Updated Turner Syndrome Guidelines Issued

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Contributing Writer

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, estrogen treatment, and more comprehensive cardiovascular evaluation—including the use of diagnostic MRI—than is typically done today.

Although 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 experts also clearly state that care for adults is more often falling short.

"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," Dr. Carolyn A. Bondy said in an interview.

Dr. Bondy, chief of the developmental endocrinology branch at the National Institute of Child Health and Human Development in Bethesda, Md., chaired the consensus conference and guideline-writing committee for the consensus group, which met last summer to update recommendations 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 described 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.

That's especially true now that the American College of Obstetricians and Gynecologists is recommending that all women, regardless of their age, be offered screening for Down syndrome. Parents who receive a Turner syndrome diagnosis from such screening (TS can be an incidental finding) must be given information about the broad phenotypic spectrum of the syndrome and the high quality of life for many patients, Dr. Brody said.

Recent reports of an often-normal quality of life for those receiving comprehensive medical care should encourage—not mitigate—the efforts of physicians to diagnose TS as early as possible and better appreciate its many consequences, she said.

The diagnosis should be considered in any female with unexplained growth failure or pubertal delay or any constellation of the syndrome's characteristic physical features, the guidelines say.

"Regrettably, late diagnosis of TS, even in adults, is still a problem. No matter what the age of the patient, a full work-up with assessment of congenital malformations should be performed, including all screening tests recommended for younger patients," the document says.

Adults with TS should then be regularly screened for hypertension, diabetes, dyslipidemia, aortic enlargement, hearing loss, osteoporosis, and thyroid and celiac diseases (J. Clin. Endocrinol. Metab. 2007:92:10-25).

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 patterns 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.

Without it, the risk of significant osteoporosis is high. "These women can have severe osteoporosis at 25," said Dr. Bondy. "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.

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. "There's a new emphasis [in the guidelines] on the fact that everyone needs cardiovascular screening—from the newborn to the woman who's 20 and just found out she's infertile [and has TS] to the woman who's 40 and just got the [TS] diagnosis."

And while echocardiography usually is adequate for screening infants and young girls, MRI also must be performed in older girls and adults.

Reports of fatal aortic dissection during pregnancy and the postpartum period have raised concern about the safety of pregnancy in TS, and "preconception assessment must include cardiology evaluation with MRI of the aorta," the experts say. ■

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