# New NIST reference material reinforces fragile-x screens

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A new Standard Reference Material from the National Institute of Standards and Technology (NIST) will help clinical genetics labs improve the accuracy of their diagnostic tests for the most common cause of hereditary mental retardation.

"Fragile X Syndrome" is a genetic mutation affecting approximately one in 3,600 males and one in 4,000 to 6,000 females. It has been linked to several physical abnormalities and to intellectual problems ranging from minor learning disabilities to severe mental retardation and autism. The mutation is characterized by an excessive number of repeats of a sequence of three nucleotides (the chemical building blocks of DNA) within a particular gene on the human X chromosome.

Proper diagnosis depends critically on accurate counts of the number of triplet repeats. Individuals with up to 44 repeats are normal; individuals with 55 to 200 repeats fall into the premutation category (unaffected, but the number of repeats can increase in their children, who can then be affected); and those with 200 or more repeats have the full mutation and Fragile X syndrome. In general, the symptoms of the disorder become more severe as the number of repeats increases.

To assist clinical diagnostic and genetic testing laboratories in accurately counting fragile-X repeat sequences, NIST has developed a new reference material that can be used as a check on test procedures and for quality control. SRM 2399, "Fragile X Human DNA Triplet Repeat Standard" consists of nine samples of DNA measured and certified by

NIST for triplet repeats ranging from 20 to 118. The triplet repeat standard joins more than 50 reference materials produced by NIST for quality control in clinical testing.

Source: NIST