

## GENETICS

# Too Much Information?

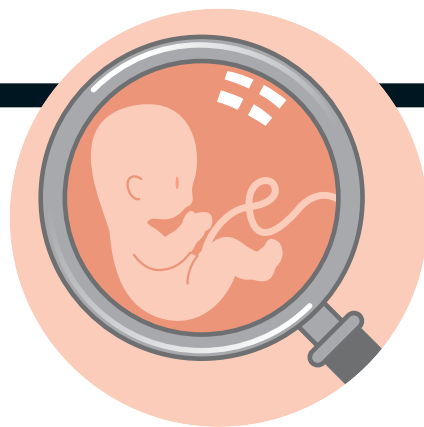
A series of recent breakthroughs means that early, noninvasive genetic tests for fetuses may be just two years away

**Today expectant parents** concerned about the diseases that could afflict their unborn children don't have a lot of options. Blood tests can determine whether parents carry mutations for such genetic diseases as cystic fibrosis and Tay-Sachs, but they can't determine whether the baby will inherit them. And although fetuses can be tested for Down syndrome and other chromosomal abnormalities using amniocentesis or chorionic villus sampling, about 1 percent of procedures cause miscarriage, so many moms opt out. But thanks to a handful of recent breakthroughs, noninvasive prenatal tests may soon be available that diagnose genetic diseases before birth using samples of a mother's blood—an exciting possibility that also raises difficult questions about how they should be regulated and administered.

What makes noninvasive tests possible is that a pregnant woman's blood contains free-floating copies of her fetus's genes, as chemical pathologist Dennis Lo of the Chinese University of Hong Kong discovered in 1997. Last December in *Science Translational Medicine*, Lo reported on a method of sequencing individual fetal genes and counting individual fetal chromosomes in a mother's blood to establish whether a fetus carries disease-causing mutations or chromosomal abnormalities. Fetal genes inherited from the mother are identifiable because they are present in higher-than-normal concentrations in the mother's blood; gene variants not shared by the mother are assumed to be inherited from the fa-

ther. In a follow-up article in the *British Medical Journal*, Lo tested his approach on 753 pregnant women. He counted the proportion of DNA molecules found in the mother's blood that were derived from chromosome 21—individuals with Down syndrome have three copies rather than the normal two—and accurately diagnosed 100 percent of the fetuses who would be born with the disorder. The test, Lo says, would prevent “98 percent of invasive procedures, such as amniocentesis.” The trial did, however, report three false positives, so all positive results would need to be followed up with more invasive tests.

San Diego company Sequenom is developing a test based on Lo's method that should be available within two years. Tests for other conditions, including cystic fibrosis, Tay-Sachs, hemophilia and sickle cell disease, may be four to five years away.



The big question is how these tests will affect parental decisions: Will couples abort affected fetuses? How will the prevention of rare diseases affect research funding for their cures? Will tests arise that allow parents to select fetuses based on superficial traits, such as eye or hair color? Stanford University law professor Henry T. Greely says that the U.S. government is doing nothing to address these questions. In addition, doctors will need guidelines to help them counsel test takers properly. Otherwise “you will end up with families getting information they're not ready to get,” says Siobhan Dolan, an obstetrician at Montefiore Medical Center in New York. In a few years, we might have too many options rather than too few. —Melinda Wenner Moyer

### BY THE NUMBERS

# 225 miles

Current length of the U.S.'s only high-speed-rail line, part of the Acela Express, which debuted in 2000.

**2,594 miles:** Current length of China's high-speed-rail system.

**4,124 miles:** Current length of high-speed-rail systems in Europe.

An advertisement for Gorilla Glue. The background is a dark, textured surface. On the right, a hand with thick, dark, hairy fingers holds a bottle of Gorilla Glue. The bottle is orange and black with a yellow label. The label features a gorilla's face and the text "INCREDIBLY STRONG", "GORILLA GLUE", and "100% WATERPROOF". Below the bottle, the text "For The Toughest Jobs On Planet Earth" is written in a bold, sans-serif font. At the bottom, it says "Bonds: Wood, Stone, Metal, Ceramics, Foam, Glass &amp; More!". On the left, the text "For the Toughest Jobs on Planet Earth" is written in a large, bold, white font. Below this, it says "With its incredible versatility and strength, Gorilla Glue is the ultimate solution for all your adhesive needs. Bonds wood, stone, metal, ceramic, foam, glass and much more!". At the bottom left, it says "FOR THE TOUGHEST JOBS ON PLANET EARTH" and "GORILLA TOUGH". At the bottom center, it says "1-800-966-3458 Made in USA". On the right side, there is a small vertical copyright notice: "© 2011 Henkel Bala Company".