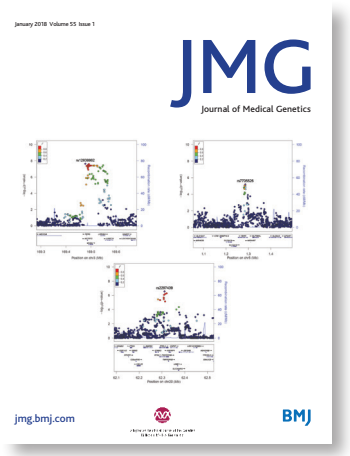
Journal of Medical Genetics



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January 2018 Volume 55 Issue 1