

OPTICAL GENOME MAPPING

Optical Genome Mapping (OGM) is a powerful technique capable of resolving almost all genomic aberrations. Two papers, both of which appeared in the Am. J. Hum. Genet., explore this technology in [constitutional cases](#)¹ and in [hematological malignancies](#)². Their conclusion is that OGM is very efficient and accurate. The only exceptions are rearrangements in which one of the breakpoints is in the (peri) centromeric region; OGM is unable to handle the repetitive nature of centromeres. A further limitation was that the authors, at least under their experimental design, could not detect mosaic aberrations below ~ 10%.

¹ <https://www.sciencedirect.com/science/article/abs/pii/S0002929721002172?via%3Dihub>

² <https://www.sciencedirect.com/science/article/abs/pii/S0002929721002238?via%3Dihub>