Misconceptions about genetic testing still too common among public

A recent Ars feature story about genetic screening generated quite a lively debate in the discussion thread. However, it also underlined just how many misconceptions people have when it comes to genetics. Public perception hasn't been helped by scientists overhyping their findings or by inaccurate portrayals in the media (*GATTACA*, anyone?). So today, I'm going to try to clear some common confusions.

First off, screening tests and diagnostic tests are not the same. Genetic (or genomic, where multiple genes are analyzed) screening tests don't always tell you if someone has a disease. Rather, they're typically probabilistic—they tell you if you've got a greater chance of a problem than the average person. Even an increased risk compared to the general population is still just that: a risk.

But humans aren't good at understanding probabilities and risks, something that's clear even with simple examples like the Monty Hall problem. In fact, there's no reason to believe that more screening would lead to more effective treatment of a given disease, which is why there are such concerns right now with the overuse of both prostate cancer screening tests and mammograms.

Hand in hand with the fact that screening isn't diagnosis goes the fact that tests are not 100 percent specific (not all positive results are true) nor 100 percent sensitive (not all negative results are true). False negative results—telling someone they're low-risk when they're not—is immediately obvious as a problem. But false positives are harmful, too, particularly if a positive result leads to an invasive biopsy or a drug therapy (and that's not considering psychosocial harms, which are real).

The GLP aggregated and excerpted this blog/article to reflect the diversity of news, opinion and analysis. Read full, original post: Misunderstanding the genome: A (polite) rant