

Title (en)

RAPID, ACCURATE IDENTIFICATION OF DNA SEQUENCE VARIANTS BY ELECTROSPRAY MASS SPECTROMETRY

Title (de)

SCNELLE, AKKURATE IDENTIFIZIERUNG VON DNA-SEQUENZVARIANTEN MITTELS ELEKTROSPRAY MASSENSPEKTROMETRIE

Title (fr)

IDENTIFICATION PRECISE ET RAPIDE DE VARIANTES DE SEQUENCES D'ADN PAR SPECTROMETRIE DE MASSE EN PHASE IONISEE PAR ELECTRO-PULVERISATION

Publication

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Application

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Abstract (en)

[origin: WO9747766A1] A method of detecting a polymorphism at a polymorphic site in the genome of an individual is described. The method involves amplifying a portion of a genomic DNA sample by polymerase chain reaction to produce an amplified segment that contains the polymorphic site, desalting the amplified segment, and determining the mass of the amplified segment by electrospray ionization mass spectrometry. Comparison of the mass of the amplified segment to a reference mass permits detection of the presence or absence of the polymorphism. Methods for heterozygosity and disease inheritance by mass spectrometry are also described. The figure shows the molecular weight transform spectrum obtained from the mass-to-charge spectrum of a 53-base oligonucleotide.

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