

## Popping the genetics bubble

The hype over the promise of genetics in medicine, like the superlatives that swirl around most “revolutions” in health care, started out as genuine enthusiasm for promising technologies. It didn’t take long, however, for researchers to come under pressure to make their work sound exciting, commercializable and immediately applicable. So research institutes and fundraising bodies began touting the promise of a brighter tomorrow in easy-to-digest language.

That fed into the always-hungry media machine, bringing about headlines bloated with sunny adjectives: groundbreaking, breakthrough, game-changing. Soon the “brave new world” of genetics seeped into sci-fi movies and other areas of pop culture. And let’s not forget private companies offering direct-to-consumer genetic testing, some with marketing campaigns best described as, ahem, creative.

All aboard the hyperbole express.

Next stop: health care utopia.

Or not.

Many of the promises made by genomic evangelists have not come to fruition. Still, the field holds much promise, say geneticists, who worry there’ll be an inevitable backlash that will slow progress in integrating genomics into medical practice. And though some parties to blame for the hype may be more interested in profit than in improving health, most people swept up in the excitement had good intentions, says Dr. James Evans, editor-in-chief of *Genetics in Medicine* and Bryson Distinguished Professor of Genetics and Medicine at the University of North Carolina in Chapel Hill.

“There is a dramatic alignment of interests that all work together to inflate things without any intent to be nefarious when a bubble like this grows,” says Evans. “We need to learn from those bubbles. We need to look at the next big thing, be it nanoparticles or something else, and take a reasonable view of its promise.”

Evans and several colleagues have



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**Overhyping the benefits of genomics led to a bit of a backlash once the hype bubble burst.**

suggested that exaggerated expectations stemming from a failure to realistically evaluate the potential for genomics to improve human health will “undermine its legitimacy, threaten its sustainability, and result in misallocations of resources” (*Science* 2011;331:861-2). To get discussions on genetics in medicine back on track, they recommended fostering a realistic understanding of the “incremental nature of science,” maintaining focus on acquiring evidence before attempting to change medical practice, and re-evaluating funding priorities to ensure more work is done in areas that may yield practical benefit.

One of the main reasons that genomics did not — and likely will not

— revolutionize medicine is that genetic testing has not proven to be highly predictive of health risks. Genetics is but one component in understanding disease, and most health care problems are caused by many factors, including economic, social and environmental contributors. “All the genetics in the world isn’t going to solve our problems,” says Evans. “Our job is not to push genetics into medicine but to pull it in when it has been shown to benefit patients.”

As for the notion that genetics would usher in an era of personalized medicine, where health care is tailored to each and every individual genome — well, don’t get your hopes up, Evans says. “Let’s just say I find that overly optimistic. ...

The idea that it's going to provide a crystal ball that will personalize everyone's care is absurd. It will guide some important medical decisions and will be an important tool in our arsenal."

Of course, promises of personalized medicine don't seem so crazy in comparison with the outlandish claims occasionally made by some direct-to-consumer genetic testing companies. DNA testing has been offered to develop personalized perfumes and tailored diets, and even to assist the lovelorn in finding an exquisite, biologically compatible mate.

"While the desire for profit lies at the heart of this phenomenon, it must be recognized that it builds on the hype about the health value of genetic testing that often flows from the research community, the representations of hype that appears in the media and throughout popular culture, and the public expectations that are fueled by this hype," Timothy Caulfield, a Canada Research Chair

in Health Law and Technology who teaches in the law faculty and school of public health at the University of Alberta in Edmonton, wrote in a commentary (*JCOM* 2011;10:C02).

"In the short term, everybody benefits from the hype," says Caulfield, adding that the abundance of unrealistic expectations shouldn't detract from the actual promise of genomics. "I have no doubt that we are going to see real benefits from the tremendous genetic research that's happening."

Actually, some of that research is already set for prime time, says Cinnamon Bloss, the lead investigator of the Scripps Genomic Health Initiative at the Scripps Translational Science Institute in La Jolla, California. "There are some aspects of genomics that are ready to help improve human health, and one area is pharmacogenics," says Bloss, referring the practice of assessing how individual genotypes will react to med-

ication. "This will enable us to better dose a drug so it would better work for a person. If you know a person's genotype, you will also be better able to tell if they might have an adverse event."

Bloss adds that criticism of a genetic service as being clinically irrelevant doesn't mean it lacks value for patients, citing the concept of "personal utility." Even if, for example, a test reveals a predisposition for a condition with no treatment, some people still want that information and may live their lives differently as a result. Some consumer advocates argue that it is not up to the medical profession to decide if genetic information is useful or not.

"Is that a decision for someone else to make for me?" says Bloss. "Shouldn't I be able to decide if that information is useful? Those types of arguments are being made." — Roger Collier, *CMAJ*

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## Who should hold the keys to your DNA?

**C**an you handle the truth? Do you really want to know the secrets hidden beneath your skin, deep down in your DNA? There might be bad news in there, lurking in your genes, concealed in your proteins, skulking in your chromosomes.

These are questions more people will likely face as direct-to-consumer genetic tests fall in price and increase in sophistication. Many physicians and geneticists believe consumers shouldn't be answering those questions alone, suggesting that regulations are necessary to ensure medical professionals are part of the process. Critics of regulation disagree, claiming that doctors are being paternalistic and that consumers deserve uninhibited access to their own genetic information.

In the early days of retail genomics, more than a decade ago, there was much concern in the medical community that consumers would be unprepared to receive potentially devastating health news from genetic tests and would suffer great anxiety. That doesn't appear to have transpired, though it was not absurd, at the time, to think that it could, says Timothy Caulfield, a Canada Research Chair

in Health Law and Technology.

"It was reasonable speculation. The thinking at the time was that people were going to be getting this information and it was going to be powerful. There would be anxiety. Perhaps people would make inappropriate health care decisions. To some degree, this drove the development of the whole field of genetic counselling," says Caulfield, who teaches in the Faculty of Law and the School of Public Health at the University of Alberta in Edmonton. "It turns out, and data is emerging to support this, that people don't get that anxious about it. Maybe a cohort of people does but, in general, there aren't those high levels of anxiety that people expected."

Prior to the proliferation of direct-to-consumer genetic tests, a systematic review of scientific literature about the mental hazards of genetic discovery uncovered few studies worth reading, though it was suggested that "those undergoing predictive genetic testing do not experience adverse psychological consequences" (*Eur J Hum Genet* 2000; 8:731-8). Little has changed in the ensuing dozen years, according to a recent

study which found that consumers who purchase genetic tests suffer few anxiety symptoms. Over 90% of participants who completed follow-up reporting indicated that they suffered no test-related distress (*N Engl J Med* 2011;364:524-34).

"This type of test has been and remains extremely controversial for a variety of reasons. One is the direct-to-consumer nature of it. You get results without a physician and without a genetic counsellor," says Cinnamon Bloss, lead author of the more recent study and lead investigator for the Scripps Genomic Health Initiative at the Scripps Translational Science Institute in La Jolla, California. "Some people feel it is a medical test and that it should be interpreted by a health care professional, because people might see their test results and have anxiety if they don't understand them. Based on our data, people aren't having strong negative reactions in terms of anxiety."

In fact, many people indicate they would take predictive genetic tests "even in the absence of direct treatment consequences" and are "willing to pay reasonably large amounts for the opportunity," according to a random survey of 1463

Americans (*Health Econ* 2012;21:238-51). When presented with various disease scenarios, such as Alzheimer disease, arthritis, breast cancer or prostate cancer, 70%–88% of respondents were still inclined to take a genetic test.

“I think there is a need to think this through and to have some balance. On the one hand, we should respect consumers’ preferences and freedom to choose,” says Peter Neumann, director of the Center for the Evaluation of Value and Risk in Health at the Institute for Clinical Research and Health Policy Studies at Tufts Medical Center in Boston, Massachusetts and lead author on the paper. “We should also inform them about the risks and benefits.”

In some countries, governments have stepped in to ensure that doctors are the ones who must inform consumers about risks and benefits. Laws in France, Germany, Portugal and Switzerland stipulate that genetic tests only be administered by physicians. There are no regulations in Canada and few in the United States, though the US Food and Drug Administration has indicated that it will be stepping up efforts in the area.

“I’m of two minds about this,” says Caulfield. “If the general social anxiety isn’t that big of a problem, maybe we don’t need an aggressive regulatory response. On the other hand, I like the idea that regulators are putting their minds to this. We want to ensure, at minimum, that accurate information is getting to consumers.”

Caulfield once surmised that policymakers who do turn their attention to genetic testing should perhaps pay less attention to the “creation-of-anxiety concern” and more to the impact “on the health care system, the possible promotion of unnecessary follow-up investigations, and the need to ensure truth in advertising” (*Hum Genet* 2011; 130:23-5).

Then again, patient anxiety levels might become of greater concern when

the price of sequencing entire genomes falls to a level that makes it a reasonable consumer option, providing people with access to more, and potentially scarier, genetic information.



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**Opinion is divided over whether doctors or patients should be receiving the results of direct-to-consumer genetic tests.**

“There is reasonably good data to suggest that people who actively pursue certain genetic tests, and are highly educated about the results, and when the delivery of those results is done in a careful way, those individuals do not suffer untoward results,” says Dr. James Evans, editor-in-chief of *Genetics in Medicine* and Bryson Distinguished Professor of Genetics and Medicine at the University of North Carolina in Chapel Hill.

Still, the potential exists for people to casually discover devastating information about their health, says Evans, and that is a legitimate concern.

While it would no doubt be easier for companies that sell genetic tests to reach consumers if they didn’t have to first go through regulators or physicians, excluding the medical profession might be unwise, Evans explains. Per-

haps those who showed early interest in their genomes suffered little mental anguish because they are more scientifically savvy than the general public, but a genetic test should still be “treated like the medical test it most assuredly is” and doctors should be involved in the process to, at very least, inform people of the risks of digging into their DNA.

As for whole-genome testing, it represents an “extraordinarily complex amalgam of multiple tests that can be at once useful, pointless, confusing, or overtly harmful,” Evans once wrote while suggesting that physicians will have little choice but to grapple with how such tests should be applied and regulated (*JAMA* 2001;3306:2376-7).

“I’m not saying that people shouldn’t have access to their genome or the information in it, but we shouldn’t casually spew potentially disturbing information to people without some reasonable expectation that they have been educated about it. Physicians are often accused of being paternalistic, but we deal with complex information that can easily be misconstrued and cause harm,” he says. “We have to be careful to do this in a responsible way. I don’t think that doing it in a responsible way is overly paternalistic.” — Roger Collier, *CMAJ*

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**Editor’s note:** Second and third of a multipart series on genetic testing.

**Part 1: Separating hype from reality in the era of the affordable genome** ([www.cmaj.ca/lookup/doi/10.1503/cmaj.109-4143](http://www.cmaj.ca/lookup/doi/10.1503/cmaj.109-4143)).

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### **Who should be privy to your privates?:**

Uncertainty about who should chaperone intimate examinations may be undercutting the protections such attendants may afford patients and doctors, experts say ([www.cmaj.ca/lookup/doi/10.1503/cmaj.109-4129](http://www.cmaj.ca/lookup/doi/10.1503/cmaj.109-4129)). — Lauren Vogel, *CMAJ*

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([www.cmaj.ca/lookup/doi/10.1503/cmaj.109-4147](http://www.cmaj.ca/lookup/doi/10.1503/cmaj.109-4147)). — Lauren Vogel, *CMAJ*

*CMAJ* 2012. DOI:10.1503/cmaj.109-4150



## Chaperones: friend or foe, and to whom?

Medical regulators have long recommended the use of “chaperones” to make patients feel more comfortable during sensitive examinations.

But standards of practice vary considerably across Canada and guidelines about when to use a chaperone and whether patient consent is required are somewhat hazy and often left to physician interpretation.

What’s more, there’s growing concern among patient advocates about who chaperone policies actually serve, particularly as there’s been increasing call in recent years by medico-legal societies for more routine use of such attendants to protect physicians from allegations of impropriety.

Undergoing an intimate examination shouldn’t feel “like being searched at the airport,” says Sholom Glouberman, president of the Patients’ Association of Canada. “If all of this is based on the growing trend towards less trust and less connection between patients and practitioners, then it makes the relationship even harder and makes the situation more unpleasant and more intrusive.”

Chaperone use is not mandated by any regulatory body in Canada, except in cases where it is a condition of an individual physician license. Medical regulatory authorities, however, do offer some advice.

For example, the College of Physicians and Surgeons of Ontario leaves it for physicians to decide whether to offer patients the option of having a third party present ([www.cpso.on.ca/uploaded/Files/downloads/cpsodocuments/policies/policies/sexual\\_abuse\\_boundaries.pdf](http://www.cpso.on.ca/uploaded/Files/downloads/cpsodocuments/policies/policies/sexual_abuse_boundaries.pdf)). The College of Physicians and Surgeons of Prince Edward Island places an onus on patients to ask for chaperones ([www.cpspei.ca/publications?action=view\\_entries&id=1](http://www.cpspei.ca/publications?action=view_entries&id=1)), while the College of Physicians and Surgeons of New Brunswick says either patients or physicians can insist on a witness in situations that make them feel uncomfortable ([www.cpsnb.org/english/Guidelines/guidelines-1.html](http://www.cpsnb.org/english/Guidelines/guidelines-1.html)).

The College of Physicians and Surgeons of British Columbia provides perhaps the most comprehensive guide-



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**The use of chaperones during physical examinations is intended to put patients at ease, while protecting physicians from liability.**

lines. A chaperone’s presence may be necessary “if the patient is mentally handicapped, subject to delusions or paranoia, exhibits seductive behaviour, exhibits anxiety about a proposed examination ... is a survivor of abuse, or has other emotional or cognitive challenges which may lead to misperception or misinterpretation” ([www.cpsbc.ca/files/u6/Sensitive-Examinations.pdf](http://www.cpsbc.ca/files/u6/Sensitive-Examinations.pdf)).

But for the most part, physicians are left to rely on their own intuition.

“What we say is weigh it all out,” says Dr. Janet Wright, assistant registrar for the College of Physicians and Surgeons of Alberta. “If you’re with a brand new patient you don’t know and your ‘spidey senses’ go off ... it’s a good idea to have someone else in the room until a long-term relationship is established.”

Others contend this intuitive approach may be exposing physicians

to increased risk of allegations from patients who, for whatever reason, don’t set off those internal alarm bells.

Many physicians forego the use of a chaperone depending on the procedure or the gender of a patient, says Dr. Ross Upshur, Canada Research Chair in Primary Care and professor of family and community medicine at the University of Toronto in Ontario. Chaperones are more commonly used with female patients, while male doctors are about 40 times more likely to use a chaperone, he explains. “Female physicians are less likely to have a chaperone present while doing intimate examinations on female patients. Similarly, male physicians are less likely to have chaperones present when they’re doing intimate examinations on men.”

Therein may lie a “false sense of security,” considering both male and

female patients have leveled accusations against physicians, in some cases, regardless of the level of intimacy of the procedure performed, Upshur says.

Dr. Ed Schollenberg, registrar for the College of Physicians and Surgeons of New Brunswick, concurs, citing a recent case in which a woman complained about a female physician because she felt unnecessarily exposed during an intimate exam. “Being the same gender doesn’t offer as much protection as one might think [because] it’s not always about sex,” he says.

“We’ve tended to see the benefit of the attendant as a function of the kind of examination that’s being done,” he adds. Yet physicians have been accused of impropriety “during the course of what otherwise would not be an exam where you would think an attendant would be necessary” such as “examining a musculoskeletal injury.”

Because of that, medico-legal experts and the Canadian Medical Protective Association have urged more routine use of chaperones and detailed documentation of sensitive examinations and discussions of intimate problems (*Can Fam Physician* 2001;47:1732-1734).

But some patient advocates worry that the emphasis on protecting doctors from liability undermines the goal of putting patients at ease. “I don’t blame doctors for this but I think they’re being put into a defensive mode that’s really not appropriate,” Glouberman says. “By placing more and more security requirements ... it’s difficult to establish the kinds of relationships that are necessary [to develop] mutual trust and a chance for that to flourish.”

There’s also a dearth of detailed guidance on how to balance the use of chaperones with patient privacy, particularly in cases where a physician wants a chaperone present but a patient does not. Such cases fall into a “little gray area” of current guidelines, Upshur says.

As a result, the College of Physicians and Surgeons of Nova Scotia urges doctors to obtain “explicit consent” from patients if third parties are to be present during an examination ([www.cpsns.ns.ca/greatphysicians.aspx](http://www.cpsns.ns.ca/greatphysicians.aspx)).

Doctors in some jurisdictions skirt the issue by simply introducing chaperones as “assistants,” says Dr. Joel Sher-

man, a cardiologist and advocate for patient privacy based in Waterbury, Connecticut. “Most male doctors doing pelvic exams feel they have no choice.”

But misleading patients in such a fashion is an “affront to patient privacy,” he says.

Still, many Canadian physicians bring chaperones into the examination room without comment or explanation, Schollenberg says.

Patients may be made uncomfortable by the direct offer of a chaperone because it hints at an expectation of impropriety, he explains. “I’m not sure it requires explicit consent because I think there’s some expectation on the part of the patient that there’s going to be other people around.”

But Dr. Samantha Kelleher, deputy registrar for the College of Physicians and Surgeons of British Columbia, says physicians who opt to have a chaperone in the examination room also have a “responsibility to explain” their rationale and offer referrals to patients who fear their privacy may be compromised.

“If physicians have a straightforward approach to their use of chaperones then I think it becomes much easier,” Kelleher says. Ensuring that patients are informed in advance, through such means as signs in waiting rooms, is preferable to last-minute notice that a physician wants a chaperone present, she adds.

Dr. Victoria Davis, a member of the Society of Obstetricians and Gynaecologists of Canada’s Social and Sexual Issues Committee, concurs. But such a policy should be applied to all patients equally, as the greatest damage to the doctor–patient relationship occurs when patients feel they are being singled out as untrustworthy, she adds. “That’s a big drawback [of using chaperones on a case-by-case basis]. At that point you’re probably breaking that relationship.” — Lauren Vogel, *CMAJ*

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**Editor’s note:** First of a two-part series.

Part 2: **Who should be privy to your privates?** ([www.cmaj.ca/lookup/doi/10.1503/cmaj.109-4129](http://www.cmaj.ca/lookup/doi/10.1503/cmaj.109-4129)).