

Musings

Musings on genome medicine: abuse of genetic tests

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Abstract

The wide general publication of a putative genetic test for athletic supremacy is clearly an abuse of genetics and reveals an undercurrent of hucksterism in biomedical science.

When we agreed to compose a series of commentaries for *Genome Medicine*, we vowed to stick with the science and discuss interesting and important developments in genome medicine. We started with genome-wide association studies, and plan commentaries on gene therapy and cancer genetics to be followed by a series of commentaries on various genetic diseases, from Crohn's disease to disorders of eye development. We did not intend to launch into *ad hoc* discussions of ethical or policy issues because we do not consider our own opinions on scientific or clinical right and wrong to be more valid than those of any thoughtful professional. We do not appreciate lectures on morality so, we concluded, why should we inflict them on our readers?

But something snapped in both of us when we read a front page story in the *New York Times* of 30 November 2008. There for all to behold was a new gene test, this one for a gene that may have some vague relationship to muscle activity. The test is being marketed to the unwary who may wish to determine whether their fertilized egg, fetus, or newborn is endowed with the "right" gene to support a future in certain kinds of athletics.

To test our own immediate negative reactions we asked one of our colleagues, a distinguished molecular, population and clinical (pediatric) geneticist, to read the *New York Times* article and give us his impressions. He had seen some of the reports on the genetics as well as the article and his response was as follows:

"Now I think I need some antihypertensives to avoid a stroke. The association with athletic ability (I seem to recall) had some intriguing hints but was very confusing to me (different effects for endurance versus speed, elite athlete versus non-elite, male versus female). I also think there are other studies, not mentioned in the NYT article, that don't back it up. Plus, there are other reasons it might have been a false positive. But, even if it were completely valid, there is a huge distinction between a valid association and marketing a genetic test to parents of potential youth soccer players. Whatever happened to having your child try out a bunch of activities and seeing what your child likes and is good at?"

I think the test is worthless and probably misleading."

The gene in question is *ACTN3*, a member of the muscle α -actinin gene family. Polymorphisms of this gene are the subjects of about 50 reports, largely in the sports medicine literature. Most of the articles make rather weak claims of associations of certain polymorphisms with either muscle strength or speed in athletes. Some of the reports dispute the associations. To make decisions on the future of a child based on such a test is clearly irresponsible.

This is not to say that genetics plays no role in athletic performance. A look at the dominance of Kenyan runners in marathons is all one needs to be persuaded that genetic endowment together with training plays a very strong role in

athletic performance capacity. But to rely in any way on a single genetic test to predict such performance is absurd. The purveyors of such a test are simply preying on the anxieties and gullibility of parents who have no biomedical training and are irrationally desperate to achieve success for their children. The marketing of such a test conjures up the ultimate abuse of genetic medicine, a nightmare scenario in which technicians sort through fertilized eggs, discarding embryos that don't have the 'right' genetic stuff to be potential football stars.

We do not see ourselves as moralizing physicians encumbered by some religious or high-minded zeal, or unwilling to explore controversial procedures. We have, in fact, devoted considerable effort to the development of prenatal diagnostic tests for the serious hemoglobinopathies such as thalassemia or sickle cell disease because we believe that parents should be able to plan a family without fear. But we share with most of our neighbors a deep reverence and respect for the fetus, the mother and the child. We would never devise a prenatal test for a trivial purpose, because that might lead to abortion for no worthy reason. We include as trivial the gender of a fetus, unless the information would inform the prospective parents about the risk of an X-linked disorder the gene for which had not yet been determined. Nor would we countenance a weak (read worthless) genetic test for anything that might be stupidly used to pigeon hole the academic or athletic future of a child. We consider conclusions drawn from single genetic tests to establish probabilities of complex traits both foolish and venial: venial because the hucksters and mountebanks who promote the tests do so knowing that they are selling snake oil.

Perhaps we are particularly sensitive when we see such nonsense promulgated by so-called 'experts' because we have been forced to accept that our own profession is in fact infiltrated by a few unscrupulous and greedy colleagues who are in 'the game' to get rich and famous and not to provide the very best they can for patients who suffer from serious disorders. The infiltration can come very close to home. At this very moment we have prominent faculty members who stand accused of selling out their reputations to drug companies in order to help the companies market drugs of disputed value for children. They accept huge consulting fees from such companies and then piously maintain that they are above the influence of mere pelf.

In no way does our disgust with such outliers reduce our respect for, and indeed reliance on, the pharmaceutical or biotechnology industries. We are thoroughly persuaded by our own experience that these two industries have made huge contributions to medicine worldwide. Without their brilliant science, their medicinal chemistry and their capacity to perform clinical trials, we would be set back decades. But the outliers in the industries and on faculties bring shame and opprobrium as well as excessive regulation upon all of us. We

are in a very tough period in investigative medicine right now. Funds are in short supply, just as our knowledge is expanding. We are on the threshold of major discoveries in the chronic diseases that afflict our society. False claims ruin our stature in society, turn government against us and, worst of all, destroy the confidence of our patients.

So we are angry when we read articles like the great muscle gene prevarication. We just don't want to co-inhabit clinical science with 'colleagues' for whom we have no respect. So we took this opportunity to say so loud and clear. Genetics has a huge potential in medicine and public health, but that potential will not be realized if genetics is twisted to put money and power ahead of quality patient care.

We had to get that anger off of our chests. Now we can return to the science.

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