

## CLAIMS

1. A process for detecting a chromosome state which represents a subgroup in a population comprising determining whether a chromosome interaction relating to that chromosome state is present or absent  
5 within a defined disease-associated region of the genome, wherein said disease is breast cancer, and wherein a specific combination of chromosome interactions are typed comprising at least 4, 5, 6 or 7 of the chromosome interactions represented by the following probes (Marker Set 3):
- TTGGAGGGAAAAGTAATTACGTTCAACTTCGACTGTATTCTACAAAGTGCTGGGATTACA;
  - GGAGGAGGCCAGAGAAAAGAGAATGTGATCGATTCTAAAATACTGTGTGTGTGTATGTA;
  - 10 - GGAGGAGGCCAGAGAAAAGAGAATGTGATCGATGACCTTAATGTCAGTGCTACTGACTCT;
  - CAGAAATGCACGCACAGGCACAACAGCATCGAAACCGTTCTTTGGAGGCTCAGTTTTTG;
  - CCAAAGACAGCCAAGGAAAACTAAAGATCGAAAGTTTTTATTACTTCCAAATTAGTAAA;
  - AGGATCTCATGATGCTTTGAATACTTTCTCGATACCTTATTATAAAATCAGCTTTGTGTT;
  - CATAATTTTTTTTTGTAGTTTATTCACCTCGACTAGATTTTAATTTTAATTTTATTTA;
  - 15 - CCTACATATACATGGAATATTCCTGGTATCGAAATATTTAGGTAATCATTATTTGTCTA;
  - ATTTCTTTCTTCTTCCATTTTCTAAAATCGATTTTAAATTAAAGGTACAAGTTAAGGC;
  - ATACTCATCATAAATGTCAGATTTATAATCGAGATCACAGTGAGCTGAGATTGCACCACT;
  - AGCTCAAATTCTTTTACTAATTGTTACATCGAAAGTTCAAATTTAAATTTTAAACGTTTT;
  - GGATGGAGGAAGAGGAGGAATTCAAGACTCGAACTAAACAAAAAGGAGATGATCCTGGGT;
  - 20 - AATTTAGAGGAAGTCTATATAAACAACATCGAACTTTGCTTCATGCACAAAATTTAAAA.
2. A process according to claim 1 comprising typing all of the chromosome interactions represented by all of the said probes of Marker Set 3.
3. A process according to claim 1 or 2 comprising typing all or at least 4, 5, 6 or 7 of the chromosome interactions represented by the following probes (Marker Set 1):
- ATTTCTTTCTTCTTCCATTTTCTAAAATCGATTTTAAATTAAAGGTACAAGTTAAGGC;
  - GGATGGAGGAAGAGGAGGAATTCAAGACTCGAACTAAACAAAAAGGAGATGATCCTGGGT;
  - AGCTCAAATTCTTTTACTAATTGTTACATCGAAAGTTCAAATTTAAATTTTAAACGTTTT;
  - 30 - CCAAAGACAGCCAAGGAAAACTAAAGATCGAAAGTTTTTATTACTTCCAAATTAGTAAA;
  - AATTTAGAGGAAGTCTATATAAACAACATCGAACTTTGCTTCATGCACAAAATTTAAAA;
  - TTGGAGGGAAAAGTAATTACGTTCAACTTCGACTGTATTCTACAAAGTGCTGGGATTACA;
  - ATACTCATCATAAATGTCAGATTTATAATCGAGATCACAGTGAGCTGAGATTGCACCACT;
  - AGGATCTCATGATGCTTTGAATACTTTCTCGATACCTTATTATAAAATCAGCTTTGTGTT.

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4. A process according to any one of the preceding claims comprising typing all or at least 4, 5, 6 or 7 of the chromosome interactions represented by the following probes (Marker Set 2):
- TTGGAGGGAAAAGTAATTACGTTCAACTTCGACTGTATTCTACAAAGTGCTGGGATTACA;
  - GGAGGAGGCCAGAGAAAAGAGAATGTGATCGATTCTAAAATACTGTGTGTGTGTATGTA;
  - 5 - GGATGGAGGAAGAGGAGGAATTCAAGACTCGAACTAAACAAAAAGGAGATGATCCTGGGT;
  - AGGATCTCATGATGCTTTGAATACTTTCTCGATACCTTATTATAAAATCAGCTTTGTGTT;
  - AGCTCAAATTCTTTTACTAATTGTTACATCGAAAGTTCAAATTAATTTTAAACGTTTT;
  - AATTTAGAGGAACTCTATATAAACACATCGAACTTTGCTTCATGCACAAAATTTAAAA;
  - CAGAAATGCACGCACAGGCACAACAGCATCGAAACCGTTCTTTGGAGGCTCAGTTTTTG;
  - 10 - CCTACATATACATGGAATATTCTGGTATCGAAATATTTAGGTAATCATTATTTGTCTA.
5. A process according to any one of the preceding claims in which the chromosome interactions are typed by detecting the presence or absence of a DNA loop at the site of the chromosome interactions.
- 15 6. A process according to any one of the preceding claims in which the chromosome interactions are typed by detecting the presence or absence of distal regions of a chromosome being brought together in a chromosome conformation.
7. A process according to any one of the preceding claims in which the chromosome interactions are typed by detecting the presence of a ligated nucleic acid which is generated during said typing and whose sequence comprises two regions each corresponding to the regions of the chromosome which come together in the chromosome interaction.
- 20 8. A process according to claim 7 wherein detection of the ligated nucleic acid is performed by PCR.
- 25 9. A process according to claim 8 wherein said PCR uses at least one of the following primers:
- TGTAGTAGTTACCCTGTTGTTG;
  - GCCTCACAGTGCTCTTATG;
  - GTGCTTTGTAAACCATGAAGTG;
  - 30 - TCGTGGGCATATGACTGAG;
  - GGCATTGCTTTGCCTTATC;
  - CAACTTCCTTGGGTGTAGAG;
  - CGCTATATGTGGTTCTGTACG;
  - CTTCTCTAAAGGGAGATTTGGG;
  - 35 - TGTTGAGCAAGATGGATAGC;
  - ATATTCAGGATGGAACCCAAG;

- TCCAGAGGTTATGGAATTTGAG;
- AAGAAACAGACTGGGCTTG;
- ACTCAAATACTGCTCTACACTG;
- AAGGAAGTTAAGCCCTATGC;
- 5 - ACCCTCCTTCACTCACATAG;
- GCACCTAATCTACCTAACATCAC.

10. A process according to any one of the preceding claims, wherein the process is carried out to diagnose breast cancer.

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11. A process according to any one of the preceding claims, wherein the process is carried out to select an individual for a medical treatment.

12. A process according to any one of the preceding claims, wherein the process is carried out on an individual who has been selected based on gender or another physical characteristic.

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13. A process according to any one of the preceding claims, wherein the process is carried out to detect malignant breast cancer.

14. A process according to any of the preceding claims wherein the typing or detecting comprises specific detection of the ligated product by quantitative PCR (qPCR) which uses primers capable of amplifying the ligated product and a probe which binds the ligation site during the PCR reaction, wherein said probe comprises sequence which is complementary to sequence from each of the chromosome regions that have come together in the chromosome interaction.

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15. A process according to claim 14 wherein said probe comprises an oligonucleotide which specifically binds to said ligated product.

16. A process according to claim 14 or 15 wherein said probe comprises a quencher covalently attached to the 3' end of the oligonucleotide.

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17. A process according to any one of claims 14 to 16 wherein said probe comprises a fluorophore covalently attached to the 5' end of the oligonucleotide.

18. A process according to claim 17 wherein said fluorophore is selected from HEX, Texas Red and FAM.

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19. A process according to any one of claims 14 to 18 wherein said probe comprises a nucleic acid sequence of length 10 to 40 nucleotide bases.

20. A process according to claim 20 wherein said probe comprises a nucleic acid sequence of length 20 to  
5 30 nucleotide bases.