

A Genetic Test for Prospective Parents

By [MICHAEL TOTTY](#)

A three-year-old Silicon Valley start-up, Counsyl Inc., won the Bronze Innovation Award for its simple, low-cost genetic test that can alert prospective parents who might be carrying genes that could cause hereditary diseases in their children.

Counsyl's technology promises a "high quality-of-life impact for families at risk for conceiving children with familial diseases," says Larry Bock, an Innovation Awards judge and special limited partner at venture firm Lux Capital. "This technology bundles previously existing capabilities but with more elegance, access and affordability."

Proponents of universal prepregnancy genetic screening make a bold claim: If these tests were widely available, they could significantly reduce, and possibly eliminate, hereditary diseases. Parents who discovered that they carried the genes that would put their children at risk for these diseases could pursue other alternatives—adoption or in vitro fertilization, for example. Widespread screening also could reduce infant mortality; 20% to 30% of infant deaths are caused by genetic illnesses.

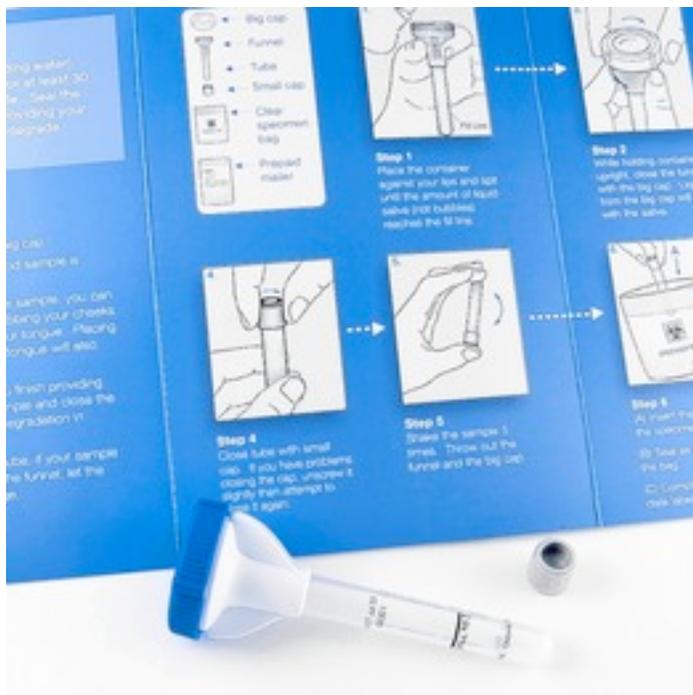
Until recently, however, most genetic tests covered only one disease at a time, required a blood sample and cost between \$500 and \$1,000 each. So both their use and their usefulness were extremely limited. Most prepregnancy genetic testing was performed for one of only a few diseases and only for certain ethnic groups, such as the screening for Tay-Sachs disease offered since the early 1980s to Jewish parents.

Counsyl, based in Redwood City, Calif., has devised a single test that covers more than 100 genetic disorders, including Tay-Sachs, cystic fibrosis and spinal muscular atrophy. The saliva-based test costs about \$350, and the cost typically is covered by most private insurance.

The company uses a customized microarray—essentially a lab on a chip that can simultaneously test for lots of genetic variations at a time—and sophisticated statistical software to hunt for the presence of disease-causing mutations in genes. For the disorders it covers, the test is at least as accurate as common single-disease tests, the company says.

Counsyl's lab received regulatory approval in the spring of 2009. The test was introduced in some clinics in May of last year and became nationally available last October.

— Michael Totty



Counsyl Inc.
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