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CONCLUSION

This case report highlights the efficacy of NGS to detect novel variants in the BRCA1/2 genes. NGS approach is an efficient, rapid, cost-effective strategy for the simultaneous detection of all types of BRCA1/2 mutations, avoiding the usual time consuming multistep approach in the routine diagnostic testing of HBOC.

REFERENCE

 Petrucelli N, Daly MB, Pal T. BRCA1- and BRCA2-Associated Hereditary Breast and Ovarian Cancer. EditorsIn: Adam MP, Ardinger HH, Pagon RA, Wallace SE, Bean LJH, Stephens K, Amemiya A, editors. SourceGeneReviews® [Internet]. Seattle (WA): University of Washington, Seattle; 1993-2019. 1998 Sep 4 [updated 2016 Dec 15].