Genetic Testing for Familial Hypercholesterolemia TODAY

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PROBLEM TRYING TO SOLVE

- Familial Hypercholesterolemia (FH) impairs the ability to clear LDL cholesterol
- FH is underdiagnosed, especially in young adults
- FH affects approximately 1 in 200 people
- Patients with FH are at 20x increased risk for coronary heart disease

OUTCOME/GOALS/IMPACT TO PATIENT

- Assess the effectiveness of a phenotype-based EHR screening approach to identify individuals with FH.
- Characterize patient interest and engagement in the genetic testing and counseling process for FH in a primary care patient population at OSF
- Evaluate downstream services completed following patient identification.
- Pilot is in planning stages

JOURNEY TO GET THERE/PLANNED JOURNEY

- Use algorithmic approach to identify patients via existing EMR data
- Conduct targeted, engaging outreach
- Discuss significance and encourage patients to seek testing
- Initiate or alter treatment based on results

DIRECT IMPACT TO PATIENT/FAMILIES

- Early diagnosis can decrease risk of coronary heart disease of those with FH to close to that of the general population
- Promotes identification and testing of first-degree relatives

OSF INNOVATION

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