

Kit expands genetic screening options for Ashkenazi Jewish couples

When a Jewish couple is planning their wedding or anticipating starting a family, they probably aren't thinking much about rare genetic conditions. But JScreen, an educational and screening program, urges couples to add genetic testing to their to-do list. And by offering home-based testing, JScreen hopes to eliminate any obstacles to this process.

Based at Emory University's Department of Human Genetics, JScreen (jscreen.org) provides a Web-based portal for individuals to request a genetic-screening kit. Participants provide a saliva sample — most genetic tests involve a blood draw — and mail it back for analysis. Before receiving the kit, participants must view an educational video and enter health information that is reviewed by an Emory genetic counselor.

"We all carry [recessive genes for] various genetic diseases. We just don't know what they are," said Karen Grinzaid, a genetic counselor and instructor at Emory University School of Medicine and the senior director of outreach initiatives for JScreen.

The problem occurs when both parents are carriers of the same disease. In that case, each of their offspring has a 25 percent chance of manifesting the condition.

According to Emory's Department of Human Genetics, about one in five Ashkenazi Jews in the U.S. carries a genetic disease. However, most don't have a family history of the disease and are unaware of their status of carriers. In fact, 80 percent of babies with genetic diseases are born to parents with no known family history of that disease.

"The only way to know if you are a carrier for a Jewish genetic disease is to have an affected child or be screened," Grinzaid said. "For the vast majority of couples, genetic screening gives couples reassurance that they're not at risk."

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