

CORRIGENDUM: Correction to [GIM.0b013e3181c684b2](#)

Laboratory guideline for Turner syndrome. Wolff DJ, Van Dyke DL, Powell CM. Working Group of the ACMG Laboratory Quality Assurance Committee. *Genet Med* 2010;12(1):52–55.

Please note that Section E5.1.2.2 of the Laboratory Standards and Guidelines which currently state “Cases being studied for possible sex chromosome abnormalities, in which mosaicism is common, should include a minimum of 30 cells counted.” has been changed to “Cases being studied for possible sex chromosome abnormalities, in which mosaicism is common, should include the standard 20-cell assessment. If mosaicism is confirmed, the analysis is complete. A minimum of 10 additional metaphase cells should be evaluated when one cell with a sex chromosome loss, gain or rearrangement is observed within the first 20 cells analyzed.”

This change is based on evidence provided by Wiktor et al. (2009) and Thangavelu and Mascarello (personal communication) in which the authors demonstrate that routine analysis of 30 cells for all patients suspected of having sex chromosome abnormalities is of limited value and that the standard 20-cell analysis used for patients with other diagnoses is sufficient to identify the clinically important cell types(s). Wiktor et al. studied 1,580 patients with a sex chromosome abnormality to determine if analysis of only 20 cells would decrease costs without sacrificing diagnostic power. Of the 1,580 cases, 453 (28.7%) were mosaic. In 1,578 of 1,580 (99.9%) cases, the sex chromosome abnormality was suspected (based on finding a single cell with a structural aberration or a gain of a sex chromosome or one or two cells with loss of a sex chromosome) or confirmed within the first 20 metaphase cells counted. Thangavelu and Mascarello (manuscript in preparation) examined records from 1,081 patients with a clinical indication of possible sex chromosome abnormality, of which 24 were mosaic. In only one case was a new cell type found after the evaluation of the first 20 cells and that finding did not have clinical significance. For five cases, there was a single cell with an extra sex chromosome or a structural aberration or two cells with loss of the X that led to a suspicion of mosaicism.

This change affects the Laboratory Guideline for Turner syndrome, sections 3.2.2.1 and 3.2.2.3 which reference the 30-cell analysis. This Guideline will be modified on the online version of the Laboratory Standards and Guidelines to include the updated change above.

1. Wiktor AE, Bender G, Van Dyke DL. Identification of sex chromosome mosaicism: is analysis of 20 metaphase cells sufficient? *Am J Med Genet A* 2009;149A: 257–259.