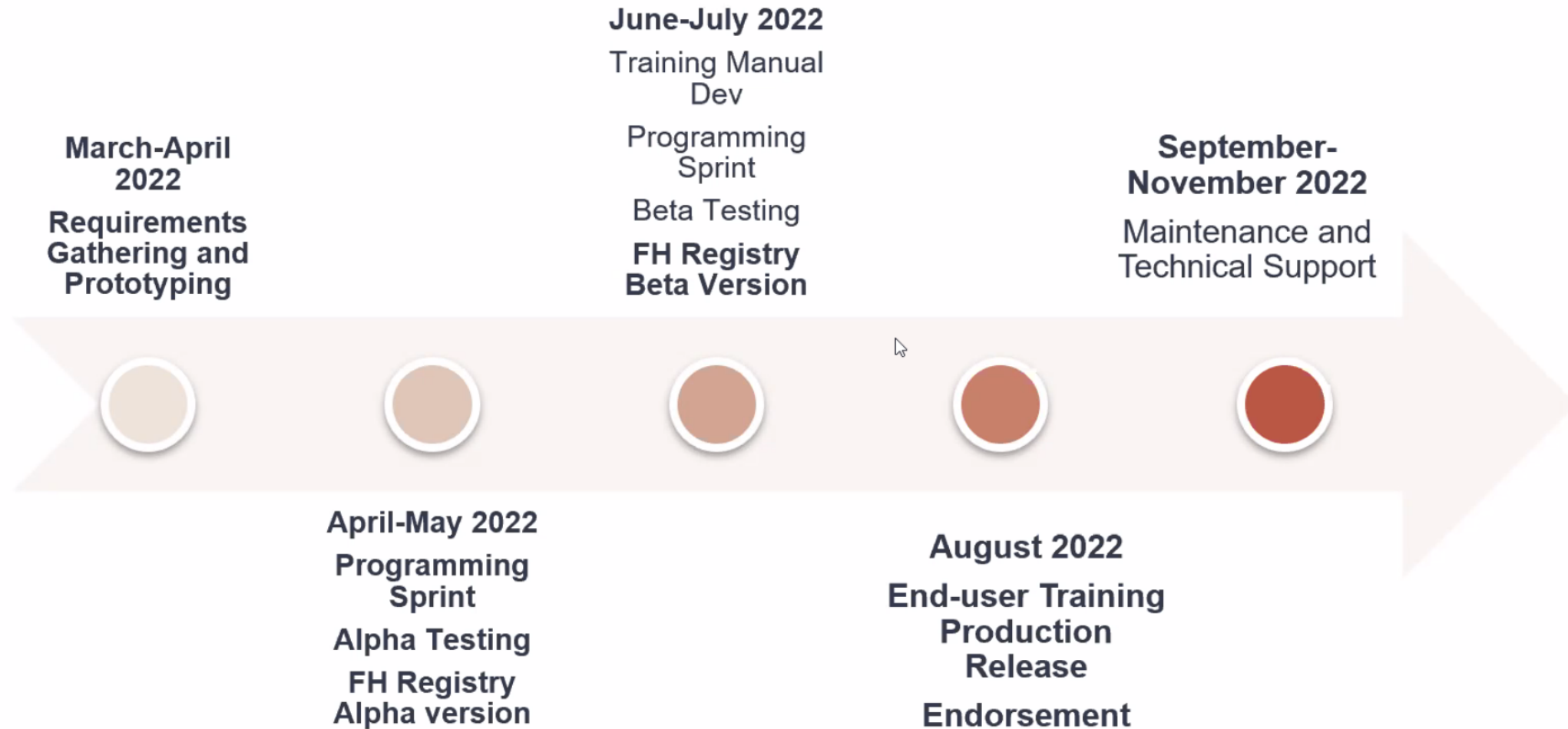
The registry will be developed by the UP Manila National Telehealth Center led by Dr. Francis Raymond Sarmiento.

FH Registry proposed system

Disclaimer: the above workflow and labels are only to depict the plans for the system. The entire context will be finalized during the actual requirements and specifications validation.

Timeline

March-April 2022

Requirements Gathering and Prototyping

April-May 2022

Programming Sprint

Alpha Testing

FH Registry Alpha version

June-July 2022

Training Manual Dev

Programming Sprint

Beta Testing

FH Registry Beta Version

August 2022

End-user Training

Production Release

Endorsement

September-November 2022

Maintenance and Technical Support

Time Lines

- April-June 2022: Set-up of the registry.

- Quarter 3: Ethics Review and Approval

- The registry itself will continue to collect data from all prospectively identified patients and will be maintained for at least the next 5 years.

- The registry itself is still to be developed to allow it to be functional and operational indefinitely.
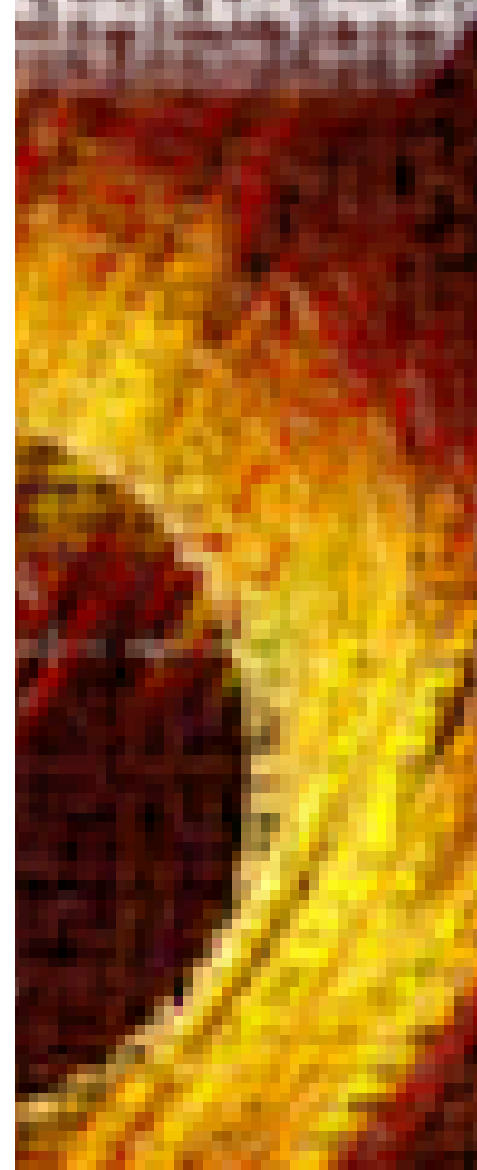
Tasks

- SJREB: will commence soon as the registry is up and running.

- For sites with no local ethics review, submission to the National Ethics Committee (NEC)

- Local Ethics Review will be done parallel to SJREB.

FH
CHAMPIONS
League

- Leaders for **advocacy** in FH: to educate the public & fellow doctors on its importance as a PREVENTABLE cause of early mortality
- **Investigators/researchers** for FH Research

Outline of the Presentation

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Future Directions

- Policy proposals for screening
  - Screen children and teens
  - Screen students prior to entry to university (college)
  - Red flagging or tagging of all LDL-chol levels $\geq$ 190
- Cascade screening for all patients already included in the registry
- Genetic testing for patients included in the registry (15K): funding
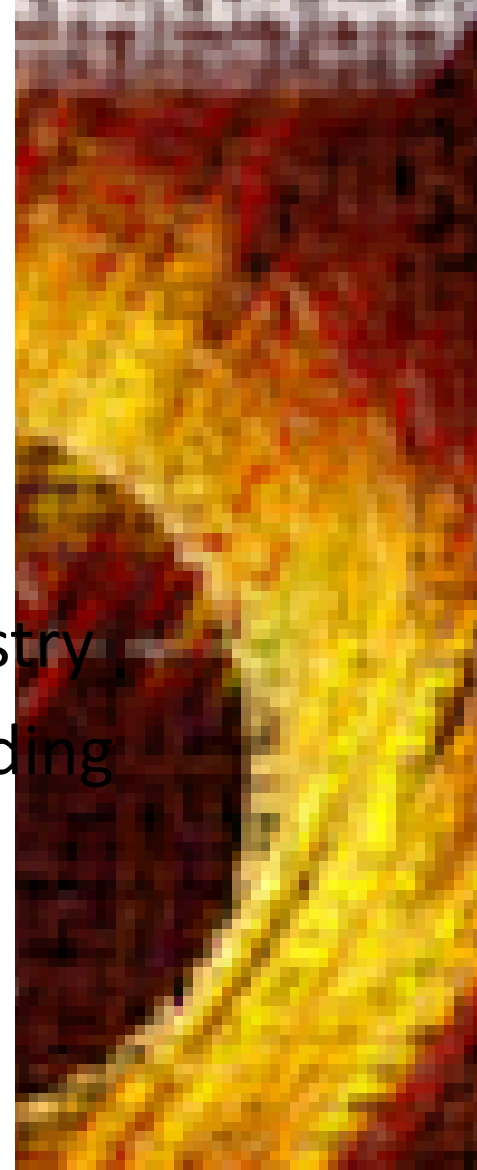
INTERESTED?

Send us an email:

- PLAS FH secretariat c/o Cess Sulit
Email: FHregistry.ph@gmail.com

- **Cecile A. Jimeno, MD**
ceciledoc@yahoo.com;
cajimeno@up.edu.ph

- Identify also your hospital or clinic; interest in advocacy and/or research