

CLAIMS

1. A method of identifying risk of breast cancer in a human subject, the method comprising:
 - 5 determining in a tumour tissue and / or blood sample of the subject, whether a copy number amplification of a region specific to human chromosome 1q21.3 is present,
wherein the presence of a copy number amplification of the region specific to human chromosome 1q21.3 represents an elevated risk of breast cancer in the subject,
wherein said risk of breast cancer is at least one of a risk of recurrence of breast cancer, a risk of metastasis of breast cancer, a risk of non-abatement of breast cancer and a risk of breast cancer-based mortality,
wherein the determining step comprises evaluating a copy number ratio
15 of at least one continuous genomic region located on human chromosome 1q21.3 in the subject to a reference continuous genomic region in the subject, wherein a copy number ratio exceeding a threshold value is indicative of a copy number amplification, and
wherein the step of evaluating a copy number ratio of at least one
20 continuous genomic region located on human chromosome 1q21.3 comprises evaluating a copy number ratio of a human TUFT1 gene, a human S100A7 gene and a human S100A8 gene.
2. The method of claim 1, wherein the threshold value is obtained by evaluating a
25 copy number ratio of the at least one continuous genomic region in a healthy subject to a reference continuous genomic region in the healthy subject.
3. The method of claim 1 or 2, wherein said evaluating a copy number ratio of the
30 at least one continuous genomic region comprises evaluating an average copy number ratio of at least two continuous genomic regions located on human chromosome 1q21.3 and wherein said evaluating an average copy number comprises:

obtaining a copy number ratio of each of the at least two continuous genomic regions; and

averaging the copy number ratios of the at least two continuous genomic regions to obtain an average copy number ratio.

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4. The method according to any one of the claims 1 to 3, wherein the threshold value is obtained by evaluating a mean copy number ratio of the at least one continuous genomic region located on human chromosome 1q21.3 based on two or more healthy reference subjects and wherein said evaluating a mean copy number ratio based on two or more healthy reference subjects comprises:

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evaluating a copy number ratio of the at least one continuous genomic region of a first healthy reference subject;

evaluating a copy number ratio of the at least one continuous genomic region of a second healthy reference subject;

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optionally evaluating a copy number ratio of the at least one continuous genomic region of one or more subsequent healthy reference subject to obtain one or more subsequent copy number ratios; and

adding the copy number ratios of the two or more healthy reference subjects; and

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computing a mean of the copy number ratios.

5. The method of claim 4, wherein the determining step comprises determining whether a copy number amplification is present in a cell free DNA.

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6. Use of a therapeutic agent capable of suppressing IRAK1, IRAK4, S100A7, S100A8 or S100A9 in the manufacture of a medicament for treating breast cancer in a subject, wherein the subject is determined to have in his/her tumour tissue and / or blood sample, a copy number amplification of a region specific to human chromosome 1q21.3,

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wherein the therapeutic agent comprises pacritinib or therapeutically effective analogs thereof.

7. The use of claim 6, wherein the subject was previously ineffectively treated for breast cancer by an earlier therapy, wherein the earlier therapy is different from a therapy using the therapeutic agent.
- 5 8. A kit for determining whether a copy number amplification of a region specific to human chromosome 1q21.3 is present in a tumour tissue and / or blood sample, the kit comprising:
oligonucleotides for hybridizing to a human TUFT1 gene, a human S100A7 gene and a human S100A8 gene.
- 10 9. The kit of claim 8, wherein the oligonucleotides comprise primers for amplifying the human TUFT1 gene, the human S100A7 gene and the human S100A8 gene.
- 15 10. The kit of claim 8 or claim 9, wherein the oligonucleotides comprise at least one of the sequences:
TTTTAATCAGAGGGTGAGGGTGAT (SEQ ID NO. 1);
GCTTCTCAATGTTGGAGGATACA (SEQ ID No. 2);
GTCAAGATTGAGGAGGAAGAAGC (SEQ ID No. 3);
20 TTCATAGATGGCTATGCCTCGG (SEQ ID No. 4);
GGTGTTCCTCCCACTAGCCA (SEQ ID No. 5);
CCCAGAGAGTGTATTGGCCC (SEQ ID No. 6);
TGCTATGTGGCCTTGGACAGATCACC (SEQ ID No. 7);
AGTTTAAAGATCTCAGAGAGAGCCGAGGCA (SEQ ID No. 8); and
25 CCTAGCGTATCACATGTGGACATGGACA (SEQ ID No. 9).
11. The kit of any one of claims 8-10, wherein the kit further comprises an oligonucleotide for hybridizing to a human RPP30 gene.
- 30 12. The kit of claim 11, wherein the oligonucleotide comprises a primer for amplifying the human RPP30 gene.
13. The kit of claim 11 or claim 12, wherein the oligonucleotide for hybridizing to a human RPP30 gene comprises at least one of the sequences:

GTTAGAGAGTCTCCAGGCCC (SEQ ID No. 10);
ACTGTAATCCAGCAAAAGCGG (SEQ ID No. 11); and
TGTCCACAGACTTTCTCAAAAGATAGGGCC (SEQ ID No. 12).

- 5 14. The method or the kit of any one of claims 1-5 or 8-13, wherein the method or the kit has a sensitivity of no less than 85% and a specificity of no less than 85% in detecting a copy number amplification of a region specific to human chromosome 1q21.3.