

CLAIMS

1. A method for assessing the risk of a human female subject for developing breast cancer comprising:
 - 5 performing a clinical risk assessment of the female subject;
 - performing a genetic risk assessment of the female subject, wherein the genetic risk assessment involves detecting, in a biological sample derived from the female subject, at least 72 single nucleotide polymorphisms associated with breast cancer, wherein at least 67 of the single nucleotide polymorphisms are selected from Table 7,
 - 10 or a single nucleotide polymorphism in linkage disequilibrium with one or more thereof, and the remaining single nucleotide polymorphisms are selected from Table 6, or a single nucleotide polymorphism in linkage disequilibrium with one or more thereof, wherein the single nucleotide polymorphism(s) in linkage disequilibrium has linkage disequilibrium above 0.9; and
 - 15 combining the clinical risk assessment with the genetic risk assessment to obtain the risk of a human female subject for developing breast cancer.
2. The method of claim 1, wherein the female is Caucasian.
- 20 3. The method of claim 2, which comprises detecting at least 72 single nucleotide polymorphisms shown in Table 9, or a single nucleotide polymorphism in linkage disequilibrium with one or more thereof, wherein the single nucleotide polymorphism(s) in linkage disequilibrium has linkage disequilibrium above 0.9.
- 25 4. The method of claim 1, wherein the female is Negroid.
5. The method of claim 4, which comprises detecting at least 74 single nucleotide polymorphisms shown in Table 10, or a single nucleotide polymorphism in linkage disequilibrium with one or more thereof, wherein the single nucleotide
- 30 polymorphism(s) in linkage disequilibrium has linkage disequilibrium above 0.9.
6. The method of claim 1, wherein the female is Hispanic.
7. The method of claim 6, which comprises detecting at least 78 single nucleotide
- 35 polymorphisms shown in Table 11, or a single nucleotide polymorphism in linkage

disequilibrium with one or more thereof, wherein the single nucleotide polymorphism(s) in linkage disequilibrium has linkage disequilibrium above 0.9.

8. The method according to any one of claims 1 to 7, wherein combining the clinical risk assessment with the genetic risk assessment comprises multiplying the risk assessments to provide the risk score.

9. The method according to any one of claims 1 to 8, wherein performing the clinical risk assessment uses a model selected from a group consisting of the Gail Model, the Claus Model, Claus Tables, BOADICEA, the Jonker Model, the Claus Extended Formula, the Tyrer-Cuzick Model, BRCAPRO, and the Manchester Scoring System.

10. The method according to any one of claims 1 to 9, wherein performing the clinical risk assessment includes obtaining information from the female on one or more of the following: medical history of breast cancer, ductal carcinoma or lobular carcinoma, age, age of first menstrual period, age at which she first gave birth, family history of breast cancer, results of previous breast biopsies, breast density, and race/ethnicity.

11. The method according to any one of claims 1 to 10, wherein the results of the clinical risk assessment indicate that the female should be subjected to more frequent screening and/or prophylactic anti-breast cancer therapy.

12. The method according to any one of claims 1 to 11, wherein if it is determined the subject has a risk of developing breast cancer, the subject is more likely to be responsive to oestrogen inhibition therapy than non-responsive.

13. The method of any one of claims 1 to 12, wherein the breast cancer is estrogen receptive positive or estrogen receptor negative.

14. The method of any one of claims 1 to 13, wherein the single nucleotide polymorphism in linkage disequilibrium has linkage disequilibrium of 1.

15. A kit when used for assessing risk of developing breast cancer according to the method of any one of claims 1 to 14, the kit comprising at least 72 sets of primers for

- amplifying 72 or more nucleic acids, wherein the 72 or more nucleic acids comprise a single nucleotide polymorphism, wherein at least 67 of the sets of primers amplify nucleic acids comprising a single nucleotide polymorphism selected from Table 7, or a single nucleotide polymorphism in linkage disequilibrium with one or more thereof, and the remaining sets of primers amplify nucleic acids comprising a single nucleotide polymorphism selected from Table 6, or a single nucleotide polymorphism in linkage disequilibrium with one or more thereof, wherein the single nucleotide polymorphism(s) in linkage disequilibrium has linkage disequilibrium above 0.9.
16. A genetic array when used for assessing risk of developing breast cancer according to the method of any one of claims 1 to 14, the genetic array comprising at least 72 sets of probes for hybridising to 72 or more nucleic acids, wherein the 72 or more nucleic acids comprise a single nucleotide polymorphism, wherein at least 67 of the probes hybridise to nucleic acids comprising a single nucleotide polymorphism selected from Table 7, or a single nucleotide polymorphism in linkage disequilibrium with one or more thereof, and the remaining probes hybridise to nucleic acids comprising a single nucleotide polymorphism selected from Table 6, or a single nucleotide polymorphism in linkage disequilibrium with one or more thereof, wherein the single nucleotide polymorphism(s) in linkage disequilibrium has linkage disequilibrium above 0.9.
17. A method for determining the need for routine diagnostic testing of a human female subject for breast cancer comprising assessing the risk of the subject for developing breast cancer using the method according to any one of claims 1 to 14.
18. A method of screening for breast cancer in a human female subject, the method comprising assessing the risk of the subject for developing breast cancer using the method according to any one of claims 1 to 14, and routinely screening for breast cancer in the subject if they are assessed as having a risk for developing breast cancer.
19. A method for determining the need of a human female subject for prophylactic anti-breast cancer therapy comprising assessing the risk of the subject for developing breast cancer using the method according to any one of claims 1 to 14.
20. A method for preventing breast cancer in a human female subject, the method comprising assessing the risk of the subject for developing breast cancer using the

method according to any one of claims 1 to 14, and an anti-breast cancer therapy is to be administered to the subject if they are assessed as having a risk for developing breast cancer.

5 21. An anti-breast cancer therapy when used for preventing breast cancer in a human female subject at risk thereof, wherein the subject is assessed as having a risk of developing breast cancer according to the method of any one of claims 1 to 14.

22. A method for stratifying a group of human female subjects for a clinical trial of
10 a candidate therapy, the method comprising assessing the individual risk of the subjects for developing breast cancer using the method according to any one of claims 1 to 14, and using the results of the assessment to select subjects more likely to be responsive to the therapy.

15 23. A computer implemented method for assessing the risk of a human female subject for developing breast cancer, the method operable in a computing system comprising a processor and a memory, the method comprising:

receiving clinical risk data and genetic risk data for the female subject, wherein the genetic risk data was obtained by detecting, in a biological sample derived from the
20 female subject, at least 72 single nucleotide polymorphisms associated with breast cancer, wherein at least 67 of the single nucleotide polymorphisms are selected from Table 7, or a single nucleotide polymorphism in linkage disequilibrium with one or more thereof, and the remaining single nucleotide polymorphisms are selected from Table 6, or a single nucleotide polymorphism in linkage disequilibrium with one or
25 more thereof, wherein the single nucleotide polymorphism(s) in linkage disequilibrium has linkage disequilibrium above 0.9;

processing the data to combine the clinical risk data with the genetic risk data to obtain the risk of a human female subject for developing breast cancer;

outputting the risk of a human female subject for developing breast cancer.

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24. A system for assessing the risk of a human female subject for developing breast cancer comprising:

system instructions for performing a clinical risk assessment of the female subject;

35 system instructions for performing a genetic risk assessment of the female subject according to any one of claims 1 to 14; and

system instructions for combining the clinical risk assessment with the genetic risk assessment to obtain the risk of a human female subject for developing breast cancer.