

Familial HyperCholesterololaemia

*Commissioning for Better Experience of
Care*

Brief history of Michael, a 37 year old young man



- Michael had a fairly healthy lifestyle, exercised regularly, did not smoke, blood pressure was fine, BMI 23.1
- However, one evening he was taken by ambulance to Accident and Emergency following a collapse.
- Diagnosis: massive heart attack.



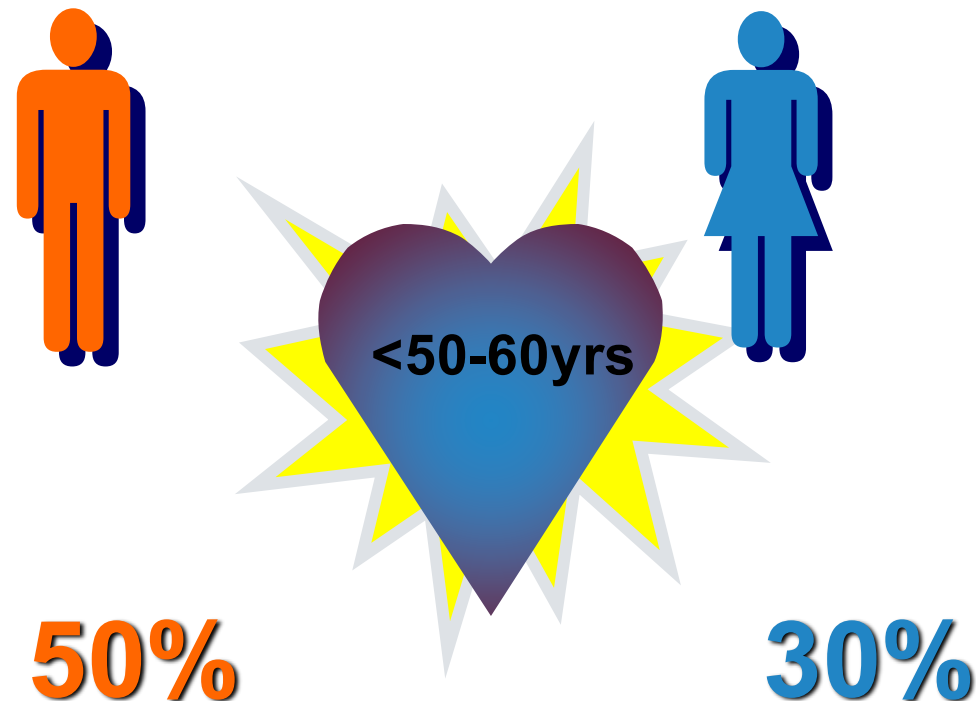
.....Michael.... passed away.

He had a daughter with his longtime fiancé. They had plans to marry four months later.

It was found out that his father also died of a heart attack at 47 years of age.

FH cause Heart attacks at Early Age

50% inheritance – passed to children



Does FH treatment really work ?

	Death mean age	Death mean age
	1978	2021
Without FH	72	79
With FH	50	78

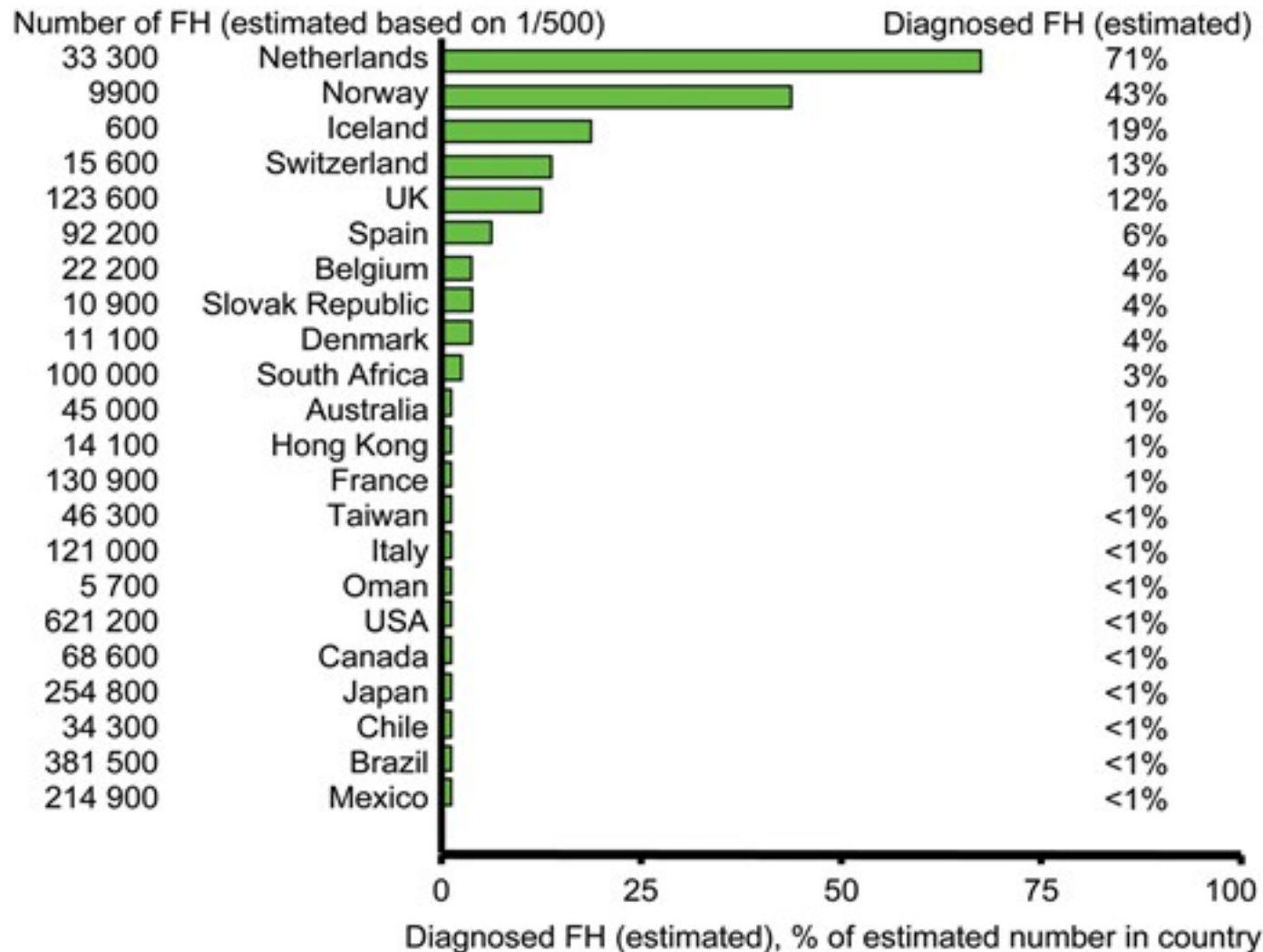
Development of ages at CHD and death from 1978 through 2021 in the entire population of Denmark

Ages at coronary heart disease and death in familial hypercholesterolaemia: a Danish nationwide study spanning 44 years. European Heart Journal, 2024

However,



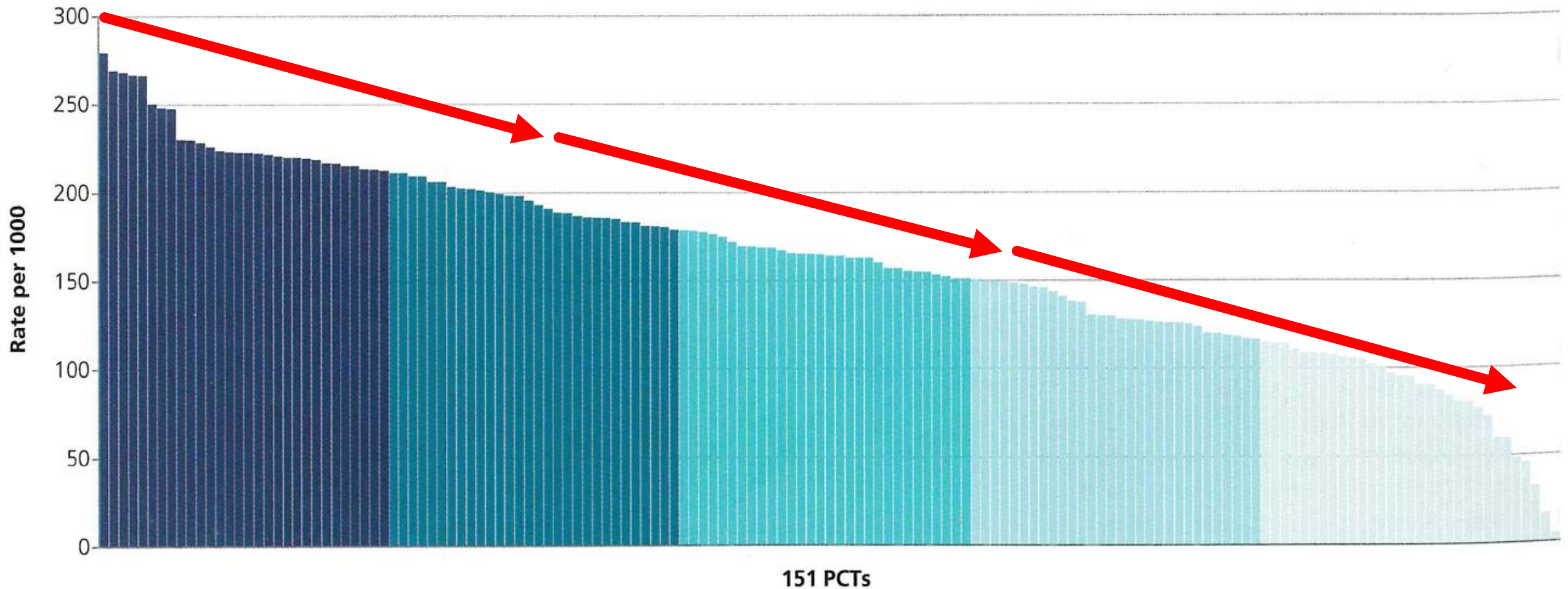
FH is One of the Most Common Genetic Disorders, Yet Remains Underdiagnosed



- Graph prepared using a HeFH prevalence 1:500
- However, prevalence of definite or probable HeFH is shown to be at least 1:250
- Recent estimated prevalence of HoFH to be 1 in 300,000

Geographical Bias of Care in England

LDL Cholesterol tests requested per 1000 people in England



Why this situation with FH?

- ▶ Poor FH-risk identification tools.
- ▶ Geographical bias of care.
- ▶ MANY patients would need to be screened.
- ▶ Very expensive and limited resources for outpatients' appointments, OP administration, booking, etc.
- ▶ Patients' insufficient awareness about FH.



Let's think outside the box

Together, for
Best Patient Care

FH
Project
N&W

Eclipse

Protect
NoW

East
Genomics

East
GMSA

Health
Innovation
East

PRIMIS

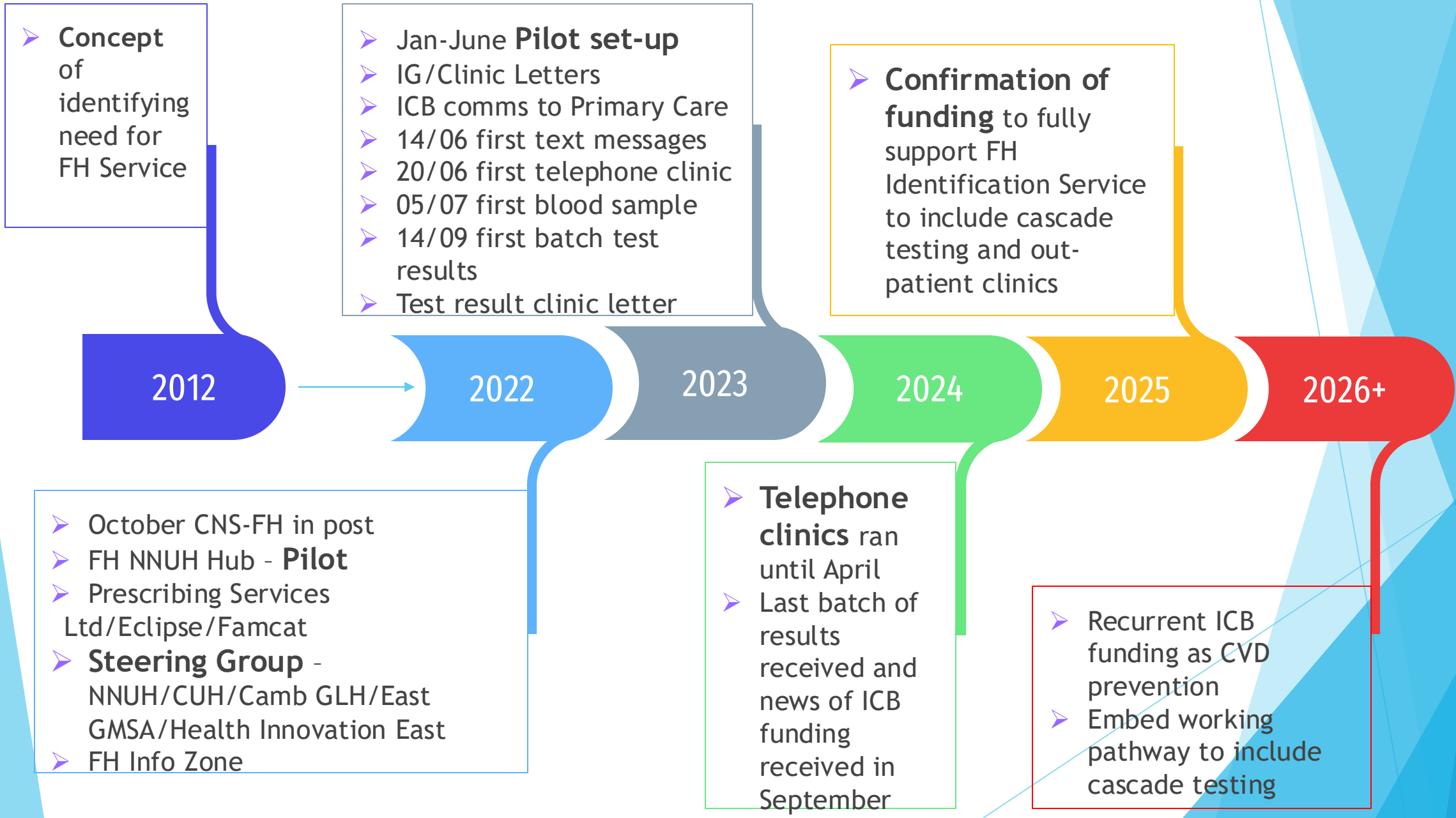
N&W
ICB

FH Hub
EPA
NNUH

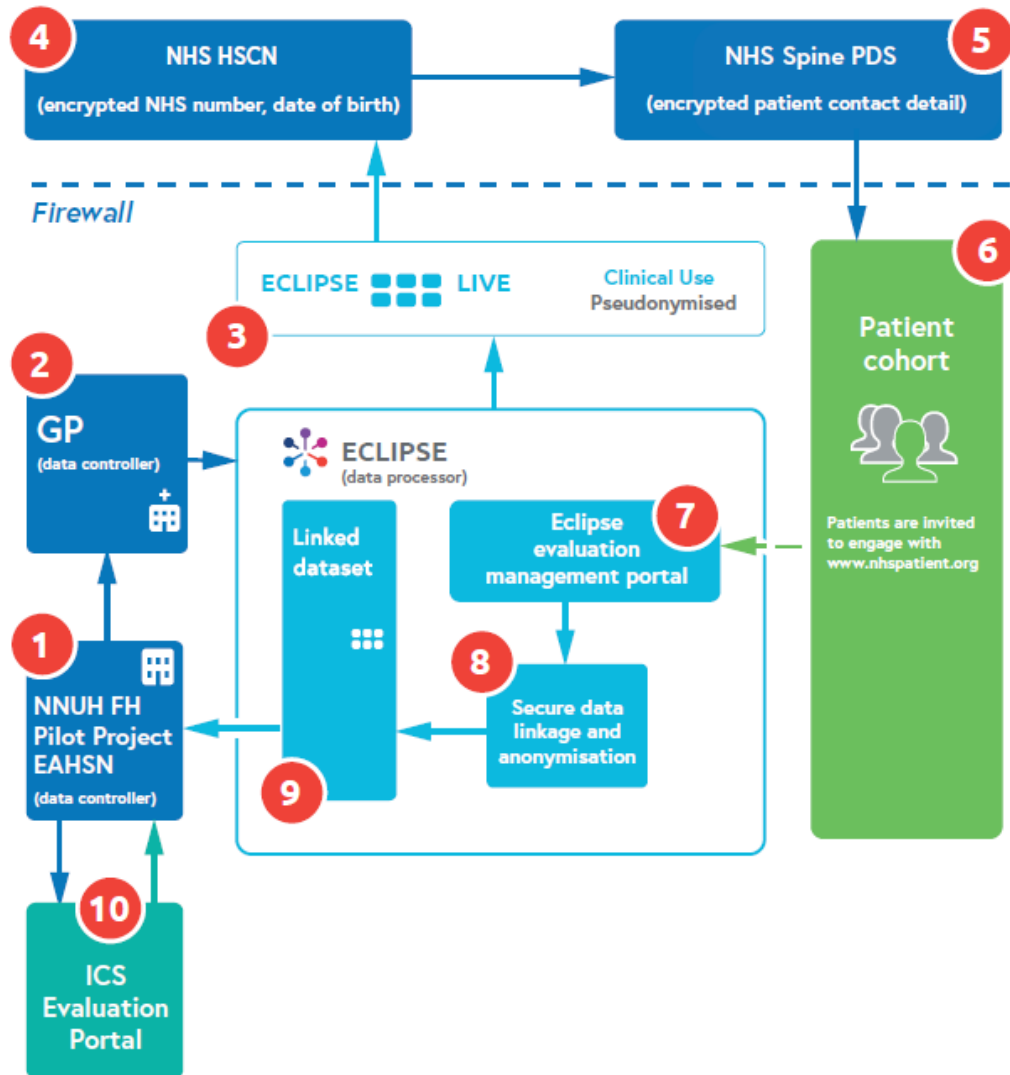
UEA



Pilot FH Identification Service Milestones Overview



FAMCAT-ECLIPSE FH HUB



1. Identifies patients at risk of FH
2. Patient receives invitation and information
3. Appointment made Tel / inperson
4. Assessment with FH Specialist Nurse, advice, guidance and genomic testing

- 1 NNUH FH Pilot Project/EAHSN - Aim is to identify patients with a total cholesterol greater than 7.5mmol/L or LDL-C greater than 4.9mmol/L in an adult. Complete FAMCAT, FTQ and clinician review and offer identified patients at high risk of FH Genomic testing.
- 2 Fully informed GPs (data controllers) instruct Prescribing Services Ltd (PSL; data processors) to securely process data for the evaluation including screening patient lists.
- 3 PSL interrogates the ECLIPSE LIVE clinical database against prescribed eligibility criteria to create a **pseudonymised** database of patients that may benefit from the intervention.
- 4 PSL-managed, automated system contacts the secure NHS HSCN server to obtain NHS number and date of birth. No details are retained.
- 5 PSL automated system accesses the secure NHS SPINE PDS (patient demographic services) for patients' contact details. No details are retained.
- 6 PSL sends secure one-way data transmission via the NHS SPINE PDS inviting patients to engage with their 'NHSPatient.org' patient portal.
- 7 Patient engagement, NHS data collation and data linkage is securely managed within the Eclipse evaluation management portal.
- 8 Intervention provider securely receives minimum required patient contact data from ECLIPSE and returns anonymised aggregate data and pseudonymised intervention data to ECLIPSE for distribution (aggregate data) or integration (individual data).
- 9 PSL conducts secure data linkage and anonymisation processes.
- 10 ICS Evaluation Portal provides linked, fully **anonymised** evaluation dataset and aggregate dataset to CPICB.

Wider Norfolk and Eastern Region Engagement

- Eastern Pathology Alliance (EPA) Newsletters
- Pulse Magazine/The Beat - NNUH intranet
- FH : Transforming Diagnostic Pathways May 2023 (Kate Downes) - Automated case-finding for FH in Primary Care
- Rare Disease Day Feb 2024 - promoting genomics
- Rare & Inherited Showcase July 2024 - Finding the missing 92%
- Pulished February 2025 [Pilot to permanent: the FH Hub's success story - Health Innovation East](#)

New service identifies people at highest risk from cholesterol condition

A new pilot at our hospital is identifying local people at risk from a condition known as Familial Hypercholesterolemia (FH).

FH is one of the most common treatable genetic disorders that increases the likelihood of premature heart disease and affects approximately 220,000 people nationally. An affected parent has a 1 in 2 chance of passing the condition on to each child they have.

Untreated, FH can lead to an increased risk of angina, heart attacks and strokes earlier in life because of high LDL, or "bad cholesterol". In FH the genes responsible for regulating cholesterol are not working effectively which, over time, causes a build-up of LDL in the arteries.

Despite the risks, nationally only 8% of people with FH have been identified. Across Norfolk and Waveney, around 4,300 people have FH, with just 300 diagnosed.

Shelina Rajan, Clinical Nurse Specialist and Consultant Dr Javier Gomez are running the pilot service from NNUH.

Shelina said: "We are supporting GPs and other healthcare professionals to identify more people at high risk of FH. This means we can do a simple genetic blood test and treat people earlier to greatly reduce their risk of developing heart disease."

The earlier someone with FH is identified, the sooner they can be started on lipid lowering treatment, which greatly improves their long-term health. The new service also offers people genetic counselling and support whilst waiting for their results.

To date the team have identified 43 people in the



highest risk group, and of those 30 have undergone genetic counselling and genetic testing. From this group, 23 have received their result which is a good outcome for them and the service.

Dr Gomez added: "People we identify as being at higher risk are invited to view a patient information portal, which explains more about our service and FH. We are now rolling out the service to all 105 GP Practices across Norfolk and Waveney."

You can find out more on the East Genomics website at www.eastgenomics.nhs.uk or scan this QR code



If you receive a text message from NHSConnect, please follow the link and help us to help you, and your family.

Information for patients can be found on the Heart UK website at www.heartuk.org.uk/fh/what-is-fh and British Heart Foundation website www.bhf.org.uk/informationsupport/conditions/familial-hypercholesterolaemia

Rare & Inherited Showcase agenda

Wednesday 10 July 2024

9.20am - 1.30pm

MS Teams

NHS
East Genomics

Time	Session	Speaker
9.20am	Opening & Welcome	Prof Dick Sandford, Clinical Director, East GMSA 
9.30am	How is genomics impacting rare and inherited conditions?	Prof Guru Aithal, Medical Lead, East GMSA & Consultant Hepatologist, Nottingham University Hospitals 
9.50am	Finding the 'missing 95%': Familial Hypercholesterolaemia in Norfolk and Waveney	Dr Javier Gomez, Consultant in Clinical Biochemistry, Norfolk and Norwich University Hospitals 

Patient Communication & Engagement

- Letter
- Text message
- 3 telephone calls
- Cholesterol Support Programme (FH) - Norfolk & Waveney
- Portal page



“Your surgery has prioritised you as someone who may benefit from a high cholesterol support programme...

...We are working with healthcare professionals across Norfolk & Waveney to help educate and, where appropriate, treat people with high blood cholesterol.

High cholesterol can be caused by a condition called familial hypercholesterolaemia. We aim to find and treat this as early as possible to prevent complications and improve your health.

”

Cholesterol Optimisation Patient Portal

The Patient Portal has a built-in accessibility tool that can:

- Read website text aloud (including PDFs)
- Download the text as an MP3 file to play
- Translate text into more than 100 different languages
- Change font sizes and colours and customise background-colours
- Access a fully integrated dictionary and thesaurus

Primary Care Communication & Engagement

- West Norfolk PCN
- Wider N&W PCNs
- Newsletter to GPs containing link to Pro-forma
- Poster for GP Practice waiting areas

Have you received a text from NHSConnect about FH, a high cholesterol condition?

We are running a new service to identify local families at risk from a high cholesterol condition known as Familial Hypercholesterolemia (FH).

FH is one of the most common treatable genetic disorders that leads to high cholesterol and increases the likelihood of early heart disease, angina, heart attacks and strokes.

DID YOU KNOW?

- 220,000 people with FH in England
- Over 4,300 in Norfolk & Waveney
- 92% are unaware they have it
- 50% chance children will inherit it

What happens if I'm contacted?

We will contact you on behalf of your GP practice via a text message that will come from NHSConnect.

The text will contain a link to a patient portal where you can find out more about FH, and enter some details to help us help you find out if you or your relatives are affected by this serious, but easily treatable condition.

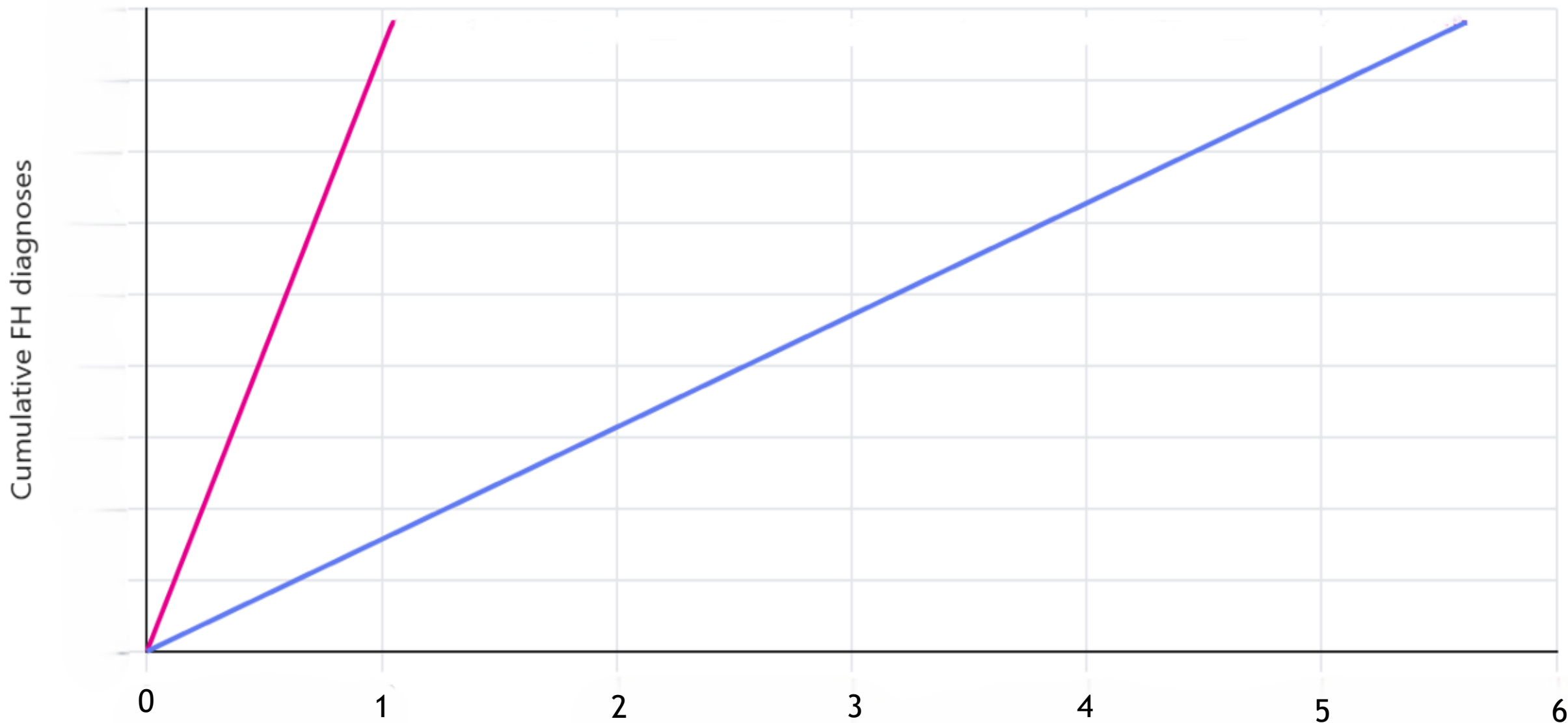
PATIENT PORTAL

To find out more scan the QR code or visit: bit.ly/FH_Info

Results

The background of the slide features abstract, overlapping geometric shapes in various shades of blue, ranging from light sky blue to deep navy blue. These shapes are primarily located on the right side of the slide, creating a modern, dynamic visual effect.

- NNUH adopted process in Norfolk and Waveney ICB
 - Standard process for FH diagnosis based on Simon Broome criteria
- 19 tests per FH diagnosis
- 102 tests per FH diagnosis



Patients experience is much better:

- ▶ Patients who don't have FH are not troubled and worried.
- ▶ FH Patients' satisfaction of their positive outcome.
- ▶ We reach >3 times patients with FH.

- ▶ 19% detection rate,
compared with only 6% based on standard SB protocol.

- ▶ Avoids geographical bias.
- ▶ Adapted to all needs: language, IT literacy, ethnicity, social background, health limitations etc.
- ▶ Equity to give all patients at risk equivalent opportunities to benefit.
- ▶ Reasonable timescales and appointments.
- ▶ Affordable to be able to reach All !

FH Pilot - Results

19% detection rate,
compared with only 6%
based on standard SB
protocol.

Norfolk & Waveney Pilot FH Identification Project 20/06/2023 to 12/09/2024

16 Primary Care Networks (46 out of 105 Primary Care Practices)
692 patients contacted (554 ♀ 138 ♂ aged 21-91 yrs) **VH Risk** 209 **H Risk** 390 **M Risk** 93
175 patients responded
150 patients screened (134 ♀ 16 ♂)
141 blood samples sent to Cambridge GLH (126 ♀ 15 ♂)
141 results received

Results		FAMCAT Risk		15 ♂ 126 ♀	Statin use pre FH genetic test	Statin use post FH genetic test	Q-Risk score of patient with FH not prescribed lipid treatment
				Sex			
LDL - R	21 pts	Very High	10 pts	0 ♂ 10 ♀	7/10	9/10	1 patient = 33%
		High	11 pts	3 ♂ 8 ♀	10/11	11/11	
Apo-B	3 pts	Very High	1 pt	0 ♂ 1 ♀	1/1	1/1	
		High	2 pts	0 ♂ 2 ♀	2/2	2/2	
LDL-R VUS	2 pts	High	1 pt	0 ♂ 1 ♀	1/1	1/1	
		Medium	1 pt	0 ♂ 1 ♀	1/1	1/1	
PCSK9 VUS	1 pt	Very High	1 pt	1 ♂ 0 ♀	0/1	0/1	1 patient = 19%
Polygenic	114 pts	Very High	28 pts	2 ♂ 26 ♀	23/28	Determined by GP based on Q-Risk score	
		High	76 pts	9 ♂ 67 ♀	49/76		
		Medium	9 pts	0 ♂ 9 ♀	8/9		

TABLE 1													Dutch Lipid	
	Sex		Deprivation Score		Current Statin		Diabetes		CKD		FAMCAT2 Risk		Criteria (DLC)	
Non genetically-confirmed FH	Range		Use										Score Range	
	Female	Male	Female	Male	Female	Male	Female	Male	Female	Male	Female	Male		
(n = 114)	103	11	1 - 10	2 - 9	76%	73%	9%	18%	5%	0%	26 ^A	2 ^A		
	Age 42-79	Age 31-71	med=5	med=6							68 ^B 9 ^C	9 ^B		
	med=62	med=56												
Age 27-35		1		4		100%		100%		0%		1 ^B		
36-44	2		1-3		50%		0%		0%		2 ^B			
45-53	8	4	1-9	2-9	75%	25%	0%	0%	0%	0%	3 ^A 5 ^B	4 ^B		
54-62	43	3	1-10	2-9	60%	66%	9%	33%	2%	0%	12 ^A 29 ^B 2 ^C	1 ^A 2 ^B		
63-71	37	3	1-9	5-7	81%	66%	8%	0%	5%	0%	9 ^A 23 ^B 5 ^C	1 ^A 2 ^B		
72-80	13		1-9		92%		15%		15%		2 ^A 9 ^B 2 ^C			
Monogenic (n=23)	Female	Male	Female	Male	Female	Male	Female	Male	Female	Male	Female	Male	Female	Male
VUS (n=3)	23	4	2 - 9	5 - 6	96%	50%	4%	50%	4%	0%	11 ^A	1 ^A	1 - 18	6 - 14
	Age 43-80	Age 55-74	med=5	med=5.5							11 ^B 1 ^C	3 ^B	med=3	m=7.5
	med=70	med=58												
Age 36-44	2		5-6		0%		0%		0%		1 ^A 1 ^B		5-6	
45-53	3		2-5		66%		0%		0%		2 ^A 1 ^B		3-4	
54-62	2	3	3-5	5-6	100%	66%	100%	66%	0%	0%	1 ^B 1 ^C	1 ^A 2 ^B	2-3	6-14
63-71	8		3-9		87%		0%		87%		4 ^A 4 ^B		1-6	
72-80	8	1	2-6	6	100%	100%	0%	0%	0%	0%	4 ^A 4 ^B	1 ^B	1-18	6
Entire cohort median value	Age 62		5											

* All values are mmol/L

¹ 17 patients in total with DLC score <6

² 1 Genetically confirmed FH in 1 sibling plus 1 sibling pair

^AVery High ^BHigh ^CMedium Risk of FH

Table 1 displays the FH pilot cohort breakdown incorporating elements of predictor variables of the FAMCAT2 algorithm and associated stratified risk scores. The DLC was added as a comparator tool and identified 17 patients with a score of <6 who may not have been referred for FH genetic testing in a clinical setting.

TABLE 3					1st Degree Fam Hx		1st Degree Fam					
	Sex		High Cholesterol		Hx IHD no MI		FAMCAT2 Risk		DLC Range			
Results	Female	Male	Female	Male	Female	Male	Female	Male	Female	Male		
Apo-B (n=3)	3		66%		33%		1 ^A 2 ^B		1-4 ¹			
LDL-R (n=21)	18	3	83% ²	66% ²	28%	33%	10 ^A 8 ^B	3 ^A	1-18 ¹	6-9		
LDL-R VUS (n=2)	2		50% ²		0%		1 ^B 1 ^C		2-8 ¹			
PCSK9 VUS (n=1)		1		0%		0%		1 ^A		14		

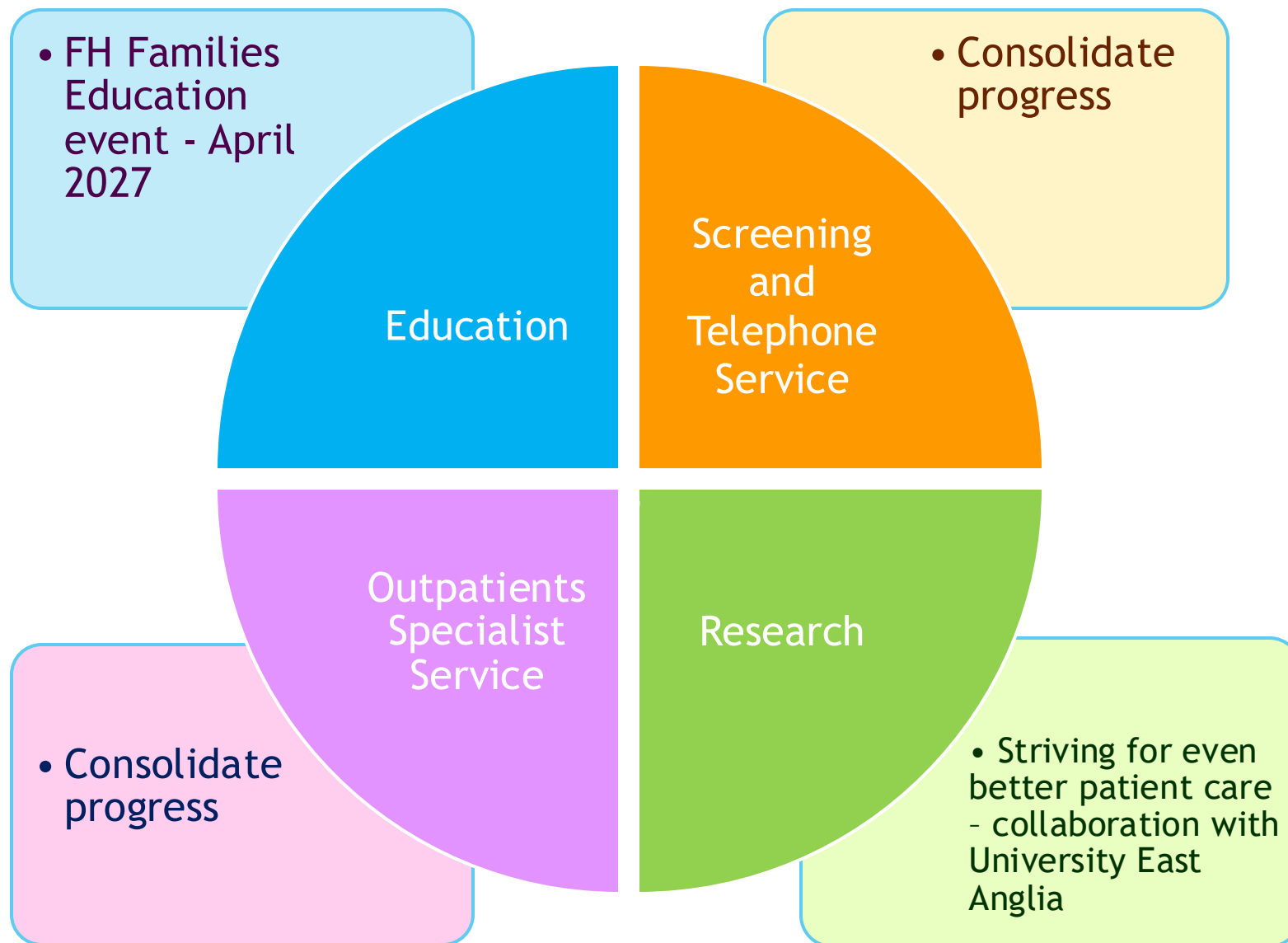
Table 3 shows the breakdown of monogenic FH and VUS with family history and the associated FAMCAT2 stratified risk scores and DLC as per Table 1.

TABLE 2								
	First Available		First Available		First Available			
Non genetically-confirmed FH	TC* Range		LDL-C* Range		Triglyc.* Range			
	Female	Male	Female	Male	Female	Male		
(n = 114)	3.9-20.2	4.9-10.4	1.6 - 8.2	2 - 3.5	0.5-5.7	0.6-7.9		
	med=7.1	med=7	med=4.7	med=3.2	med=1.8	med=1.8		
Age 27-35		6.5		3.5		1.5		
36-44	8.1-8.9		4.2-5.1		1.1-2.7			
45-53	5.1-10.7	5.6-10.4	2.5-8.2	2.9	0.8-3.3	1.8-2.3		
54-62	4.3-20.2	5.9-9.0	2.1-6.3	3.1-4.5	0.4-5.6	1.2-7.9		
63-71	3.9-11.0	4.9-9.3	1.6-7.2	2.0-3.4	0.4-5.7	0.6-1.48		
72-80	5.1-9		2.7-6.7		1.1-4.9			
Monogenic FH (n=24)	Female	Male	Female	Male	Female	Male		
VUS (n=3)	5.1-13.2	7.5-10.4	2.5 - 11.7	4.3 - 21.0	0.5-3.4	0.7-2.0		
	med=8	med=9	med = 6.2	med=7	med=1.0	med=1.0		
Age 36-44	8.6-9.4		6.7-7.8		0.5-1.7			
45-53	7.7-8.5		5.9-6.4		0.8-1.2			
54-62	6.9-8.3	7.5-10.4	2.5-4.2	7.2-21.1	1.7-3.4	1.3-2.0		
63-71	5.1-10.3		3.3-8.3		0.8-1.8			
72-80	6.8-13.2	9.2	4.2-11.7	4.3	0.9-1.7	0.77		
Entire cohort median value	7.3		4.6		1.6			

Table 2 shows the first available TC and LDL values. Were unable to establish with accuracy if patients were taking any lipid-lowering medication historically.

*All values are mmol/L.

Our plans 2026-27



Some testimonies received from patients:

- Having lost four family members to heart attacks, I was keen to take part in the FH assessment. I would encourage anyone who receives an invitation to do so. From patient
- Shelina explained the whole process fully and made sure I understood the reasons behind it. She was a joy to chat with and very informative. From patient
- I would encourage anyone who has been invited to take part in this FH scheme if they can. From Christine, patient



A straight line is made of many points,
one united to another: engage others for
Better Patient Care!



Katie Blakeney

Regional Programme Manager

Joe Power
ICB Senior
Manager



Ian Kingsbury

Communication and
Public Involvement Lead



Javier Gomez
FH Service Lead



Shelina Rajan
FH Specialist Nurse



Cat Hamlet

Protect NoW Manager

Jackie Cole
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Officer



Silvia Pruteanu
VTS Call Agent



Rebecca Ritchie
FH Officee



Jon Saward
VTS Call Agent