

CLAIMS

1. A process for detecting the stage or presence of scleroderma;
 - wherein the stage of scleroderma is detected by determining the presence or absence of at least 10 chromosome interactions represented by any probe shown in:
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 - Table 1.a2 or Table 1.b2,
 - Table 3.a3 or Table 3.b3, or,
 - Table 4.a3 or Table 4.b3; or
 - wherein the the presence of scleroderma is detected by determining the presence or absence of at least 10 chromosome interactions represented by any probe shown in:
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 - Table 2.a2 or Table 2.b2,
 - Table 5.a2 or Table 5.b2, or
 - Table 6.a3 or Table 6.b3 .
2. A process according to claim 1 in which the presence or absence of the chromosome interactions is determined:
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 - in a sample from an individual, or
 - by detecting the presence or absence of a DNA loop at the site of the chromosome interactions, or
 - detecting the presence or absence of distal regions of a chromosome being brought together in a chromosome conformation.
- 20 3. A process according to claim 1 or 2 in which the presence or absence of each of the chromosome interactions is determined by detecting the presence of a ligated nucleic acid which is generated during said detecting and whose sequence comprises two regions each corresponding to the regions of the chromosome which come together in the chromosome interaction.
4. A process according to claim 3, in which detection of the ligated nucleic acid is by a probe with
 - 25 the sequence of any of the specific probe sequences shown in:
 - Table 1.a2 or Table 1.b2,
 - Table 2.a2 or Table 2.b2,
 - Table 3.a3 or Table 3.b3,
 - Table 4.a3 or Table 4.b3,

- Table 5.a2 or Table 5.b2, or

- Table 6.a3 or Table 6.b3.

5. A process according to any one of the preceding claims, wherein said determining of whether each of the chromosome interactions are present or absent is by a process comprising the steps of: -

5 (i) cross-linking of chromosome regions which have come together in the chromosome interaction;

(ii) subjecting the cross-linked regions to cleavage;

(iii) ligating the cross-linked cleaved DNA ends to form ligated DNA; and

10 (iv) detecting the presence or absence of the ligated DNA to thereby determine presence or absence of the chromosome interaction.

6. A process according to claim 5, wherein detecting the presence or absence of the ligated DNA is by quantitative PCR (qPCR) which uses primers capable of amplifying the ligated DNA 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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7. A process according to claim 6 in which said probe comprises:

- a fluorophore covalently attached to the 5' end of the probe, or

- a quencher covalently attached to the 3' end of the probe.

8. A process according to claim 7 in which said fluorophore is selected from HEX, Texas Red and FAM.

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9. A process according to any one of claims 6 to 8 in which said probe comprises a nucleic acid sequence of length 10 to 40 nucleotide bases.