The greatest thing in family life is to take a hint when a hint is intended, and not to take a hint when a hint isn’t.

Robert Frost

Collection of family history information has long held a hallowed place in the classic “History and Physical” learned by students in the medical profession. But do clinicians really get the full possible mileage out of this time-honored tool? In this CD Summary, we review several key clinical settings in which family history can help identify patients at high risk for diabetes, colorectal cancer, breast cancer, and hyperlipidemia, and can guide targeted screening for these conditions.

DIABETES

Type 2 diabetes and its fore-runner, pre-diabetes, provide fertile ground for incorporating family history information into an efficient screening strategy. There is a strong correlation between the number of family members with type 2 diabetes and a person’s risk of developing the condition. In general, a history of diabetes in a first-degree relative doubles an individual’s risk of developing diabetes. The collection of family history information may provide an opportunity to prevent or delay the onset of disease in apparently healthy people, especially given recent evidence that lifestyle interventions can reduce the risk of developing diabetes by 58%. In light of this, family history of type 2 diabetes can be an indication to emphasize weight control and periodic monitoring of glucose metabolism.

The American Diabetes Association (ADA) recommends screening for glycemic control in those with a body mass index of ≥25 kg/m² at 3-year intervals beginning at age 45, but testing should be considered at an earlier age and/or with greater frequency in patients with a first-degree relative with a history of diabetes or additional diabetes risk factors.

Public surveillance data from Oregon suggest that clinicians have an opportunity to make a big difference by identifying high-risk individuals. Among people who were overweight, over age 45, and who had a positive family history for diabetes, less than one-third were worried that they might develop diabetes in the next 10 years, and only one in five had discussed diabetes risk with a health care provider in the past 12 months.

HYPERLIPIDEMIA

Heart disease and stroke are leading causes of death, and, along with smoking and hypertension, hyperlipidemia is a key controllable risk factor for these conditions. Lipid screening among men aged ≥35 years and women ≥45 is strongly recommended by the U.S. Preventive Services Task Force (USPSTF). However, the Task Force also recommends screening in men or women over the age of 20 if they have either a family history of cardiovascular disease in a male relative prior to age 50, a female relative before the age of 60, or if there is a family history suggestive of familial hyperlipidemia. (A person with diabetes or multiple coronary artery disease risk factors, such as tobacco use or hypertension, should also get lipid screening at a younger age.)

COLORECTAL CANCER

A positive family history also ups the ante where colorectal cancer screening is concerned. While many persons with colorectal cancer have sporadic disease, with no apparent evidence of an inherited disorder, 25% have a family history of colorectal cancer that suggests a genetic contribution, common environmental exposures, shared behaviors such as dietary and physical activity habits among family members, or a combination of all factors. A family history of colorectal cancer, or even adenomatous polyps, in a first-degree relative prior to age 60 or in two or more first-degree relatives at any age effectively doubles a person’s lifetime risk of colorectal cancer. Based on the recommendations of the American Cancer Society, this tips the scales in favor of starting screening using colonoscopy for this person at age 40, or ten years before the youngest case in the immediate family, whichever comes first.

BREAST CANCER

In Sept. 2005, the USPSTF weighed in on a heretofore controversial issue: In what settings (if at all) should a woman be referred for genetic counseling and testing for the presence of BRCA1 or BRCA2 genes based on her family history? The USPSTF recommends referral for genetic counseling and consideration of BRCA testing based on family history in certain settings (see flip side). While this may seem like a rare condition, in fact, about 2% of adult women are in the categories for whom evaluation is recommended. One wrinkle
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to be aware of: family history is pertinent whether cancer occurred in a relative on the maternal or paternal side.

TAKE-HOME MESSAGE FOR HEALTH CARE PROVIDERS

Providers have long embraced the notion that common diseases have genetic as well as environmental components. In a busy practice though, it’s not always easy to use family history to its full potential. Here are a few ways to incorporate it efficiently in your practice:

- Based on ADA recommendations, consider screening for diabetes with a fasting plasma glucose in adults age 45 and older who have a body mass index $\geq 25$ kg/m$^2$ and at a younger age in people who are overweight and have a family history of diabetes in a first-degree relative. (For good measure, the USPSTF recommends screening for diabetes in adults with hypertension or hyperlipidemia.)

- Based on USPSTF Guidelines, consider lipid screening in men aged 20 to 35 years and women aged 20 to 45 years in the presence of any of the following:
  - Diabetes
  - A family history of cardiovacular disease before age 50 years in male relatives or age 60 years in female relatives,
  - Family history suggestive of familial hyperlipidemias,
  - Multiple coronary heart disease risk factors (e.g., tobacco use, hypertonension).

- Based on American Cancer Society recommendations, consider screening colonoscopy in adults with a family history of colorectal cancer or adenomatous polyp in a first-degree relative prior to age 60. Initial screening should be considered at age 40 or 10 years prior to diagnosis of cancer in the first-degree relative, whichever comes first.

- Based on USPSTF recommendations, consider referral for genetic counseling and consideration of BRCA gene testing for any women with:
  - 2 first-degree relatives with breast cancer if one of them was diagnosed at $\leq 50$ years,
  - 3 or more first or second-degree relatives with breast cancer,
  - A combination of breast and ovarian cancer among first- and second-degree relatives,
  - A first-degree relative with bilateral breast cancer,
  - $\geq 2$ first or second-degree relatives with ovarian cancer,
  - A first or second-degree relative with both breast and ovarian cancer,
  - History of breast cancer in a male relative, or
  - Ashkenazi Jewish heritage and either a first-degree relative or 2 second-degree relatives on the same side of the family with breast or ovarian cancer. (No one ever said this was all going to be easy...)

Still, by simply recognizing key patterns in family history, you can identify patients at increased risk for debilitating and potentially lethal conditions, and set them up with the screening services they need to identify or rule out these conditions.

For more information about the Oregon Genetics Program, please visit:

http://www.oreongenetics.org/

REFERENCES