Genetic Testing: Pass or Fail?

Testing Genetic Testing Companies for Substantiated Evidence behind Testing

Tag Words: genetic testing, DNA, Genetic Information Nondiscrimination Act, Myriad Genetic Laboratories, Atlas Sports Genetics, GeneLink, Dermagenetics, ACTN3, MMP1, SOD2, GPX1, EPHX, TNFa, BRCA1, BRCA2, MLH1, MSH2, MSH6, EPCAM, PMS2, P16, DPYD, TYMS

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Summary

Genetic tests comprise analysis of human DNA, RNA, or protein. Genetic tests are known for early detection of a mutant or variant in the genome that is known to cause or contribute to a debilitating phenotype, or disease. However, recently companies are claiming that they can detect more than just disease based on genotype (genetic makeup), including future abilities and talents in young children, the age at which wrinkles will appear, the optimal diet for an individual, and no-fail quit-smoking plans. Certain claims lack substantial scientific backing and therefore the validity of those tests are questionable. The majority of genetic testing devices and kits can be found on the internet on the company website. Some companies advertise clinical tests directly to buyers. The major flaw inherent in this process is that consumers often put blind faith in the product and obtain clinically significant test results before consulting a physician, and therefore consumers do not have proper instruction in understanding or utilizing the testing results.

Video Link

Genetic Testing: Pass or Fail?: http://www.youtube.com/watch?v=jpYPO838Tn4
The Issue: Genetic Testing

Laws and Agencies Behind Testing
Federal agencies have taken the responsibility in supervising of genetic testing. The Centers for Medicare and Medicaid Services (CMS) regulates clinical laboratory testing to guarantee laboratory accordance with the Clinical Laboratory Improvement Amendment of 1988. This amendment calls for “quality standards for all laboratory testing to ensure the accuracy, reliability and timeliness of patient test results regardless of where the test was performed showing accuracy and reliability in conducting assays”. The Federal Trade Commission (FTC) manages advertising of clinical tests and items. The Food and Drug Administration (FDA) regulates tests that are marketed as “diagnostic devices,” or, tests produced and assembled by a company and then sold as a kit to scientific laboratories for genetic testing. However, “home-brew” tests, which are both manufactured and implemented by the same laboratory, are not in the realm of the FDA’s parameters. Familiar genetic tests, like the BRCA breast cancer gene tests, are included in this category. Due to this this regulatory immunity, FDA oversight is not required for genetic testing facilities that use, advertise, and vend home brew tests directly to the medical community and the public. The National Human Genome Research Institute (NHGRI) is specifically concerned with issues surrounding the incorporation of clinical utility and validity into genetic testing. The NHGRI is partaking in the creation of advisory guidelines and procedures for the direct marketing of tests to potential clients. The NHGRI is part of the Secretary's Advisory Committee on Genetics, Health, and Society (SACGHS), which is staffed by the National Institutes of Health (NIH) Office of Biotechnology Activities. This organization offers policy recommendations to the Department of Health and Human Services (DHHS) on the extensive range of elaborate medical, ethical, legal and social issues brought forth by genetic testing. With the prospect of health insurance companies categorizing people into risk categories based on genotype and past testing, and thus raising certain costs, society's capability to utilize novel genetic approaches to advance human health and the capacity to conduct the research behind it is endangered. The NHGRI advocates for a federal resolution to this apprehension. The Genetic Information Nondiscrimination Act of 2008 (GINA) is U.S. federal legislation that safeguards Americans from prejudice (in health insurance and employment evaluations) on the grounds of genetic information. It was put into effect in 2009.

Myriad Genetic Laboratories
Myriad Genetic Laboratories help their customers predict risks of acquiring particular hereditary cancers. Myriad can perform tests for breast and ovarian cancer, colorectal cancer, melanoma, and risk for detrimental chemotherapy side effects. A germline mutation in either the BRCA1 or BRCA2 genes can predict susceptibility to breast and ovarian cancer. Because these genes are known as tumor suppressor genes, an inherited mutation will therefore interfere with the body’s natural ability to suppress tumors, and therefore the chance of the person gaining malignant tumors increases greatly. These mutations are inherited in an autosomal dominant pattern. Hereditary Nonpolyposis Colorectal Cancer (HNPCC), or Lynch syndrome is caused by an inherited mutation in a mismatch repair gene, which consequently reduces the body’s ability to repair base-pairing errors that happen during DNA replication. Thus, other mutations may accumulate to augment the probability of cancer development. Mutations in the MLH1, MSH2, MSH6, EPCAM, and PMS2 genes are associated with HNPCC. Gene p16 on chromosome 9p21 is responsible for about 40% of inherited melanoma cases. P16 is a tumor suppressor gene, and
mutations that arise within it lead to uncontrolled cell growth. These mutations follow an autosomal dominant pattern. 5-fluorouracil is common in chemotherapy treatments, but oftentimes chemotherapy can do more harm than good in damaging normal, healthy bodily cells. Mutations in the DPYD and TYMS genes are known to increase risk of dose-limiting adverse effects to 5-fluorouracil chemotherapy. One in four patients has these mutations, and testing for them allows for customization of medical remedies based on the individual’s genotype. A blood sample is needed for most of these tests, and it can be done by a doctor or at home before being sent to Myriad Genetic Laboratories. However, the breast and colorectal cancer risk tests have an oral testing option via a sample of saliva available. Results are usually returned between 7 and 14 days. The website makes it clear that these tests do not assess for cancer, but rather for the level of risk that runs through a family. Though very expensive and requires extensive thought before doing, tests like these that evaluate disease risk should be advocated in cases of family planning, and are more beneficial to families than tests estimating ability.

Atlas Sports Genetics
Atlas Sports Genetics has developed a test that monitors the ACTN3 gene for the R577X variant. The company claims that two copies of this variant gives the individual an inclination for success in endurance events. One copy of this variant allegedly means an equal predisposition to endurance and sprint/power events, and no copies suggest a proclivity for sprint/power events. Atlas Sports Genetics makes this claim off of a knockout study of the gene in mice for mouse muscle metabolism. However, this is just one study in a model organism that needs further exploration (see MacArthur, D.G., et. al). For $169.00, a test kit is provided in the mail within a few days after the customer places the order. The test involves a simple swab of the inner cheek which is then mailed back to the company in a plastic envelope. Roughly two weeks later results are mailed back. For $999.00, the same thing is mailed with an additional home electronic timer and assessment chart. Listed under privacy policy on the company website, statements are included that declare that customer information will be shared with third parties and may be used by them, and personal information may be released to enforce website policies or to protect our the company’s rights, property, or safety. Also, there is the possibility that if the company sells or buys online stores or other assets, information about customers will be among the transferred assets during such transactions. If a third party acquires the company’s assets, consumer information will be transferred and thus made available to other parties. Therefore, by submitting a completed kit back to the company, the customers do not know exactly who can discover their genetic information.

GeneLink
The company GeneLink patented Dermagenetics Skin Health Assessment to measure gene-single nucleotide polymorphisms involved in collagen breakdown, oxidative stress, skin irritation, sun damage, and toleration of environmental and biological pollutants. MMP1, SOD2, GPX1, EPHX, and TNFa are the genes analyzed in this testing process. If there are not any variants in any of these genes, that means the skin is functioning normally and optimally. If there is one variant in any of these genes, it is interpreted that skin is functioning less healthily and thereby aging more rapidly, and two variants in any gene mean that aging is most rapid. Along with test results, the company also provides a skin care system to help manage and treat skin based on genotype.
Conclusion
In essence, any genetic test offered by a large company should be explored with caution and skepticism before actually being bought and used. People with familial diseases who believe they are at high risk may want to discover their genotype. However, this information should be used with caution. Any risk assessment does not imply actually having, or obtaining, a disease. Sometimes a test should be avoided altogether if a person in a high risk category develops anxiety after verifying this information altogether. However, this may be useful for family planning so that two people in high risk categories are aware of the risks if they decide to have children, and take precautionary measures. Tests verifying a more cosmetic or non-detrimental phenotype or ability should be examined with even more caution, because in many cases companies base these tests around very few studies in need of further information and application. Companies may develop these tests for profit, hoping that the public will give in to expensive testing backed by unsubstantiated evidence under the guise of cutting edge and novel science. They play on the minds of curious people, especially parents, who will want to find an aptitude or appealing trait to highlight or emphasize in themselves and future generations. It is as if there is illusion that fate, consisting of future ability, agility, looks, intelligence, disease, deterioration, and so on, relies totally and completely on variants within the genome. One must never ignore the fact that environmental factors play a role in determining fate, and no matter what mutations or variations are present in DNA, one has the power to change lifestyle and habits in order to live a happy, healthy, and fulfilling life.

Myriad Genetic Laboratories: Pass
Atlas Sports Genetics: Fail
GeneLink Dermagenetics: Fail

The Service Project: Raising Awareness

In order to promote awareness on the issue, I looked up potential blogging sites geared toward mothers and parents in general. These are the people most likely to engage in accessory genetic testing for their children and families. They might believe that it is for the greater good of their children, and therefore would be willing to spend copious amount of money in order to get them tested. Yet not all tests have valid backing, and some are what we just might call a “rip off”. Since many are ignorant on the subject (and have no time to do extensive research on the issue because they are busy with their families), I have decided to blog about my findings so that families can save their time and income for more important things. I started by looking up three “mom” oriented blogs and registering for all of them. My user name is listed as Saziz. They are Mom Spark, Mom Zone, and Single Parents Forum. Because of issues of validation of my registration and lengthy waiting times, the only blog where I could post was on Mom Spark. A link from Mom Spark directed me to Mom it Forward, an associated site, and that is where I posted my information. See: <http://momitforward.com/problem-solving-10-tips-to-being-proactive/comment-page-1#comment-74089>. My comment is still awaiting moderation as of June 22, 2011. I do hope to post on the other sites as well, as soon as I become a confirmed member.
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References


Editorial

(Sent to Star Ledger):
Editor,
Use caution with genetic testing! Many tests are designed for companies to profit, and lack substantiated scientific evidence. These tests are not the sole determinant of your future. Use testing only when necessary, like in assessing disease or chemotherapy risk. Do not be fooled by tests that claim to tell you your abilities or accessory traits. Your life is more than your DNA.
~Siana Ziemba