**Supplementary tables:**

Table S1: Highly predisposed families selection criteria. yo = years old.

|  |  |  |  |  |
| --- | --- | --- | --- | --- |
|  | **Number of generation** | **Degrees** | **Number of cancers\*** | **Type and age of cancers**  at least: |
| **a** | ≥ 2 generations | First-degree or second-degree through a male relative | 2 cases | * 1 triple negative breast cancer ≤ 40 yo |
| **b** | 2 cases | * 2 ovarian cancers with at least one ovarian cancer ≤ 50 yo |
| **c** | 3 cases | * 1 breast cancer ≤ 40 yo or * 1 ovarian cancer ≤ 50 yo |
| **d** | 4 cases | * 1 breast cancer ≤ 50 yo or * 1 ovarian cancer ≤ 60 yo |
| **e** | 5 cases | * 1 breast or ovarian cancer without age criteria |

\* Number of cancers in the restricted HBOC spectrum (breast, ovarian)

Table S2: Custom panel gene list with associated reference transcript NMs. bp = base pairs.

|  |  |  |  |
| --- | --- | --- | --- |
| **Genes** | **Number of probes** | **Reference transcript NMs** | **Transcript lengths** |
| *ATM* | 12 | NM\_000051.4 | 12,915 bp |
| *BAP1* | 8 | NM\_004656.4 | 3,600 bp |
| *BRCA1* | 10 | NM\_007294.4 | 7,088 bp |
| *BRCA2* | 13 | NM\_000059.3 | 11,954 bp |
| *BRIP1* | 14 | NM\_032043.3 | 8,182 bp |
| *CDH1* | 16 | NM\_004360.5 | 4,811 bp |
| *CHEK2* | 20 | NM\_007194.4 | 1,844 bp |
| *MLH1* | 12 | NM\_000249.4 | 2,494 bp |
| *MLH3* | 14 | NM\_001040108.2 | 7,820 bp |
| *MRE11* | 17 | NM\_005591.4 | 6,841 bp |
| *MSH2* | 20 | NM\_000251.3 | 3,115 bp |
| *MSH6* | 12 | NM\_000179.3 | 4,265 bp |
| *MUTYH* | 11 | NM\_001048171.2 | 1,858 bp |
| *NBN* | 15 | NM\_002485.5 | 4,622 bp |
| *PALB2* | 10 | NM\_024675.4 | 4,008 bp |
| *PMS1* | 14 | NM\_000534.5 | 3,156 bp |
| *PMS2* | 12 | NM\_000535.7 | 5,093 bp |
| *PTEN* | 13 | NM\_000314.8 | 8,515 bp |
| *RAD50* | 21 | NM\_005732.4 | 8,722 bp |
| *RAD51* | 10 | NM\_001164269.2 | 2,335 bp |
| *RAD51B* | 11 | NM\_133509.5 | 2,679 bp |
| *RAD51C* | 16 | NM\_058216.3 | 2,562 bp |
| *RAD51D* | 12 | NM\_002878.4 | 9,966 bp |
| *RINT1* | 15 | NM\_021930.6 | 2,860 bp |
| *STK11* | 14 | NM\_000455.5 | 3,293 bp |
| *TP53* | 11 | NM\_000546.6 | 2,512 bp |
| *XRCC2* | 5 | NM\_005431.2 | 4,771 bp |
| *XRCC3* | 9 | NM\_001100119.2 | 2,604 bp |

Table S3: Primer pair designations and sequences. ex = exon, F = forward, R = reverse, bp = base pairs.

|  |  |  |  |
| --- | --- | --- | --- |
| **Primer pair designations** | **Primer localizations** | **Sequences** | **Product lengths** |
| 1 | BRCA1\_ex14\_F | GTTGTTGATGTGGAGGAGCA | 433 bp |
| BRCA1\_ex16\_R | TGATGTGGTGTTTTCTGGCA |
| 2 | BRCA1\_ex5\_F | TACGAGATTTAGTCAACTTG | 420 bp |
| BRCA1\_ex9\_R | ATTTCATCCCTGGTTCCTTG |
| 3 | BRCA1\_ex12\_F | AGCCAGCCTTCTAACAGCTA | 239 bp |
| BRCA1\_ex14\_R | GGGCATTTAGAAGGGGATGAC |
| 4 | BRCA1\_ex10\_R | GACATTAAGGAAAGTTCTGCT | 908 bp |
| BRCA1\_ex13\_R | CCTTCTGGATTCTGGCTTATAGGG |
| 5 | SVA\_F | CTGTCTGGGATGTGAGGA | 119 bp |
| BRCA1\_ex13\_R | AAGGCCTTCTGGATTCTGG |
| 6 | BRCA1\_ex12\_F | CCTTGAGGACCTGCGAAA | 2,018 bp |
| SVA\_R | CTCCCCACATCTCAGACGAT |