

CLAIMS

1. A method of testing a nucleic acid sample of a human, comprising:
testing a nucleic acid sample obtained from a human and determining presence of a somatic mutation in a promoter of a *telomerase reverse transcriptase (TERT)* gene, wherein the somatic mutation is located at chr5 1,295,228 ("228"), chr5 1,295,229 ("229"), or chr5 1,295,250 ("250"), and the somatic mutation is selected from the group consisting of C228T, C250T, C228A, and C229A, wherein the nucleic acid is from a cancer selected from the group consisting of: a myxoid liposarcoma, a fibrosarcoma, a myxofibrosarcoma, a chondrosarcoma, an osteosarcoma, a urothelial carcinoma of the upper urinary epithelium, a head and neck cancer, a medulloblastoma, a glioma, an astrocytoma, an oligodendrogliomas, and an oligoastrocytoma ; and further comprising a step selected from:
 - (a) diagnosing the cancer;
 - (b) identifying progression or recurrence of a cancer;
 - (c) characterizing the cancer as a subtype of tumor; and
 - (d) recommending a treatment or monitoring plan for the cancer.
2. The method of claim 1 wherein the cancer is a sarcoma.
3. The method of claim 1 wherein the cancer is a head and neck cancer.
4. The method of claim 1 wherein the cancer is a medulloblastoma.
5. The method of claim 1 wherein the cancer is a glioma.
6. The method of claim 2 wherein the cancer is a myxoid liposarcoma.
7. The method of claim 1 wherein the cancer is a urothelial carcinoma of the upper urinary tract.
8. The method of claim 3 wherein the cancer is a squamous cell carcinoma of the tongue.
9. The method of claim 5 wherein the cancer is a primary glioblastoma.
10. The method of claim 5 wherein the cancer is an astrocytoma.
11. The method of claim 5 wherein the cancer is an oligodendrogliomas.
12. The method of claim 5 wherein the cancer is an oligoastrocytoma.
13. The method of claim 1 wherein the nucleic acid sample is obtained from a primary tumor.

14. The method of claim 1 wherein the nucleic acid is tested by amplifying the promoter or part of the promoter to form an amplicon.
15. The method of claim 14 wherein the part of the promoter comprises at least one nucleotide selected from the group consisting of chr5 1,295,228 ("228"), chr5 1,295,229 ("229"), and chr5 1,295,250 ("250").
16. The method of claim 14 wherein the amplicon is sequenced.
17. The method of claim 14 wherein the amplicon is hybridized to a mutation specific oligonucleotide.
18. A nucleic acid probe comprising at least 18 contiguous nucleotides of a human TERT promoter at human chromosome 5 comprising positions 1,295,228 ("228"), chr5 1,295,229 ("229"), or chr5 1,295,250 ("250"), wherein the 18 contiguous nucleotides include mutations C228A or C229A.
19. A nucleic acid primer comprising at least 18 contiguous nucleotides of a human TERT promoter at human chromosome 5 comprising positions 1,295,228 ("228"), chr5 1,295,229 ("229"), or chr5 1,295,250 ("250"), wherein the 18 contiguous nucleotides include mutations C228A or C229A.
20. The probe of claim 18 which comprises a detectable label.
21. The primer of claim 19 which additionally comprises a sequence of at least 10 nucleotides which are not contiguous to the 18 contiguous nucleotides of the human TERT promoter.
22. The method of claim 1 wherein prior to the step of testing, nucleic acids are extracted from a primary tumor sample.

23. A method of testing a nucleic acid sample of a human, comprising:

testing a nucleic acid sample obtained from a human and determining presence of a somatic mutation in a promoter of a *telomerase reverse transcriptase (TERT)* gene, wherein the somatic mutation is located at chr5 1,295,228 ("228"), chr5 1,295,229 ("229"), or chr5 1,295,250 ("250"), and the somatic mutation is selected from the group consisting of C228T, C250T, C228A, and C229A, wherein the nucleic acid is from a cancer selected from the group consisting of: a myxoid liposarcoma, a fibrosarcoma, a myxofibrosarcoma, a chondrosarcoma, an osteosarcoma, a urothelial carcinoma of the upper urinary epithelium, a head and neck cancer, a medulloblastoma, a glioma, an astrocytoma, an oligodendroglioma, and an oligoastrocytoma.