Electronic Health Record Algorithm Enhances Early Detection of Turner Syndrome

A simple algorithm embedded in a pediatric practice's electronic health record (EHR) system could improve early detection of Turner syndrome, concluded a study conducted at Cincinnati Children's.

One in 2,000 girls are born with a chromosome abnormality that causes Turner syndrome. With early detection and initiation of growth hormone therapy, most girls with Turner syndrome can reach an adult height within normal range.

"At the Turner Syndrome (TS) Center at Cincinnati Children's, we see patients being diagnosed all the time between 7 to 9 years of age," says Philippe Backeljauw, MD, director of the center and clinical director of the Cincinnati Center for Growth Disorders (CCGD) at Cincinnati Children's.

"Using an EHR algorithm is very effective in identifying patients with Turner syndrome who otherwise would have been missed," he explains. "If this can be used in health systems and large primary care practices, many more patients will be picked up at an earlier age."

Early Detection Encouraged in Primary Care Settings

"Algorithm-Driven Electronic Health Record Notification Enhances the Detection of Turner Syndrome" was accepted for publication in 2019 by the Journal of Pediatrics. It studied electronic health records of 216 girls ages 3 to 18 years with idiopathic short stature (ISS).

The study showed that a best practice alert guided by an algorithm based on both absolute height standard deviation and a deflection from the mid-parental height (MPH) percentile (or target height curve) will successfully identify patients at risk for Turner syndrome.

"It's a decision support system that could be really valuable in the primary care setting. Any EHR system that collects data on a child's height and parents' heights can use the algorithm for female patients," Backeljauw says. "If the patient fits the criteria, there will be a pop-up to alert the physician to think about a Turner diagnosis."

As the paper states, there are multiple advantages to an early diagnosis: "In addition to improving height outcomes, prompt detection of Turner syndrome is crucial, as some patients have unsuspected cardiovascular anomalies and are, with advancing age, at increased risk for aortic dilation, and even dissection and rupture.

"Health-related quality of life measures also improve when treatments such as hormone-replacement therapy for primary ovarian failure are initiated within an age-appropriate time frame, allowing for pubertal induction and progression in congruence with peers. Furthermore, early recognition of cognitive and psychosocial deficits can allow for tailored interventions that help promote optimal educational attainment and adaptive skills."

For more information, contact Cincinnati Children's Division of Endocrinology at **513-636-4744.**

The Turner Syndrome Algorithm:

Identify patients with a height standard deviation (SD) <-2, body mass index (BMI) >5th percentile, and absence of chronic illness, as evidenced by International Statistical Classification of Diseases and Related Health Problems, 10th Revision. Further select patients who have height data available on both parents are >1 SD below mid-parental height (MPH) and have not yet received genetic testing. The MPH is defined as the mean height standard deviation score (HSDS) of both parents.