

A SIMPLE MOLECULAR ASSAY FOR RHD GENOTYPING

Rhesus (Rh) status is routinely determined by serological analyses, but molecular analysis of the RHD gene for phenotype prediction is required when discrepancy and/or ambiguity occurs. EFS has identified a novel predominant RHD allele resulting in the weak D phenotype in the Indian population. A new RHD genotyping method based on the detection of this novel allele is proposed.

INVENTION:

A method for detecting a novel allele of the RHD gene associated with a weak D phenotype in the Indian population. Thanks to its design the test is also valuable for identifying gene rearrangement and copy number variation, including whole RHD gene deletion and hybrid RHD-RHCE alleles. This invention was developed by the EFS laboratory located in Brest.

KEYWORDS:

RHD gene, RHD genotype, weak D phenotype, Indian population, wild-type, D-, D+, genotyping, D antigen, primers, rapid, simple, low-cost, Rh D-positive, Rh D-negative, multiplex PCR

DESCRIPTION:

This invention is a method for detecting the presence of a novel Indian-specific RHD variant allele, including duplication of a 12-kilobase region embedding RHD exon 3, in a DNA sample. This specific allele is the main genetic cause resulting in a weak D phenotype in the Indian population.

The proof-of-concept was achieved in 223 human genomic DNA samples of Indian origin.

cells stroma-dependent and thereby allow to reduce chemotherapy doses.

ADVANTAGES:

- Rapid test: Processing time is around 2 hours after genomic DNA extraction. Only one amplification step is necessary per test.
- Simple protocol: PCR mix ready to use, minimal material is required to carry out the test.
- Low cost genotyping test: cost <1 euro (reagent/test).

APPLICATIONS:

- Identification of the most prevalent weak D variant allele in the Indian population
- Identification of RHD copy number variation /RHD-RHCE gene rearrangement
- Clinical management of transfusion
- Pregnancies at risk

REFERENCE

- Y. FICHOU et al., Transfusion 2018

CONTACTS

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INTELLECTUAL PROPERTY

Pending

WO 2018/162516
US16/491645
EP18711859,1
IN201917040421
CA3053875
ZA3053875

