Reply to "Practice guidelines for short stature"

I read the practice guidelines for the evaluation of short stature in the June issue of *Genetics in Medicine* with great interest. As a geneticist and a woman with Turner syndrome, I appreciated the number of comments in the body of the article about Turner syndrome and the importance of karyotype in the evaluation of short stature but was somewhat disappointed to see no karyotype mentioned in the diagnostic algorithm. This practice guideline would otherwise be useful not only to geneticists but also to other physicians, but I fear that quickly referring to the convenient algorithm might lead to overlooking this important step early in the workup of every child (especially girls) with growth failure.

On behalf of the Turner Syndrome Society of the United States, I applaud ACMG for addressing this important issue.

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Disclosure: The author declares no conflict of interest.

To the Editor:

Thank you for your comment on the "Genetic evaluation of short stature" guideline published in the June 2009 issue. As you know, we mentioned Turner syndrome as the most common diagnosis likely to present with short stature in the text of the guideline. Because this guideline was written with the intent of being directed toward medical geneticists in creating the algorithm, we considered the diagnosis of Turner syndrome to fall within the algorithm as a "recognized syndrome" that leads to appropriate/specific testing. For those individuals with Turner syndrome with isolated short stature, there is some controversy whether a karyotype should be performed first or if array comparative genomic hybridization (CGH) should now be the first option and that point was discussed in detail through the committee process. The wording in the algorithm (specific testing if available) would allow the geneticist some latitude to order either a karyotype or array CGH as a first-line test in this situation recognizing that there may be institutional and regional differences in practice. Obviously, the array CGH will detect Turner syndrome and variants, although we recognize that this may not be the most cost-effective test in that case. In general, for other individuals with short stature with or without other obvious minor or major malformations, and no recognizable diagnosis, the array CGH is the most efficient diagnostic test.

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