Educating Patients about Familial Hypercholesterolemia: The Role of the Cardiovascular Nurse

- Genetic disease often goes undiagnosed until symptoms become severe. –

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Oct. 8, 2012 - Earlier this year, a group of people affected by FH led an effort to establish the FH Foundation as the first patient organization in the United States to focus on the needs of people living with familial hypercholesterolemia (FH). The mission of the FH Foundation is to help more people learn about FH through education, advocacy, and research – ultimately helping them to get a proper and early diagnosis and treatment. And a central consideration in the effort to raise awareness of FH is the important role that cardiovascular nurses can play in that effort.

Familial hypercholesterolemia (FH) is a genetic disorder that often results in premature heart disease, and for many with this genetic disorder death may be the first symptom and awareness that they have FH. While it is one of the most common genetic diseases, it remains widely undiagnosed. According to the National Lipid Association only about 20 percent of those affected are diagnosed, and of that group many are not aggressively treated. The link between high Low Density Lipoprotein-c (LDL-C) levels and increased risk of cardiovascular disease is well established, but according to some clinicians including cardiovascular nurses, there are significant challenges in efforts to educate both patients and colleagues about the health risks associated with the disorder.

In just one example, a focus group consisting of members from the Preventive Cardiovascular Nurses Association (PCNA) met last year during the association’s annual symposium in Florida to learn more about and to discuss efforts to expand community awareness of FH. According to their experience, there continue to be major challenges in educating patients about FH and implementing strategies to improve patient education and quality of care related to the diagnosis and treatment of associated high LDL cholesterol.

According to Dr. Lola Coke, PhD, ACNS-BC, RN-BC, FAHA, FPCNA, President of the PCNA board of directors, “The discussion revealed major hurdles nurses must clear in order to properly educate patients about the risks associated with inheriting FH. Problems arise because those who have inherited the disorder often don’t show symptoms until experiencing a major cardiovascular event like stroke or heart attack at a young age. We need to encourage people to investigate their family history and seek screening, especially if they find a pattern of high cholesterol.”

About high cholesterol and FH

High cholesterol is among the most common global health issues, affecting 71 million people in the U.S. alone. FH is a genetic condition that often causes dangerously high levels of LDL-C to build up in the blood, due to inherited genes that impair the liver’s ability to clear cholesterol. It affects about one in 500 people, making it four times more common than sickle cell disease, and five times more common than cystic fibrosis. FH is an autosomal dominant disorder, meaning that all first degree relatives of
people with FH have a 50 percent chance of also having the disorder. More than 1.6 million people in the U.S. and Europe are living with FH today. But despite its prevalence, FH remains relatively unknown both within the population as a whole and among healthcare professionals.

Many people with FH do not exhibit visible symptoms, and usually do not conform to the typical image of someone at high risk for cardiovascular disease. Additionally, children and younger adults are not typically screened for high cholesterol in routine health maintenance. Without treatment, the risk of developing premature cardiovascular disease is 20 times higher in people with FH than that of the general population. vi People with FH in their 40s often have narrowing of the arteries similar to that found in a 70-year old. vi, vi While treatment can help many people with FH to balance cholesterol levels, others may develop a more severe form of the disease in which cholesterol levels remain dangerously high, even following treatment.

Challenges in counseling patients about FH

According to cardiovascular nurses who participated in the roundtable discussion, the challenges in providing proper care often begin with the term “familial hypercholesterolemia” itself. Many nurses reported that they seldom use this term in discussions with patients because it can be confusing and intimidating. Instead, they often refer to FH using more conversational phrasing, such as “high cholesterol in the family,” “genetic high cholesterol,” or “something they inherited.” While this phrasing may be more comforting to patients, nurses also note that it could imply that the disease is not serious. As a result, patients may not understand the severity of the risk and consequently avoid seeking specialized treatment.

Some nurses also noted that colleagues often are not aware that FH can cause dangerously high cholesterol levels in children as young as age two, and consequently do not raise the issue of FH with parents. To address the problem, many healthcare providers call for broader access to information about FH overall and in children specifically. Nurses indicate that this information should be available to colleagues in primary care as well as multiple specialty areas including pediatrics, gynecology, obstetrics and cardiovascular health. In the discussion, some called for resources that highlight the prevalence of FH, the benefits of screening for high cholesterol beginning in childhood, and the importance of screening family members of those who have been diagnosed. One positive note is that some nurses reported that support for screening in children may be increasing. This could be attributed to the American Academy of Pediatrics (AAP), which now encourages cholesterol testing after the age of two for all children from families where a pattern of premature heart disease or FH are identified. Additionally, AAP recently endorsed guidelines calling for universal cholesterol screening in children beginning at age 9. vi

The image problem of FH

The focus on FH in the healthcare community may be further limited by the fact that patients often show no visible symptoms of disease and appear healthy. For this reason, some nurses report that FH lacks the “fear factor” associated with some less common diseases that are equally dangerous, but where patients present with visible symptoms that inspire them to seek appropriate medical care.
According to one nurse who treats FH patients, “It’s an ongoing challenge to alert patients with FH that their health may be at risk and that they may require lifelong health management strategies.” Some nurses also find that diagnosed FH patients who have experienced cardiac events wrongly conclude they are “cured” following treatment with stents or even bypass surgeries.

Historically, treatment for FH has largely been based on NIH guidelines, which recommend aggressive intervention to treat high cholesterol only when there is evidence of heart disease and a risk of atherosclerosis, heart attack, or stroke. But nurses familiar with FH also call for early intervention including proper screening, diagnosis, and treatment to help patients reduce the risk of cardiovascular disease and manage cholesterol levels over the long term. Many nurses saw an advantage in creating a stronger sense of urgency regarding the dangers of high cholesterol to encourage more patients and healthcare providers to appreciate the risks and take action. But they concede that efforts to build awareness of FH that can result in more preemptive action by patients and healthcare providers will continue to be a challenge.

**Diagnosis dilemmas**

Among nurses who treat FH patients, there is a concern that lack of awareness of FH among healthcare providers increases the risk of non-diagnosis or misdiagnosis. They report that many families are tested only after a relatively young family member experiences an ischemic event. Leading up to such an episode, some people with FH are diagnosed with “high cholesterol” and treated with lifestyle modifications including diet and exercise which are not sufficient to reduce extremely high cholesterol levels caused by FH related to the nature of the genetic defect. In addition to ongoing lifestyle management of diet, exercise and non-smoking, statin therapy is the pharmaceutical intervention of choice, which up-regulates LDL receptors and influences the cholesterol that the body is making by interrupting an essential step in its production. As noted by the National Lipid Association in their recommendations for the identification and treatment of FH, those with this genetic disorder that are diagnosed early and treated aggressively have the same lifetime risk of heart disease than another person without FH.¹

The possibility of non-diagnosis is particularly hazardous in the case of FH because the disease is hereditary and essentially invisible. Some nurses express doubt that healthcare providers are taking the proper steps to reach all first-degree family members following a diagnosis of FH. They stressed that nurses can play a key role in getting more at-risk patients screened for FH by identifying and reaching out to first-degree family members following a diagnosis.

To improve strategies in screening and diagnosis, some nurses conclude that streamlining the current patient advocacy framework is paramount. Primary care providers (PCP) should be on the frontline in primary prevention of FH, but there is a lack of effective referral protocols when LDL cholesterol levels are unmanageable. FH patients who require more intensive management and monitoring should be treated by a lipidologist in collaboration with a PCP. The scarcity of experienced lipidology practices in the U.S. continues to present challenges in patient access to appropriate treatment and coordination of care.
The opportunities for nurses

Nurses at the forum indicated that there may be significant opportunities for both cardiac and primary care nurses to play major roles in expanding awareness of FH and in managing FH patients. They are often well positioned to lead family outreach and education initiatives, which are especially important relative to genetic diseases. Nurses can also play a key role directing patients and families to appropriate care resources. PCNA stands at the forefront leading the way for education in the nursing community.

In efforts to expand awareness of FH and access to care for patients, nurses said they can help push for more clarity in the division of responsibilities related to patient care involving PCPs, nurses, lipidologists, dieticians, and others. They also called for more efficient coordination of care while sharing patient information with other members of the care team. Cardiac nurses also saw an essential need to create a greater sense of urgency about FH and to determine terminology and broader messaging about the disease that will resonate with other clinicians, patients, and at-risk populations.

The insights presented in these discussions show clearly that all nurses and specifically cardiovascular nurses can take an active and leadership role in educating people about FH. But they also highlight a critical need to help more healthcare providers to learn about FH and the best strategies to counsel patients and families. Information that can help healthcare providers to learn about FH include The FH Foundation (www.fhfoundation.org) and the National Lipid association (www.lipid.org).

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i Goldberg, Anne. NLA Recommendations for the Diagnosis and Management of Familial Hypercholesterolemia in Children and Adults. August 27, 2011.


