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Screening companies pitch genetic testing for all couples planning to have children

By ACSH Staff — January 7, 2015

Carrier screening is a type of genetic testing performed on couples who are expecting or planning for a baby to see if they may be at risk for passing a genetic disorder on to their children. Carrier screening was previously targeted at people from certain ethnic groups, for example Ashkenazi Jews, who are at higher risk for some diseases including Tay-Sachs disease and Canavan disease. But companies who offer these screening options are now promoting them [1] for everyone.

The tests allow for a quick screening using saliva or blood samples. The tests cost between $600 and $1500, and detect heightened risk for between 98 and 300 diseases. However, carrier screening is usually covered by health insurance, resulting in an out-of-pocket bill of between $150 and $300. *Counsyl*, one of the companies offering carrier screening, says it has screened more than 350,000 people in the past 5 years. After both partners are screened, about 2 percent of couples are identified as carriers for the same genetic disease (many genetic diseases, being recessive, require both parents to supply a gene for the disorder for it to actually appear).

Many experts worry that screening everyone may cause more harm than benefits. Most of the diseases tested for are rare diseases, which are defined by the National Institutes of Health as affecting less than 200,000 Americans. Often the tests find genetic variations in parents, but these variations are not likely to affect the offspring. Genetic screenings can also give false positive and false negative results. Although testing companies are trying to encourage couples to get tested before becoming pregnant, most carrier screening is performed on pregnant women. Detecting a disease in this case leads to the incredibly difficult decision of whether to end the pregnancy. Detecting positive results in both parents before becoming pregnant, however, leads to more options, such as preimplantation genetic diagnosis. PGD allows for genetic profiling of embryos prior to implantation.

Currently, the American College of Obstetricians and Gynecologists (ACOG) recommends universal screening for only cystic fibrosis, although based on family history, other tests might also be recommended. ACOG, together with three other medical-professional groups, expects to issue

an official statement on guidelines for universal screening early this year.

ACSH s Ariel Savransky adds, The question of just what to do with the results of genetic testing is one that is being asked more frequently now, and not only [2]in the case of genetic testing related to individuals planning to have children. Patients should be counseled on just what the results of these tests may mean prior to getting the tests done and be prepared to either be left with indefinite answers or be prepared to take action. Ultimately, just because we have the technology to do something does not mean that we should be using it.

Links

[1] http://www.wsj.com/articles/new-genetic-tests-for-women-who-are-expecting-1420502078

[2] http://acsh.org/2014/09/can-really-learn-undergoing-multigene-testing/