

Electronic Supplementary Information

Mass Spectrometry based Trinucleotide Repeat Sequence detection using Target Fragment Assay

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Table S1. Sequences of the synthesized oligonucleotides.

Strand	Sequence(5'-3')	CAG Repeat numbe r
Forward primer (P1)	5'- ATGAAGGCCTTCGAGTCCCTCAAGTCC-3'	
Reverse primer (P2)	5'-biotin-CGGTGGCGGCTGTTGCTGCTGCTGCTG-3'	
HTT-10	5'-AGCTGATGAAGGCCTTCGAGTCCCTCAAGTCCTTC (CAG) _n CAACAGCCGCCACCGCCGCCGCCGCC-3'	n=10
HTT-19	5'-AGCTGATGAAGGCCTTCGAGTCCCTCAAGTCCTTC (CAG) _n CAACAGCCGCCACCGCCGCCGCCGCC-3'	n=19
HTT-30	5'-AGCTGATGAAGGCCTTCGAGTCCCTCAAGTCCTTC (CAG) _n CAACAGCCGCCACCGCCGCCGCCGCC-3'	n=30
HTT-40	5'-AGCTGATGAAGGCCTTCGAGTCCCTCAAGTCCTTC (CAG) _n CAACAGCCGCCACCGCCGCCGCCGCC-3'	n=40
HTT-50	5'-AGCTGATGAAGGCCTTCGAGTCCCTCAAGTCCTTC (CAG) _n CAACAGCCGCCACCGCCGCCGCCGCC-3'	n=50
HTT-60	5'-AGCTGATGAAGGCCTTCGAGTCCCTCAAGTCCTTC (CAG) _n CAACAGCCGCCACCGCCGCCGCCGCC-3'	n=60

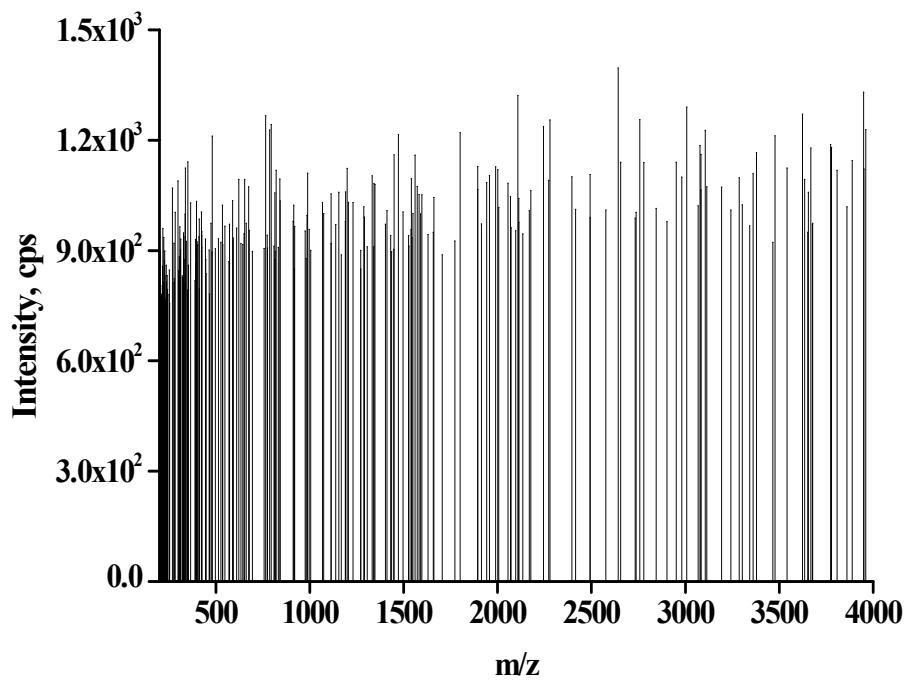


Figure S1. The full scan mass spectrum of PCR amplicons obtained by TFA approach using DNA target HTT-19 (without preliminary acidic degradation). Sample was dissolved in 10 nM ammonium acetate buffer, detected in negative ion mode.

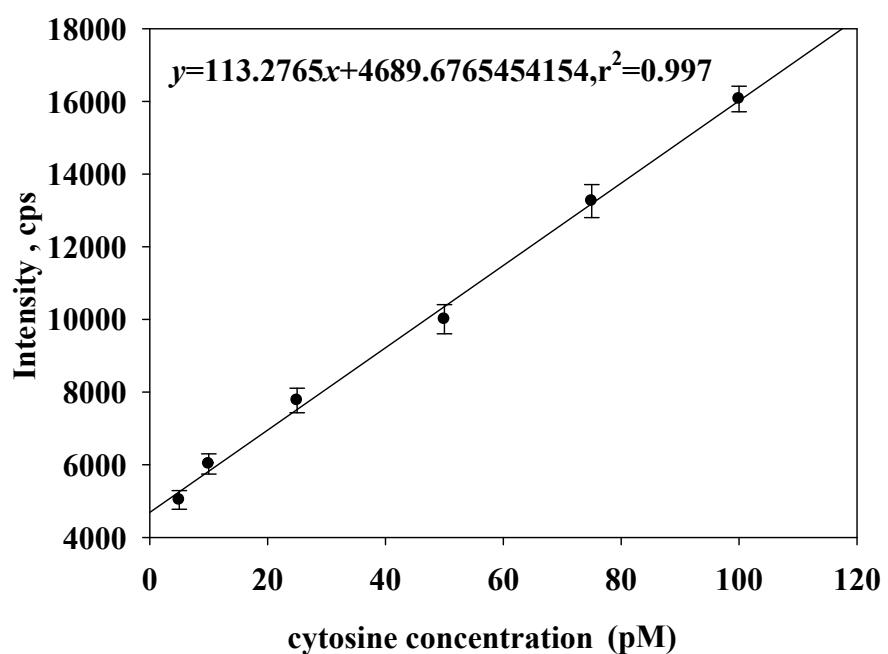


Figure S2. Plot of MS/MS peak areas versus concentrations of DNA target HTT-19 (expressed in cytosine concentration).

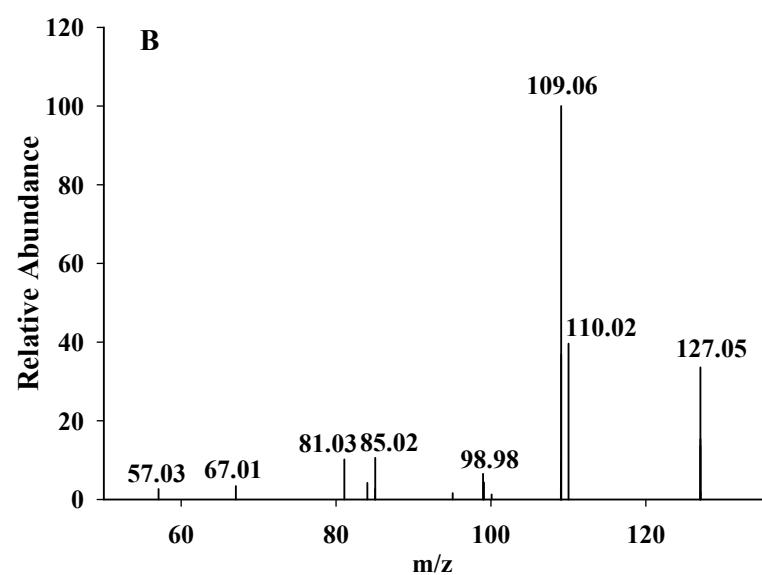
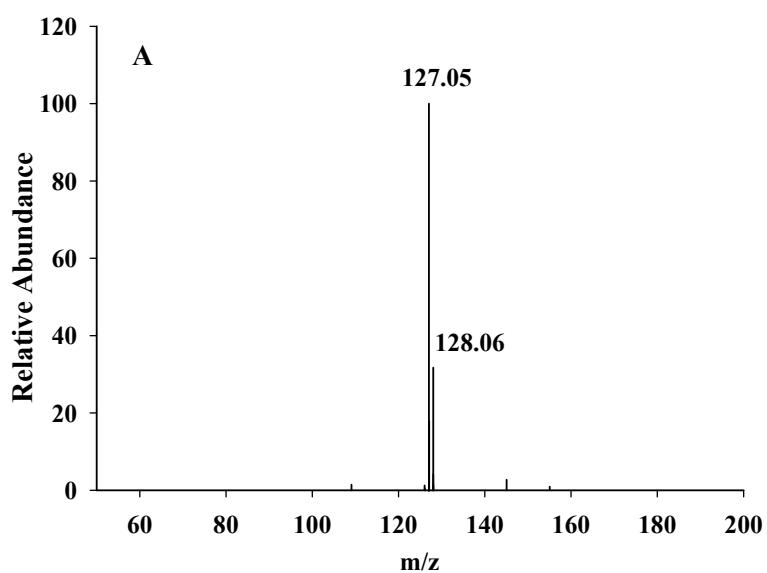


Figure S3. (A) Full scan mass spectrum of thymine. (B) Product ion scan spectrum of cytosine using protonated molecular ion of thymine (m/z 127.05) as precursor ion.

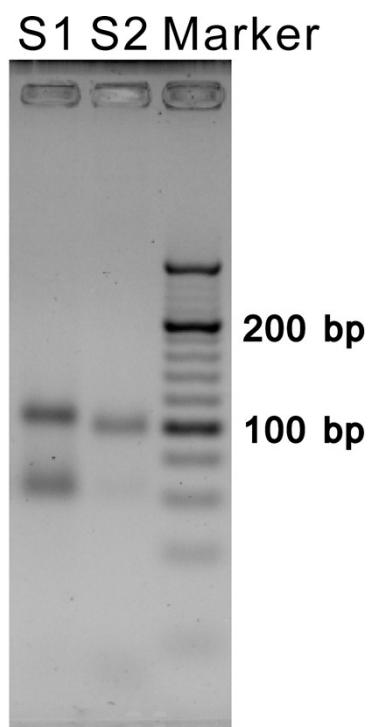


Figure S4. Agarose gel electrophoresis image of PCR products using genomic DNA extracted from two blood samples of two healthy people. Lane 1: sample 1 (S1); Lane 2: sample 2 (S2); Lane Marker is the DNA size marker.