Test could screen for more than 400 childhood diseases

Jan. 12, 2011
Courtesy of Science Translational Medicine
and World Science staff

A new genetic test could be used routinely by doctors to screen for 448 severe childhood diseases in couples before they have children, according to a study.

The test checks for recessive diseases—conditions caused by mutations that generally don't affect the parents but can affect their children if both parents are carriers. The test is designed to tell parents whether this risk exists.

Combined with genetic counseling, the technology may reduce the incidence of severe recessive childhood diseases and help speed up diagnosis of these disorders in newborns, researchers said.

But scientists also fret that the psychological burden of such a test, along with other issues related to the dynamics of reproduction, will need to be thoroughly addressed before preconception screening is available to the general public.

Inherited childhood diseases as a whole account for about one in five infant deaths and one in 10 child hospitalizations, according to the researchers, who reported their preliminary findings in the Jan. 12 issue of the research journal *Science Translational Medicine.*

In the last few decades, over a thousand genes implicated in these diseases have been identified, yet preconception screening for couples in the United States is only recommended for five diseases in specific populations. These conditions include fragile X syndrome, cystic fibrosis, and Tay-Sachs disease.

Now, Stephen Kingsmore at the National Center for Genome Resources in Santa Fe, N.M. and colleagues have developed a preconception carrier test capable of screening several hundred DNA samples simultaneously for 448 recessively inherited childhood diseases.

The test is based on a combination of techniques, including "next-generation" gene sequencing, the scientists said. Using the new sysytem to screen around 100 unrelated people, they found that on average, each person tested harbored two to three severe childhood disease mutations. They also discovered that about 10 percent of disease mutations in commonly used databases are incorrect, suggesting, they said, that disease mutation databases need careful scrutiny.