Despite early predictions of huge public demand for direct-to-consumer genetic testing, it’s safe to say personal genetic data isn’t clogging the postal system in quantities large enough to predispose mail carriers to back pain. Still, many health experts suggest that genetics remains poised to play a larger role in day-to-day medicine. While that may sound encouraging to companies that make genetic tests, it’s likely less so for family physicians with poor genetic literacy.

“Primary care clinicians, and many other types of health care providers, are generally inadequately prepared to explain the complexities of new genomic technologies,” says Dr. Greg Feero, special advisor to the director for genomic medicine for the National Human Genome Research Institute, a branch of the United States’ National Institutes of Health. “Generally, they are interested in this and feel there is potential for their patients. They probably should know more but, at this time, they are generally not convinced there is enough clinical value.”

That may prove to be a problem if personalized DNA tests become bigger sellers in the future, because primary care physicians will likely be who patients go to for guidance on genetic matters. “Should it become more popular or more used among the general population, then, yes, it’s something they will need to know about,” says Karen Powell, a genetic counselor and project coordinator at the University of North Carolina at Greensboro.

Patients will need guidance due to the uncertainty in value and interpretation of genetic tests. The science underlying many of these tests is ambiguous and there is a “virtual abyss” of information about how to use them in clinical settings, Feero noted in a commentary (Genet Med 2008;10:81-2). It is only natural that patients uncertain of the value or meaning of genetic tests results would bring questions to their family doctors. “When patients query their primary care provider regarding the genetics of common disease, they are likely to be disappointed in the answer,” he stated. “Evidence suggests that, on average, primary care providers are ill-equipped to deal with topics in genetics and genomics both from the standpoint of time and education. Some within both the primary care and genetics communities, recognizing that primary care would be faced with this challenge, have been calling for increased attention to genetic literacy among providers for some time. This call has met, and continues to meet, with ambivalence in segments of both primary care and genetic communities.”

In a genetics primer for physicians, Feero and colleagues proposed a hypothetical scenario involving a 40-year-old woman named Cathy who visits her physician for an annual physical, bringing with her a commercial genomewide scan that suggests she has a slightly elevated risk of breast cancer (N Engl J Med 2010;362:2001-11). As genomic data become more available to patients, this type of situation may become more common in doctors’ offices. “Regardless of where medicine is practiced, genomics is inexorably changing our understanding of the biology of nearly all medical conditions,” the primer
states. “How can any clinician understand the diagnosis and treatment of breast cancer, much less explain it to a patient such as Cathy, without a rudimentary understanding of genomic medicine?”

Currently, few general practitioners appear to possess that rudimentary understanding. In a recent survey of primary care physicians’ “awareness, experience, opinions and preparedness” to answer questions about genetic testing — conducted by Powell and colleagues in North Carolina — only 15% of the 382 respondents claimed they felt prepared to discuss the topic with patients (J Genet Counsel 2012;21:113-26). Only 39% professed to even be aware of direct-to-consumer genetic testing. “Education about clinical utility, privacy issues, and the pros and cons of testing is needed if DTC genetic testing becomes more widely used,” the paper concluded.

How much education on genetics family physicians need is not clear. Rather than setting a goal of making them pseudo-experts, though, it might be wiser to educate them enough to know when and how to use tools and systems to facilitate communication with genetic experts, says Feero. “As an analogy, it could be a little like MRI testing,” he says. “We train primary care doctors to know when to order a MRI. This makes sense in a clinical context. We don’t train them to take MRI film and read it.”

New technologies may also enable physicians to incorporate genetics into their practices. If electronic health records contained patients’ genetic information, for instance, it could help doctors prescribe more accurate doses of drugs, the primary goal of the field of pharmacogenetics.

“In an ideal world, your individual genomic information would already be stored in a data file,” says Feero. “When a clinician sits down to prescribe a drug, the system sees the dose and queries the person’s data and comes back and tells the health care provider the patient is very sensitive to the drug and the dose should be cut by 50%, or something like that.”

As for the means of educating primary care physicians on existing and emerging genetic technologies, no single approach is likely to suffice. Providing more genetic information in general medical journals and educational materials targeted to family physicians is important, as is an increased focus on genomics in continuing medical education programs. Teaching more about genetic testing to medical students would also help.

“There are very few medical schools that are preparing physicians for direct-to-consumer testing, but, really, it’s only been around since about 2007,” says Powell.

It is important to keep in mind, however, that certain family doctors will require more education on genetic testing than others. In areas with highly educated populations, patients tend to be more aware of genetic testing. “In a rural population with low discretionary funds, the chance of seeing a patient bring in a direct-to-consumer test is slim to none,” says Powell.

Furthermore, no amount of genetic education will change the fact that family physicians have limited time with each patient, and must use that time wisely. “The odds of finding people who have an actual syndrome are small,” says Powell. “So they focus on lifestyle, on smoking cessation and on nutrition. They focus on what will help the most patients.” — Roger Collier, CMAJ

Editor’s note: Last of a multipart series on genetic testing.
Part 1: Separating hype from reality in the era of the affordable genome
Part 2: Popping the genetics bubble
Part 3: Who should hold the keys to your DNA?
Part 4: A race-based detour to personalized medicine
Part 5: Race and genetics in the doctor’s office
Part 6: Predisposed to risk but not change
Part 7: Unhealthy behaviours influenced by genes and environment
Part 8: Young women with breast cancer genes face tough choices
Part 9: The downside of genetic screening
Part 10: Surge in Down syndrome prenatal testing anticipated
Part 11: Screening embryos made lead to stigma
Part 12: Moos your daddy?