



Name : Visit No. :202309236  
Patient ID : Gender/Age : F /56 yrs Visit DateTime :30/08/2023 00:00  
Local Patient No : Ref No : L36834 Sample Type :PB  
Physician Name : ORPHANOS GEORGE Collected Date Time :30/08/2023 00:00  
Physician Email : Physician Fax :  
Hospital/Clinic/Lab GERMAN ONCOLOGY CENTRE Hospital Fax :25208006

## MOLECULAR CANCER PATHOLOGY AND GENETICS

| Test Description  | Result   | Comments / Reference   |
|---|--|--|
| <b>12613 - Hereditary Cancer Investigation (NGS+Digital MLPA)</b> |  |  |
| Reason for Referral   | Hereditary cancer predisposition investigation |  |
| D001-Digital MLPA Cancer Panel                                    | No copy number aberrations detected            | D001 Digital MLPA cancer Panel 1 includes a total number of 690 probes for the detection of copy number alterations in 30 genes involved in hereditary cancer. |
| RESULTS SUMMARY   | NO PATHOGENIC VARIANTS DETECTED                |  |

### REMARKS

#### CLINICAL INFORMATION

Strong family history of cancer.

#### TEST RESULTS AND INTERPRETATION

#### VARIANTS OF CLINICAL SIGNIFICANCE

No clinically relevant variants have been detected in the genes tested.

#### RECOMMENDATIONS - FURTHER TESTING ADVICE

It is possible that this patient has a pathogenic variant outside of the genetic regions or genes analysed. Clinical exome sequencing or high-resolution CGH array with LOH analysis may be able to determine the presence of pathogenic variants that could contribute to the patient's phenotype.

#### DIGITAL MLPA

NGS based digital MLPA analysis is utilized to detect CNVs in 30 genes associated with hereditary predisposition to breast, ovarian, colorectal, gastric, prostate, pancreatic, endometrial cancer or melanoma. Target genes included in Digital MLPA: APC, ATM, BAP1, BARD1, BMPR1A, BRCA1, BRCA2, BRIP1, CDH1, CDK4, CDKN2A, CHEK2, EPCAM, GREM1, MTF, MLH1, MSH2, MSH6, MUTYH, NBN, PALB2, PMS2, POLE, PTEN, RAD51C, RAD51D, SMAD4, STK11, TP53.

Authorised By

Paul Costello (FRCR)  
Director

Approved Date