

# New test shows promise for accurate diagnosis of Turner syndrome

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As a child grows, a short stature is not usually cause for concern, but it is often the only sign of a condition called Turner syndrome. Prevalent in girls, Turner syndrome is a genetic defect that short-circuits normal growth and leads to cardiac and renal problems. It is not commonly detected until age 10 or older when a youngster's unusually short height raises suspicions.

This lag before diagnosis of the condition can delay the start of growth hormone therapy, which can help in achieving normal or near-normal adult stature. Yale School of Medicine researchers are aiming to close this lag time with a new inexpensive, accurate and practical [diagnostic test](#) for Turner syndrome that can be done in a doctor's office. Their research is published online and in an upcoming March 2011 print issue of The Endocrine Society's [Journal of Clinical Endocrinology & Metabolism](#).

"We've developed a practical test that can be used to test large numbers of girls, and is much quicker and less expensive than the current method called cytogenetic analysis by karyotype," said the study's lead author Scott Rivkees, M.D., professor of pediatrics at Yale School of Medicine. "The new test would also provide the benefit of early detection of other health conditions associated with TS, such as potential renal and cardiac problems."

Turner syndrome occurs when an X-chromosome is completely or partially deleted. The syndrome affects one in 1,500 to 2,000 females.

Untreated girls with TS achieve an average height of 4 feet, 8 inches.

Rivkees and his co-authors developed the test based on a quantitative method of genotyping to detect X-chromosome abnormalities. The test was developed and validated from DNA samples from more than 500 individuals. Of 90 clinically confirmed TS individuals tested, the assay correctly identified 87.

"Because of the small amount of DNA needed for the test, sample DNA can be extracted from cheek swabs, or from newborn screening blood spots that are routinely collected," Rivkees said. "If broadly used in the clinical setting at young ages, this test can prevent the delayed recognition of TS."

A U.S. patent was recently issued to Yale University for "DNA Diagnostic Screening for [Turner Syndrome](#) and Sex Chromosome Disorders."

Provided by Yale University

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