World Changing Ideas

Ten thoughts, trends and technologies that have the power to transform our lives

TECHNOLOGY IS ALL AROUND US, EXPANDING THE LIMITS OF WHAT IS POSSIBLE. BUT EVERY ONCE in a while, some invention or insight has an outsize effect; it creates a large discontinuity, dividing history into "before" and "after." The steam engine, the transistor, the World Wide Web—each of these ideas seemed to emerge from nowhere to change our world in fundamental ways. Which key technology will arise from today's vast cauldron of innovation to become tomorrow's world changing idea? It's impossible to know, of course, but we know it will come.

Here are 10 candidates—10 new ideas and technologies that could rewrite the rules. What if we could build robots that turn waste into fuel? Or harness the power of video games (yes, video games) to make ourselves do the right thing? What if the "junk" in our DNA is actually as important as our genes? What if insects hold the secret to fending off cyberattacks? Welcome to the World Changing Ideas 2010 edition. —*The Editors*

Photograph by Mark Hooper

HEALTH AND MEDICINE

One Hundred Tests

A cheap diagnostic warns couples against passing rare genetic diseases to their offspring *by Mary Carmichael*

WHAT WOULD YOU PAY to ensure that your children would not be born with disabling or fatal recessive genetic diseases? The obvious answer is "anything," but that's not what most people actually do. Individual screening tests can already identify silent carriers of many single faulty recessive genes—the kind that, when inherited in double (one copy from each parent), can lead to conditions such as cystic fibrosis and Tay-Sachs disease. But almost no one gets tested for all these mutations before conceiving because it would be too expensive—the dozens of tests cost several hundred dollars apiece. Because each potentially dangerous mutation is rare, most people choose instead to play the odds and hope their children will be healthy—a strategy that sometimes results in tragedy.

That isn't necessary anymore, thanks to a simple saliva test made by a company called Counsyl that interrogates the genome for more than 100 disease-causing recessive traits. In one sense, it is like having many traditional, separate tests combined; from a medical standpoint, it yields essentially the same results. But it does so in one go, at a cost of $350.

Traditional tests for recessive variants work by zooming in on specific genomic regions associated with each disease. In some cases, the tests sequence the genes to determine if mutations are present. Counsyl's test, on the other hand, does not involve sequencing. Instead it looks for single-nucleotide polymorphisms (SNPs), tiny typos in the genome where one base has been replaced with another. Some SNPs contribute to disease; others are linked to genes that do. Because SNPs are small, it is cheaper to identify one of them than it is to sequence an entire gene or region of a chromosome, which may consist of millions of bases. The company says the test picks up mutations with greater than 99 percent sensitivity and specificity—that is, it rarely yields false positives or negatives—and has recently begun to publish results to that effect.

So far Counsyl's test has mostly been used by infertility patients. Pasquale Patrizio, director of the Yale Fertility Center, is one of the doctors offering it. (He is also on Counsyl's board of advisers.) He says it is useful in treating couples who have suffered repeated miscarriages but do not know why. In some cases, their losses may turn out to be caused by recessive genes that prevent the fetus from coming to term. "For us it was really a breakthrough to have such a comprehensive screening test," Patrizio says. But of course, many people who carry recessive genes manage to conceive without the assistance of a fertility clinic. They find out about their genetic bad luck later, once their children become ill.

Couples who test positive can plan ahead. They might choose in vitro fertilization, combined with preimplantation genetic diagnosis, to choose embryos that do not carry disease genes. Or they might decide to adopt. Either way, the numbers of ill children in the population at large would drop. Most of the double-recessive diseases are research "orphans"; because they are rare, little money is put into studying them. The Counsyl test is the best present hope for ensuring that fewer people are afflicted with them.

Counsyl may run into some roadblocks on its way to wide use. Some people fear it will open the door to "designer babies." Widespread testing for rare genetic diseases, the argument goes, opens the door to testing for traits that do not indicate disease, such as height and intelligence.

Counsyl's technology can't produce designer babies, however, because it tests for single genes, not the poorly understood, multilevel genetic networks involved in complex phenomena such as intelligence. "There isn't going to be an IQ gene or a musical ability gene," says Harvard University psychologist Steven Pinker, who is advising the company on the ethical issues surrounding personal genomics. Besides, he notes, "if any group would have fears about eugenics it would be the Jews"—yet as a group they have embraced the old, expensive recessive-gene tests because Ashkenazi Jews are more likely to carry some deleterious recessive variants. Pinker, who is Jewish, carries the one that causes familial dysautonomia, an incurable disease that halts neuron development. He found out only when he took the Counsyl test. "My wife is a carrier, too," he says. "We met too late in life to have children, but if we had met a few years earlier we would have been playing roulette." At least now other couples can choose not to.

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