TOOLBOX

Quick, cheap tests decode fragile X mutation

BY **NICHOLETTE ZELIADT**

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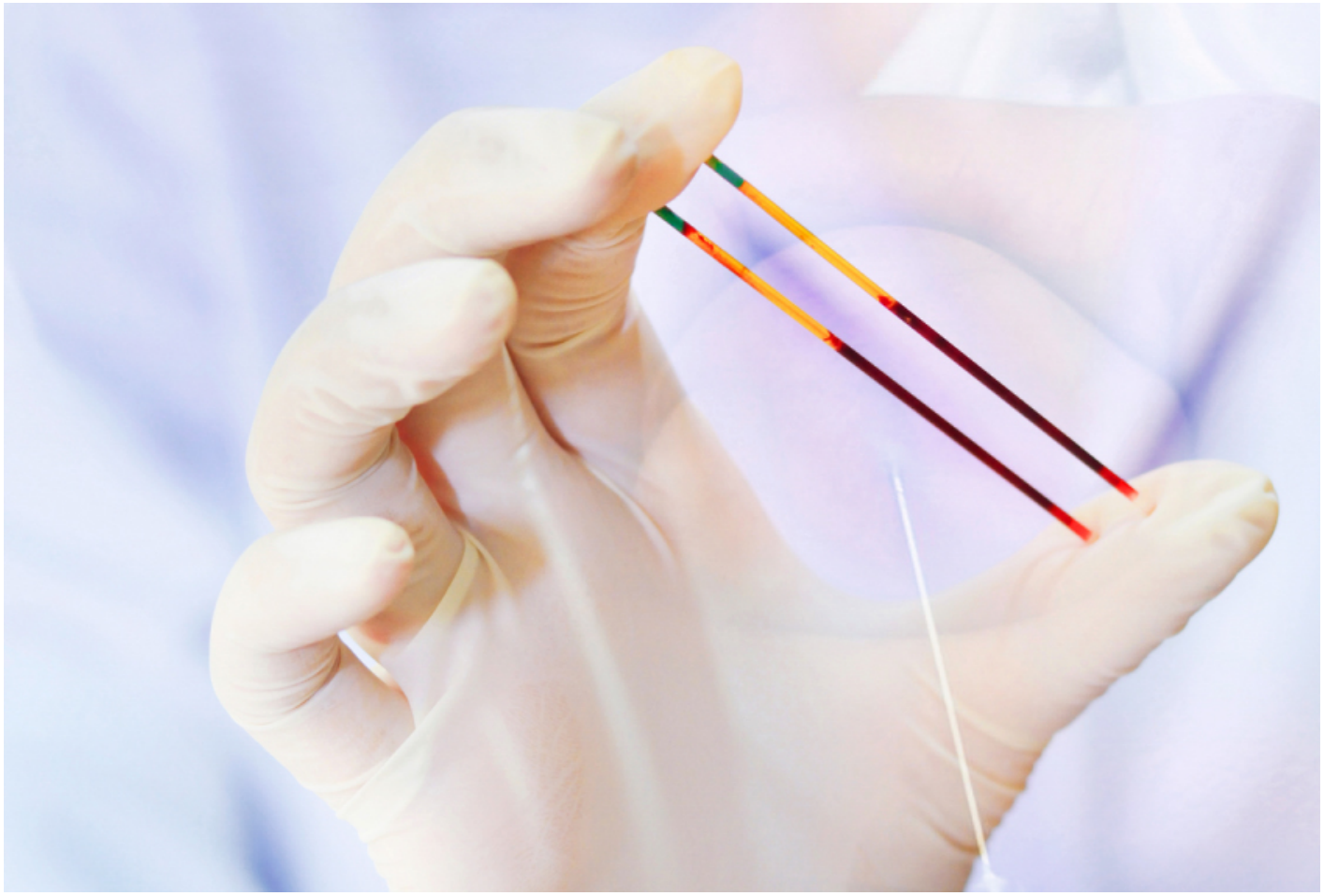

An inexpensive new set of tests allows researchers to rapidly analyze the genetic glitch underlying **fragile X syndrome**. The syndrome is one of the most common **inherited causes** of intellectual disability and autism.

The tests take less than 48 hours and cost under $5 per sample, opening the door to population-based screening for fragile X and related conditions.

The researchers describe their tests in the September issue of the *Journal of Molecular Diagnostics*[1].

Fragile X syndrome typically arises from expansion of the **FMR1** gene: People with the syndrome carry more than 200 repeats of a particular DNA sequence. Having 55 to 200 repeats can also result in **related conditions**. The excess repeats attract markers called methyl groups that shut down the gene's expression. The extent of methylation can vary among individuals.

Researchers typically analyze repeat length using a DNA-amplifying technique called polymerase chain reaction (PCR), but the method often fails when there are more than 100 repeats. To solve this problem, researchers sometimes add a second method called **a Southern blot**. The combination can cost hundreds of dollars and take up to several weeks to complete.

In the new study, researchers tweaked an established PCR assay so that it can amplify long repeats. When they tested it on isolated DNA and on cultured cells from people with fragile X syndrome, they detected more than 900 repeats in one sample.

The researchers also developed a modified version of the test that can simultaneously estimate methylation levels.

REFERENCES:

1. Hayward B.E. *et al. J. Mol. Diagn.* **18**, 762-774 (2016) **PubMed**