

## GHSNZ Hereditary Genetic Testing Guidelines: Ovarian Cancer

Version 4.0 Last reviewed 11/12/2023

Note: these guidelines are for Oncology Mainstreamed testing \* and GHSNZ use

Individuals with high grade non-mucinous epithelial ovarian, or primary peritoneal cancer at any age (includes individuals with high-grade clear cell or endometrioid ovarian cancer)

**BRCA1, BRCA2, BRIP1, PALB2<sup>1</sup>, RAD51C, RAD51D<sup>2</sup>**

P/LP variant, or variant of uncertain significance (VUS) detected

No P/LP variant or VUS detected

- Personal history of bowel cancer or FDR with bowel cancer<sup>3</sup>
- Ovarian cancer (CC + EC)<sup>3</sup>
- Amsterdam positive Fhx<sup>4</sup>
- Personal or Fhx of breast cancer under 30<sup>5</sup>

Referral to Genetic Health Service NZ NOT required

No

Yes

### Refer to Genetic Health Service NZ 6

#### Northern Hub

Fax: (09) 307 4978 Email: GenSec@adhb.govt.nz

#### Central Hub

Fax: (04) 385 5822 Email: genetic.services@ccdhb.org.nz

#### South Island Hub

Email: genetic.servicenz@cdhb.health.nz

GHSNZ – refer to GHSNZ Hereditary Breast/Colorectal Cancer testing guidelines.

GHSNZ = Genetic Health Service New Zealand

P/LP/VUS = Pathogenic/likely pathogenic/variant of uncertain significance

MMR = Mismatch repair genes

dMMR = Deficient MMR (absent staining on IHC test)

IHC = Immunohistochemistry

Fhx = Family history

LFS = Li-Fraumeni Syndrome

FAP = Familial Adenomatous Polyposis

FDR = First degree relative

CC = clear cell ovarian cancer

EC = endometrioid ovarian cancer

\* Post-completion of oncology in-service genetics education

1. Truncating variants

2. GHSNZ – add MMR genes if IHC unavailable or dMMR

3. Arrange IHC (MMR). Only refer if dMMR. For loss of MLH1 expression only, request MLH1 promoter methylation studies, and only refer if methylation absent

4. Amsterdam positive Fhx = (At least 3 relatives with colorectal cancer, 1 of whom is a first degree relative of the other (FAP excluded), with at least 2 successive generations involved and at least 1 of the cancers diagnosed before age 50)

5. Consider LFS. Significant family history of breast / other cancer types – advice available from GHSNZ

6. Please provide detailed family history information