

## RECOMMENDATIONS

- Genetic counselling is recommended for the patient and adult at-risk relatives
- Genetic testing for the identified variant in affected relative(s) (if any) and adult at-risk relatives is recommended
- A consultation with an oncologist is recommended to discuss about surveillance measures and management options
- In patients with BRCA1 germline mutations (pathogenic/likely pathogenic variant), Olaparib, a PARP inhibitor, has been approved by FDA for the below indications;
  - *Advanced ovarian cancer, who have been treated with three or more prior lines of chemotherapy*
  - *HER2-negative metastatic breast cancer, who have been treated with chemotherapy*
  - *Triple negative breast cancer patients*

## INTERPRETATION

- Individuals with BRCA1 germline pathogenic variant are at an increased risk of hereditary breast and ovarian cancer (HBOC), which is inherited in an autosomal dominant manner
- BRCA1-associated HBOC is characterized by an increased life time risk for female breast cancer (46%-87%) and ovarian cancer (39%-63%), and to a lesser extent other cancers such as prostate cancer, pancreatic cancer, male breast cancer and melanoma [PMID: 20301425]
- Molecular analysis of BRCA1/2 genes is essential for familial breast and ovarian cancer prevention and treatment. A shorter turn around time of test results is crucial when patients with breast and/or ovarian cancer are addressed for appropriate therapy guidelines and their unaffected family members for surveillance and due management