

CLAIMS

1. A method for assessing the risk of a human subject for developing colorectal cancer comprising:

performing a genetic risk assessment of the subject, wherein the genetic risk assessment involves detecting, in a biological sample derived from the subject, the presence of at least 28 single nucleotide polymorphisms selected from Table 1, or a single nucleotide polymorphism in linkage disequilibrium with one or more thereof, wherein at least three of the single nucleotide polymorphisms are rs3987, rs35509282 and rs744166, or a single nucleotide polymorphism in linkage disequilibrium with one or more thereof and, wherein the single nucleotide polymorphism in linkage disequilibrium has linkage disequilibrium above 0.7.

2. The method of claim 1, wherein the presence of at least 45 single nucleotide polymorphisms are detected.

3. The method of claim 1 or claim 2, wherein the genetic risk assessment comprises detecting the presence of single nucleotide polymorphism rs5934683, or a single nucleotide polymorphism in linkage disequilibrium thereof, wherein the single nucleotide polymorphism in linkage disequilibrium has linkage disequilibrium above 0.7.

4. The method of any one of claims 1 to 3 which further comprises performing a clinical risk assessment of the subject and combining the genetic risk assessment with the clinical risk assessment to obtain the risk of a human subject for developing colorectal cancer.

5. The method of claim 4, wherein performing the clinical risk assessment involves obtaining information from the subject on one or more of the following: medical history of colorectal cancer, age, family history of colorectal cancer, results of previous colonoscopy or sigmoidoscopy screening and race/ethnicity.

6. The method of claim 5, wherein performing the clinical risk assessment involves obtaining information from the subject on age and/or first degree relatives history of colorectal cancer.

7. The method of any one of claims 1 to 6, wherein the subject has had a positive fecal occult blood test.
8. The method of any one of claims 1 to 7, wherein the subject is at least 40 years old.
9. The method of any one of claims 1 to 7, wherein the subject has a family history of colorectal cancer and is at least 30 years of age.
10. The method of any one of claims 1 to 9, wherein the subject is male.
11. The method of any one of claims 1 to 10, wherein the results of the risk assessment indicate that the subject should be enrolled in a screening program or subjected to more frequent screening.
12. The method of any one of claims 1 to 11, wherein the method performance is characterized by an area under the curve (AUC) of at least about 0.63.
13. The method of any one of claims 1 to 12, wherein the single nucleotide polymorphism in linkage disequilibrium has linkage disequilibrium above 0.9.
14. The method of any one of claims 1 to 12, wherein the single nucleotide polymorphism in linkage disequilibrium has linkage disequilibrium of 1.
15. A method for determining the need for routine diagnostic testing of a human subject for colorectal cancer comprising assessing the risk of the subject for developing colorectal cancer using the method of any one of claims 1 to 14.
16. The method of claim 15, wherein, when factoring in that each of the single nucleotide polymorphisms may be present up to twice in the somatic diploid genome of the subject, a subject having at least 41, at least 42, at least 44, at least 46, at least 50, at least 55, at least 60, at least 65, or at least 70, of the single nucleotide polymorphisms should be enrolled in a fecal occult screening, colonoscopic or sigmoidoscopic screening program.

17. The method of claim 15, wherein if the assessment places the subject in the top 20% of subjects in a population at risk of developing colorectal cancer the subject is enrolled in a fecal occult screening, colonoscopic or sigmoidoscopic screening program.
18. The method of claim 15, wherein if the assessment places the subject in the top 10% of subjects in a population at risk of developing colorectal cancer the subject is enrolled in a fecal occult screening, colonoscopic or sigmoidoscopic screening program.
19. A method of screening for colorectal cancer in a human subject, the method comprising assessing the risk of the subject for developing colorectal cancer using the method of any one of claims 1 to 14, and routinely screening for colorectal cancer in the subject if they are assessed as having a risk for developing colorectal cancer.
20. An anti-colorectal cancer therapy for use in preventing colorectal cancer in a human subject at risk thereof, wherein the subject is assessed as having a risk for developing colorectal cancer according to the method of any one of claims 1 to 14.
21. A kit when used for performing the method of any one of claims 1 to 19, the kit comprising at least 28 sets of primers for amplifying 28 or more nucleic acids, wherein the 28 or more nucleic acids comprise a single nucleotide polymorphism selected from Table 1, or a single nucleotide polymorphism in linkage disequilibrium with one or more thereof, wherein the single nucleotide polymorphism in linkage disequilibrium has linkage disequilibrium above 0.7.
22. A genetic array when used for performing the method of any one of claims 1 to 19, the genetic array comprising at least 28 sets of probes for hybridising to 28 or more nucleic acids, wherein the 28 or more nucleic acids comprise a single nucleotide polymorphism selected from Table 1, or a single nucleotide polymorphism in linkage disequilibrium with one or more thereof, wherein the single nucleotide polymorphism in linkage disequilibrium has linkage disequilibrium above 0.7.
23. A computer implemented method for assessing the risk of a human subject for developing colorectal cancer, the method operable in a computing system comprising a processor and a memory, the method comprising:

receiving genetic risk data for the subject, wherein the genetic risk data was obtained by detecting, in a biological sample derived from the subject, the presence of at least 28 single nucleotide polymorphisms from Table 1, or a single nucleotide polymorphism in linkage disequilibrium with one or more thereof, wherein the single nucleotide polymorphism in linkage disequilibrium has linkage disequilibrium above 0.7;

processing the data to determine the risk of the human subject for developing colorectal cancer;

outputting the risk of the human subject for developing colorectal cancer.

24. The method of claim 23, further comprising, receiving clinical risk data for the subject;

processing the data to combine the clinical risk data with the genetic risk data to obtain the risk of the subject for developing colorectal cancer;

outputting the risk of the subject for developing colorectal cancer.

25. The computer implemented method of claim 23 or 24, wherein the risk data for the subject is received from a user interface coupled to the computing system.

26. The computer implemented method of any one of claims 23 to 25, wherein the risk data for the subject is received from a remote device across a wireless communications network.

27. The computer implemented method of any one of claims 23 to 26, wherein the user interface or remote device is a SNP array platform.

28. The computer implemented method of any one of claims 23 to 27, wherein outputting comprises outputting information to a user interface coupled to the computing system.

29. The computer implemented method of any one of claims 23 to 28, wherein outputting comprises transmitting information to a remote device across a wireless communications network.