

EVALUATION OF THE PRECISION ID WHOLE MTDNA GENOME PANEL FOR FORENSIC ANALYSES



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Introduction

Degraded samples require the use of short amplicons, which can be achieved using the Precision ID mtDNA Whole Genome Panel (Thermo Fisher Scientific, CA). This kit comprises 162 amplicons with an average targeted fragment size of 175 bp, suitable for most forensic samples. The selected samples were processed as routine forensic caseworks and yielded limited or no result with conventional Sanger Type Sequencing.

Materials and Methods

<u>Samples:</u> Seven (cold) casework samples from Austria and Sweden (4 hair shafts, 1 floor swab and 2 reference samples) and eight ancient samples from a medieval grave in Austria (7 teeth and 1 femur)

<u>Process:</u> sample libraries were processed using the Ion Ampliseq Library Kit, emulsion PCR and sequencing were performed on the One Touch 2 and the PGM respectively. 6 to 10 samples were loaded on an Ion 318 chip <u>Analysis:</u> Torrent Suite 4.4.3 and Torrent Suite 4.6 / variant caller (v4.4.3.3 and v5.0.4.0)

PER BASE COVERAGE VALUES • high quality sample: inside ring mean coverage value of 12,266 • ancient DNA sample: outside ring mean coverage value of 3

Results

General performance

- the samples showed significant patterns of degradation and were useful for evaluating the performance of the panel
 - full mitogenomes for all casework and two out of eight ancient samples could be sequenced with this kit
 - the remaining six ancient samples yielded partial profiles
 - full concordance (STS-CE results) for all samples in overlapping regions could be achieved
 - more forensically relevant information was generated
 - the panel increased the discrimination power and resulted in more specific haplogroup estimation

Time management

This approach would allow generating 10 full mitogenomes even from highly degraded DNA in the same time required to generate a single full mitogenome from a high quality sample using STS-CE.

Discussion

This method proved to be useful for the application on severely degraded samples. It matched the results from previous STS-CE and brought additional information in the control and coding regions. The generation of full mitogenome haplotypes from these degraded specimens brought valuable information and demonstrates the usefulness of such an approach to forensic research.

A cold case investigation from 1991 where STS-CE analysis yielded a partial HVS-I haplotype, matching the victim's profile (from a hair which had been in seawater for over 10 years), could be solved by using the Precision ID mtDNA Whole Genome Panel which resulted in a full mitogenome haplotype and excluded the victim

CASEWORK

codR

as donor of the bloodstain

bloodstain

victim

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