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Letter to the Editor

Congenital Nystagmus in a [46,XX/45,X] Mosaic Woman From a Family With X-Linked Congenital Nystagmus

To the Editor:

We wish to comment on the letter to the editor by Berry and Docherty regarding our publication, "Congenital Nystagmus in a (46,XX/45,X) Mosaic Woman From a Family With X-Linked Congenital Nystagmus" [Gutmann et al., 1991]. It is correct that in the 1960s and 1970s it was well established that loss of X and Y chromosomes occurred regularly in association with increasing age. Berry and Docherty suggest that the mosaicism in our patient is likely to be secondary to the woman's age of 76 and were surprised that her expression of X-linked congenital nystagmus was attributed to the finding that 12% of her cells showed monosomy X. She is a member of a multigenerational family with numerous males who exhibit X-linked congenital nystagmus. The percentage of cells that were 45,X in this patient (6/50, 12%) is significantly different from that in females of comparable age (23/999, 2.3%) using Fisher's exact test $P = .002$ [Galloway and Buckton, 1978]. Thus, we propose that her level of mosaicism, in addition to potential skewing of X-chromosome inactivation, is contributory to her expression of an X-linked disease.

REFERENCES

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Gutmann DH, Brooks ML, Emanuel BS, McDonald-McGinn DM, Zackai ES (1991): Congenital nystagmus in a [46,XX/45,X] mosaic female from a family with X-linked congenital nystagmus. Am J Med Genet 39:167–169.

David H. Gutmann
Department of Neurology
The University of Michigan
Ann Arbor, Michigan

Elaine H. Zackai
Clinical Genetics Center
Beverly S. Emanuel
Cytogenetics Laboratory
The Children's Hospital of Philadelphia
Philadelphia, Pennsylvania

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Address reprint requests to Dr. Elaine H. Zackai, Clinical Genetics Center, The Children's Hospital of Philadelphia, 34th and Civic Center Blvd, Philadelphia, PA 19104.