

New test shows promise for accurate early diagnosis of Turner syndrome

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A recent study accepted for publication in The Endocrine Society's *Journal of Clinical Endocrinology & Metabolism* (JCEM) has demonstrated a novel and accurate test for early diagnosis of Turner syndrome. Turner syndrome affects one in 1,500 to 2,000 female live births and early diagnosis allows for the timely management of short stature and co-morbid conditions including cardiac and renal problems.

Turner syndrome (TS) is the most common genetic problem affecting girls with short stature. Average adult height in untreated girls with TS is 4 feet, 8 inches, yet with early diagnosis and initiation of growth hormone therapy, normal or near-normal adult stature can be achieved. Unfortunately, the vast majority of girls with TS go unrecognized until after 10 years of age. This new study suggests a new way to diagnose TS to help prevent delayed recognition.

"We have developed a novel approach for diagnosing TS that can be used to practically test large numbers of girls and is much quicker and less expensive than the current methods," said Scott Rivkees, MD, of Yale University School of Medicine in New Haven, Conn. and lead author of the study. "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."

TS occurs when an X-chromosome is completely or partially deleted. In this study, researchers developed a test based on a quantitative method of genotyping to detect X-chromosome abnormalities. Of 90 clinically-

confirmed TS individuals tested, the assay correctly identified 87 (96.7 percent).

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

More information: The article, "A Highly Sensitive, High-Throughput Assay for the Detection of Turner Syndrome," appears in the March 2011 issue of *JCEM*.

Provided by The Endocrine Society

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