

Supplementary Table 1A. Germline aberrations currently in clinical trials

| Clinical trial ID | Condition | Germline query | Context |
|-------------------|---|--|--|
| NCT02032823 | Breast cancer | gBRCA_del_mt | Olaparib |
| NCT02184195 | Pancreatic cancer | gBRCA_del_mt | Olaparib |
| NCT02476968 | Ovarian cancer | gBRCA_del_mt | Olaparib |
| NCT02855697 | High-grade serous/endometrioid ovarian cancer (HGS/EOC) | gBRCA_del_mt | Olaparib |
| NCT02693535 | Lymphoma, non-Hodgkin | gBRCA_del_mt/ gATM | Olaparib |
| NCT02489006 | Ovarian cancer, fallopian tube cancer | gBRCA_del_mt1/2, RAD51B, RAD51C, RAD51D, PPM1D, FANCM, BRIP1, PALB2, and BARD1 | Olaparib |
| NCT02789332 | Breast cancer | gBRCA_del_mt/ homologous recombination deficiency (HRD) genes | Olaparib and paclitaxel |
| NCT01945775 | Breast neoplasms | gBRCA_del_mt | Talazoparib |
| NCT01905592 | Carcinoma of breast | gBRCA_del_mt | Niraparib |
| NCT02505048 | Breast cancer | gBRCA_del_mt WT | Rucaparib |
| NCT02157792 | Advanced solid tumor, breast cancer | gBRCA_del_mt WT | VX-970 |
| NCT02608346 | Women with gBRCA_del_mt1 mutations | gBRCA_del_mt | Circulating DNA |
| NCT01670500 | Breast cancer | gBRCA_del_mt | Cisplatin |
| NCT02282020 | Ovarian cancer | gBRCA_del_mt | Cisplatin |
| NCT01982448 | Triple negative breast cancer | gBRCA_del_mt WT/ HRD assay | Cisplatin |
| NCT01525589 | Breast cancer | gBRCA_del_mt | PM01183 |
| NCT00496288 | Breast cancer | gBRCA_del_mt | Prophylactic irradiation |
| NCT02053805 | Prostate cancer | gBRCA_del_mt (and Lynch) | Prostate cancer risk |
| NCT02557776 | Hereditary breast cancer | gBRCA_del_mt | Screening |
| NCT02497820 | Colorectal cancer | gMMR/Lynch syndrome genes | Chemoprevention by aspirin |
| NCT02813824 | Lynch syndrome patients with colorectal adenomas | gMMR/Lynch syndrome genes | Chemoprevention by aspirin |
| NCT02401347 | Breast cancer | HR pathway: PTEN, PALB2, CHEK2, ATM, NBN, BARD1, BRIP1, RAD50, RAD51C, RAD51D, MRE11, ATR, FANC group genes | Talazoparib |
| NCT02664389 | Breast cancer | cancer predisposition gene panel | Early onset cancers with sporadic or hereditary presentation |
| NCT02759835 | Lung adenocarcinoma | gEGFR T790M | Local ablative therapy after treatment with osimertinib |
| NCT02282917 | Vestibular schwannoma | gNF2 | AR-42 histone deacetylase inhibitor |
| NCT02823652 | Advanced malignant neoplasm | germline tests | Genetic counselling |
| NCT02019693 | Papillary renal cell carcinoma | gMET activating | Reference |
| NCT00579514 | Breast, bladder, kidney, colon, prostate, and lung cancer incidence | PCR/PCR/LDR: polymorphisms in BRCA1, BRCA2, PTEN, TGFβ1, TGFβ-1, ATM, CHK2, APC, ER, PR, MCP-1, MPIF, CCR2/5 and CCR3, cytokine signaling, and apoptosis regulatory pathways | Cancer incidence |
| NCT01003522 | Non-small cell lung cancer | whole genome | Lung cancer |
| NCT01372696 | Colorectal cancer | whole genome | Colorectal cancer |
| NCT02285179 | Breast cancer | whole genome | Pharmacogenetics |
| NCT02747888 | Thyroid cancer | whole genome | Thyroid cancer risk |
| NCT01855477 | Solid tumors | background reference | - |
| NCT01904916 | Solid tumors | background reference | - |
| NCT01946100 | Lung neoplasms | background reference | - |
| NCT02695459 | Neuroendocrine carcinomas | background reference | - |