

WHAT IS CLAIMED:

1. A reaction mixture for detecting corneal dystrophy in a human subject, the reaction mixture comprising:
(A) at least a first amplification primer pair for amplifying and determining a first TGF β I gene sequence from a biological sample from the subject; and
(B) a first set of at least four detection probes including a first detection probe comprising the sequence of SEQ ID NO:25, a second detection probe comprising the sequence of SEQ ID NO:26, a third detection probe comprising the sequence of SEQ ID NO:48 and a fourth detection probe consisting of the sequence of SEQ ID NO:49.
2. The reaction mixture according to claim 1, further comprising a second set of at least three detection probes includes a fifth detection probe comprising a sequence selected from SEQ ID NO:45 and SEQ ID NO:47, a sixth detection probe comprising the sequence of SEQ ID NO:46, and a seventh detection probe comprising the sequence of SEQ ID NO:50, and at least a second amplification primer pair for amplifying and determining a second TGF β I gene sequence from the biological sample from the subject.
3. The reaction mixture according to claim 2, wherein the first amplification primer pair comprises a first amplification primer and a second amplification primer, the first amplification primer comprises nucleotide sequence SEQ ID NO:1, and the second amplification primer comprises nucleotide sequence SEQ ID NO:2.
4. The reaction mixture according to claims 2-3, wherein the first TGF β I gene sequence comprises nucleotides encoding amino acid residue 124 from the biological sample from the subject and the second TGF β I gene sequence comprises nucleotides encoding amino acid residue 555 from the biological sample from the subject.
5. The reaction mixture according to any of claims 2-4, wherein the second amplification primer pair comprises a third amplification primer and a fourth amplification primer, the third amplification primer comprises a nucleotide sequence SEQ ID NO:43, and the fourth amplification primer comprises a nucleotide sequence SEQ ID NO:44.
6. A method for detecting corneal dystrophy comprising:
(A) amplifying a first TGF β I gene sequence from a biological sample from a human subject using a reaction mixture comprising at least a first amplification primer pair for amplifying a TGF β I gene sequence and a set of at least four detection probes including a first detection probe comprising the sequence of SEQ ID NO:25, a second detection probe comprising the sequence of SEQ ID NO:26, a third detection probe comprising the sequence of SEQ ID NO: 48 and a fourth detection probe consisting of the sequence of SEQ ID NO:49;
(B) hybridizing one detection probe of the set of at least four detection probes to the first TGF β I gene sequence; and

(C) detecting a mutation in the first TGFβ1 gene sequence based on (i) the hybridization of the detection probe of the set of at least four detection probes that has hybridized in step (B) to the first TGFβ1 gene sequence and the failure of the other three detection probes of the set of at least four detection probes to hybridize to the first TGFβ1 gene sequence.

7. The method according to claim 6, wherein the reaction mixture comprises a reaction mixture as recited according to any of claims 1 to 5.
8. A method for detecting a genomic mutation associated with corneal dystrophy in a sample from a human subject, the method comprising:
 - (A) using epithelial cells of a human subject adhered to a tip of a substrate;
 - (B) agitating the tip of the substrate in a lysis solution that lyses cells adhered to the substrate;
 - (C) removing the substrate from the lysis solution upon completion of the agitating (B);
 - (D) incubating the lysis solution after the removing (C);
 - (E) isolating genomic DNA from the lysis solution to form a gDNA solution; and
 - (F) determining an identity of at least a nucleotide present in the TGFβ1 gene using at least a first primer pair for amplifying a TGFβ1 gene sequence, a first set of at least four detection probes including a first detection probe comprising the sequence of SEQ ID NO:25, a second detection probe comprising the sequence of SEQ ID NO:26, a third detection probe comprising the sequence of SEQ ID NO:48 and a fourth detection probe consisting of the sequence of SEQ ID NO:49, and the gDNA solution by concurrently exposing the gDNA solution to the first set of at least four detection probes, wherein: the at least a nucleotide is located at a particular position of the TGFβ1 gene corresponding to a single nucleotide polymorphism (SNP) associated with corneal dystrophy.
9. The method according to claim 8, further comprising: (G) determining an identity of at least a second nucleotide present in the TGFβ1 gene using at least a second primer pair, a second set of at least three detection probes including a fifth detection probe comprising a sequence selected from SEQ ID NO:45 and SEQ ID NO:47, a sixth detection probe comprising the sequence of SEQ ID NO:46, and a seventh detection probe comprising the sequence of SEQ ID NO:50, and the gDNA solution by concurrently exposing the gDNA solution to the first set of at least four detection probes and the second set of at least three detection probes, wherein: the at least second nucleotide is located at a second particular position, independent of the position of the at least a nucleotide, of the TGFβ1 gene corresponding to a second single nucleotide polymorphism (SNP) associated with corneal dystrophy.