TOOLBOX

Quick test for fragile X may be ideal for developing nations

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Dried spots of blood taken from infants at birth can help clinicians screen for fragile X syndrome in countries with limited resources, according to a study published 11 October in *Genetic Testing and Molecular Biomarkers*[1].

Fragile X syndrome is a developmental disorder caused by expansion of the FMR1 gene and afflicts about 1 in 250 individuals. It is characterized by intellectual disability, language delay and, often, autism.

FMR1 typically has up to 55 repeats of a set of three DNA base pairs. The full mutation, which comprises more than 200 repeats, leads to the syndrome. Repeats in the 55-200 range result in a pre-mutation that can cause **infertility in women**.

Fragile X syndrome is diagnosed using a combination of two methods: polymerase chain reaction (PCR), which amplifies a section of DNA to detect its length, and Southern blots, which probe for specific DNA regions within an individual's genome. Southern blotting is required because PCR cannot easily amplify repeat regions, but the two methods together are time-consuming and expensive.

Several pharmaceutical companies have proposed **quick screens for fragile X syndrome** based on optimized PCR techniques that would not require Southern blots.

Researchers refined one of these methods, published in 2008, to detect DNA from dried blood spots[2]. This approach could lower the cost of screening from hundreds of dollars to about $5 per individual, they say.

In the new study, researchers in Indonesia used samples from 176 individuals, including 112 diagnosed with intellectual disability, 32 with a diagnosis of autism and 32 with a family history of

fragile X syndrome.

The method diagnosed one individual with intellectual disability as having fragile X syndrome and detected six full mutations and seven pre-mutations among individuals with a family history of the disorder. Standard diagnostic techniques confirmed the results in these 14 individuals.

The testing method will be especially valuable for screening populations and for determining the **prevalence** of fragile X syndrome in countries that do not have access to genetic testing, the researchers say.

References:

1: Winarni T.I. *et al. Genet. Test Mol. Biomarkers* Epub ahead of print (2011) **PubMed**

2: Tassone F. *et al. J. Mol. Diagn.* **10**, 43-49 (2008) **PubMed**