

Engaging Individuals with Familial Hypercholesterolemia and their Families to Design a Direct Contact Program (DCP) to Improve Cascade Testing Uptake

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Introduction	Participant Recommendation Themes	Description	How Recommendations were Applied to Create a DCP
<ul style="list-style-type: none"> Familial hypercholesterolemia (FH) is one of the most common genetic disorders, causing increased risk of premature atherosclerotic cardiovascular disease Early diagnosis and treatment can significantly improve prognosis and health outcomes Strategies that facilitate family communication about FH can reduce proband burden and improve cascade testing uptake Active, provider-mediated methods like “direct contact” can result in a higher rate of new relatives identified with FH per proband compared to proband-led methods AIM: Identify key input from individuals and families with FH to design a patient-centered direct contact program (DCP) to improve cascade testing uptake 	1. An FH expert with a connection to the family should perform direct contact	<ul style="list-style-type: none"> Discussed how an expert in FH would have authority Described wanting the proband’s clinician, especially a PCP, to perform direct contact Suggested clinician explain their connection to the family Highlighted the importance of the tone and communication skills of the clinician 	<ul style="list-style-type: none"> Genetic counselors, who are experts in FH, perform direct contact It is not feasible to ask PCPs to add direct contact to their workload Genetic counselors explain how they are connected to the proband and their expertise in genetics and FH Genetic counselors are well-trained in communication skills and bedside manner
	2. Prime relatives in writing that direct contact will occur and allow an opt-out option	<ul style="list-style-type: none"> Described wanting a primer letter to be sent to relatives to alert them that they will receive a phone call from a clinician 	<ul style="list-style-type: none"> Developed a primer letter to be sent to relatives via secure email, the patient portal, or by mail at the direction of the proband
	3. Coordinate internally if probands and relatives are in the same healthcare system	<ul style="list-style-type: none"> Recommended automatically contacting a relative’s clinician if they and the proband are in the same healthcare system without explicit permission 	<ul style="list-style-type: none"> Provided the option for the genetic counselor to directly contact the relative’s clinician if the proband and/or relative provides contact information Made option available even if proband and relative were not in the same healthcare system
	4. Provide probands the opportunity to be involved in outreach	<ul style="list-style-type: none"> Expressed desire to review what the clinician will say during direct contact Described wanting to help the clinician tailor conversations to relatives 	<ul style="list-style-type: none"> Provided a flyer to describe the DCP to probands to review in advance Genetic counselors do tailor and personalize care based on information about the relative provided by the proband
	5. Encourage probands to give a pre-emptive “heads up” to relatives	<ul style="list-style-type: none"> Explained how the proband should give a “heads up” to relatives Recommended allowing the proband enough time to gather relatives’ contact information and get permission from family members 	<ul style="list-style-type: none"> Coached probands at multiple touchpoints to give a “heads up” Sent primer letter to relative before direct contact Genetic counselors provide time in between when the prime letter is sent and the first attempt at direct contact in case the relative wishes to opt out
	6. Provide relatives with additional informative resources on FH	<ul style="list-style-type: none"> Recommended including multimedia sources of information Described wanting links to reputable sources of information on FH 	<ul style="list-style-type: none"> Provided links to the Family Heart Foundation’s website and the healthcare system’s FH webpage Offered a chatbot as an additional resource that the genetic counselor can send directly to the relative
	7. Help relatives understand and navigate next action steps	<ul style="list-style-type: none"> Suggested providing information about testing options for at-risk relatives outside of the state and/or healthcare system Recommended presenting both lipid testing and genetic testing options for next steps 	<ul style="list-style-type: none"> Described the importance and benefits of lipid and genetic testing Offered to place a genetic testing order or to send a link to a chatbot to order genetic testing at-home by mail Provided links to find a genetic counselor and the Family Heart Foundation’s “Find a Specialist” webpage
Methods	<ul style="list-style-type: none"> Parallel mixed method design (interviews and surveys) was used to gather input from individuals and families with FH to design a DCP Participants were recruited from Geisinger’s Multidisciplinary Lipid Clinic, MyCode precision health project, and the Family Heart Foundation Data were analyzed iteratively 		
Results	<ul style="list-style-type: none"> 11 dyads (22 individuals) completed phone interviews 98 participants responded to surveys Thematic analysis resulted in 7 themes (see Table) 		

Conclusion

Created the FH Outreach & Support Program to facilitate direct contact

Ongoing research is investigating how the program was used by families with FH and its impact on cascade testing uptake via a prospective, pragmatic trial

Findings from this FH DCP may be translatable to improve cascade testing for other genetic conditions

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Laney K. Jones is a consultant for Novartis.
Amy C. Sturm is an employee & shareholder of 23andMe.

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