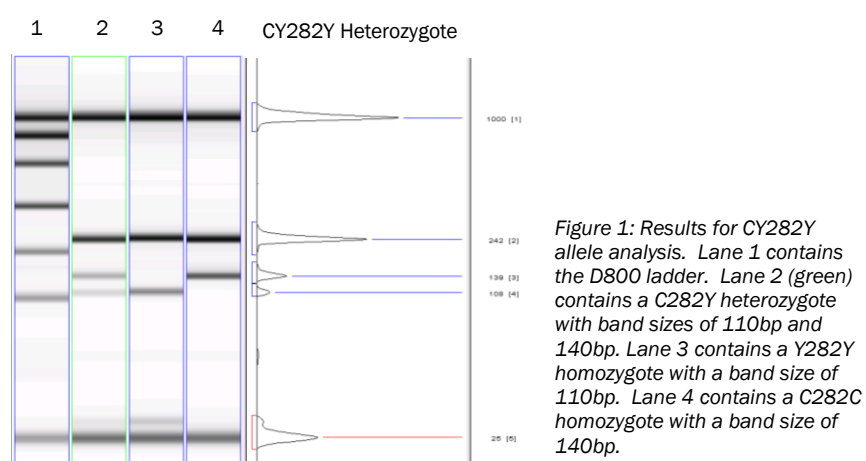


## Molecular typing of hereditary haemochromatosis is simplified with ScreenTape®



### ScreenTape used

D800

### Convenient set up

No reagent preparation

No gel loading

No manual analysis

### Total time

10 minutes

### Sample volume used

1 µl

The Lab901 ScreenTape System simplifies the analysis of molecular genetic testing in the laboratory. After HFE specific PCR and restriction enzyme digest, ScreenTape separates, images, analyses and creates GLP standard reports in 10 minutes. Bar-coding facilities create immutable traceability for essential test results.

### Introduction

Hereditary haemochromatosis (HH) is a recessively inherited disorder of iron consumption, causing the accumulation of iron in a number of body tissues. Type 1 HH is predominantly due to mutations in the HFE gene found on chromosome 6 in location 6p21.3, and mainly affects Northern European males between the ages of 40 and 60 years. Approximately 90% of clinically symptomatic UK patients are homozygous for a mutation at position 282 of the HFE gene, whereby a cysteine to tyrosine substitution has occurred (HFE mutation C282Y). The remaining patients are compound heterozygotes for C282Y and for a histidine to aspartic acid change at position 63 (H63D).

### Materials and Methods used in PCR

After DNA extraction, PCR is performed to amplify fragments containing the relevant mutation. RsaI or NdeII restriction enzymes (RE) are then used to identify C282Y and H63D mutant alleles respectively.

### ScreenTape DNA Analysis Procedure

After the PCR and RE digestion, samples were mixed 1:4 with Lab901 loading buffer and placed in the TapeStation along with D800 ScreenTape. Full analysis of 7 samples was achieved, with no user intervention, within 10 minutes.

### Results

The analysis software launched immediately (see Figure 1). Sample 1, edged with green in lane 2, contains a C282Y heterozygote. The electropherogram on the right hand side of the screengrab details the sizes of each fragment along with relative quantitative information for each fragment in the sample.

### Benefits of using ScreenTape for haemochromatosis

The ScreenTape system allows fully automated, walk-away analysis of HH samples with results analysed and presented in less than 10 minutes. Full traceability and secure file formats allow GLP standard reporting. Pre-packaged reagents decrease manual intervention, save time and increase safety whilst proven reproducibility instils confidence in assay results.