

Response to Metcalfe and Archibald

To the Editor: My thanks to Drs. Metcalfe and Archibald for their supportive comments in their letter, “Fragile X Population Carrier Screening,”¹ and for drawing attention to their earlier and ongoing studies on population-based carrier screening for fragile X (*FMR1*) trinucleotide repeat expansions. Although not explicitly stated in my commentary,² what I had in mind, perhaps provincially, was National Institutes of Health– or professional organization–sponsored pilot studies in the US population, along the lines of the cystic fibrosis carrier screening pilot studies in the 1990s that set the stage for nationwide carrier screening for that disorder. The experiences of Metcalfe and Archibald in Australia, along with other fragile X screening pilots in Taiwan, Israel,

Finland, and the United States as summarized in their earlier meta-analysis,³ are extremely valuable as we begin to formulate evidenced-based practice guidelines in this area.

Wayne W. Grody, M.D., Ph.D.¹

¹UCLA School of Medicine, Los Angeles, California, USA.

E-mail: wgrody@mednet.ucla.edu

REFERENCES

1. Metcalfe SA, Archibald AD. Fragile X population carrier screening. *Genet Med* 2012, this issue.
2. Grody WW. Expanded carrier screening and the law of unintended consequences: from cystic fibrosis to fragile X. *Genet Med* 2011;13: 996–997.
3. Hill MK, Archibald AD, Cohen J, Metcalfe SA. A systematic review of population screening for fragile X syndrome. *Genet Med* 2010;12: 396–410.

doi:[10.1038/gim.2012.1](https://doi.org/10.1038/gim.2012.1)