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COMMENTARY

A Commentary on Confirmation and refinement of an autosomal dominant congenital motor nystagmus locus in chromosome 1q31.3–q32.1

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Nystagmus is involuntary rhythmic movements of the eyes.[1] It is usually pendular or jerky movements of both eyes. The presence of this eye movement reflects an imbalance in the systems that maintain stability of gaze. From functional point of view, the nystagmus could be caused by abnormalities in the steady gaze of the primary position, the system for holding eccentric gaze or the vestibulo-ocular reflex that maintains foveal fixation of images against the head movements.[1] Nystagmus can be caused by various reasons acquired during life. Nystagmus in early onset before 6 month of birth is classified into three: one of which is congenital idiopathic (or motor) nystagmus characterized by the absence of concomitant visual or neurological impairment.[2] In this issue of *Journal of Human Genetics*, Lin *et al.*[3] describe one form of congenital motor nystagmus maps to the human chromosome 1q31.1–q32.1. They have collected a large three generation kindred with the nystagmus presenting from birth. By using a set of microsatellite DNA markers, they have found that their family maps to a 24.6-cM region in 1q25.2–q32.1 limited by markers D1S218 and D1S2655. The physical distance of this region was estimated to be 28.06 Mb.

Fortunately, they had previously mapped the first autosomal dominant congenital motor nystagmus family to an 11.39-Mb region in 1q31.1 and q32.2.[4] This led them to narrow the most likely interval down to a 5.92-Mb region between D1S2816 and D1S2655. There are at least eight forms of inherited congenital motor nystagmus. Discovery of a gene causing the 1q31.1–q32.2-linked form of dominantly inherited congenital nystagmus would open a new avenue for our understanding of the eye movement controls.

1 Ropper, A. H. & Samuels, M. A. (eds). *Adams and Victor's Principles of Neurology.* 9th edn (McGraw Hill, New York, 2009).
2 Casteels, I., Harris, C. M., Shawkat, F. & Taylor, D. Nystagmus in infancy. *Br. J Ophthalmol.* **76,** 434–437 (1992).
3 Li, L., Xiao, X., Yi, C., Jiao, X., Guo, X., Hejtmancik, J. F. *et al.* Confirmation and refinement of an autosomal dominant congenital motor nystagmus locus in chromosome 1q31.3–q32.1. *J. Hum. Genet.* **57,** 756–759 (2012).
4 Xiao, X., Li, S., Guo, X. & Zhang, Q. A novel locus for autosomal dominant congenital motor nystagmus mapped to 1q31–q32.2 between D1S2816 and D1S2692. *Hum. Genet.* **131,** 697–702 (2012).

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