

Turner Syndrome Guidelines Target Osteoporosis

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Updated guidelines on evaluating and treating girls and women with Turner syndrome advise against the practice of delaying puberty to increase height and emphasize the importance of early diagnosis and estrogen treatment.

The guidelines from the international, multidisciplinary Turner Syndrome Consensus Study Group detail how children should be evaluated and cared for, emphasizing, for example, the importance of comprehensive educational evaluation in early childhood.

The guidelines offer age-specific suggestions for ovarian hormone replacement and say that “ideally, natural estradiol and progesterone, rather than analogs, should be delivered by transdermal or transmembranous routes so as to mimic age-appropriate physiological pat-

Delaying estrogen therapy until a girl is 15 years old in order to maximize height undervalues the psychosocial importance of age-appropriate puberty.

terns as closely as possible.” Regimens can vary to meet individuals’ tolerance and preference, however, and “the most important consideration is that women actually take ovarian hormone replacement,” the authors say (J. Clin. Endocrinol. Metab. 2007;92:10-25).

Without it, the risk of significant osteoporosis is high. “These women can have severe osteoporosis at 25,” Dr. Carolyn A. Bondy said in an interview.

“I have a 30-year-old patient who has lost 2 inches of height and has a hump.”

Estrogen therapy often is required to induce pubertal development (30% or more will undergo some spontaneous pubertal development), but experts used to recommend delaying estrogen therapy until age 15 to optimize height potential.

Today, Dr. Bondy said, the consensus is that such delay undervalues the psychosocial importance of age-appropriate puberty. Recent evidence also suggests that low-dose estrogen does not inhibit growth hormone-enhanced increases in stature. “There’s a new focus on natural, sensitive, and timely puberty induction,” she said.

“The care of adults with TS has received less attention than [has] the treatment of children, and many seem to be falling through the cracks with inadequate cardiovascular evaluation and estrogen treatment,” say the new guidelines, published in the *Journal of Clinical Endocrinology & Metabolism*.

On the other hand, while medical care must be improved and while many questions about care “remain unanswered,” the experts “realize now that we have a lot more well-functioning people with TS,” according to Dr. Bondy, who is chief of the developmental endocrinology branch at

the National Institute of Child Health and Human Development in Bethesda, Md. She chaired the consensus conference and guideline-writing committee for the consensus group, which met last summer to update the recommendations that were issued in 2001. The guidelines mainly represent “consensus judgments” rather than evidence-based conclusions, the committee noted in its document.

The clinical spectrum of TS is “much broader and often less severe than that de-

scribed in many textbooks”—a finding that seems at odds with a “high elective abortion rate for incidentally diagnosed 45,X and 45,X/mosaic fetuses,” the guidelines say. This means that the content of prenatal counseling “needs updating” with the input of TS patient and parent groups, the document says.

Recent reports of an often-normal quality of life for those receiving comprehensive medical care should encourage—though mitigate—the efforts of physicians to di-

agnose TS as early as possible and better appreciate its many consequences, she said. Adults with TS should then be regularly screened for hypertension, diabetes, dyslipidemia, aortic enlargement, hearing loss, osteoporosis, and thyroid and celiac diseases. Recent studies have also suggested a broader spectrum of cardiovascular abnormalities than were previously recognized, and the consensus group agreed to bring “the heart to the forefront,” Dr. Bondy said. ■



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