

Sensitive and Accurate Sizing of CAG Repeats in the HTT Gene

The AmplideX® PCR/CE HTT Kit makes the detection and sizing of CAG trinucleotide repeats in the HTT gene easier than ever. Accurate sizing of the CAG expansions in HTT is challenged by the presence of adjacent SNPs and variable CCG repeats, which can complicate primer binding and cause allele dropouts. With its unique, two-primer design, the AmplideX PCR/CE HTT Kit overcomes these challenges to accurately and reliably report the number of repeats in one day, from as little as 10ng gDNA.

REDUCED COMPLEXITY

- Proprietary PCR solutions for GC-rich amplification and detection
- Eliminates need for multiple PCRs one result, straightforward analysis
- Resolves zygosity and detects large expansions

OPTIMIZED WORKFLOW

- Fully kitted, end-to-end solutions that significantly reduce hands-on-time
- Sample-to-result possible within a single shift
- Identical PCR and CE conditions as the AmplideX® PCR/CE DMPK Kit

QUALITY PERFORMANCE

- Reliable, unambiguous results and a robust stutter peak pattern
- Accurate sizing across the entire CAG repeat range
- Software converts raw base pair data into number of repeats



Figure 1. AmplideX PCR/CE HTT Kit Workflow.





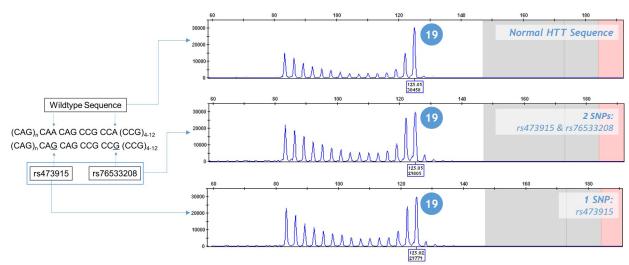


Figure 2. Repeat profile is conserved across known SNPs

Sample ID	Reported	Reported	Observed
Number	Genotype	Alleles	Alleles
NA20206	NORMAL	17, 18	18, 18
NA20207	NORMAL	19, 21	19, 21
NA20208	EXPANDED	35, 45	35, 45
NA20209	EXPANDED	45, 47	45, 47
NA20210	EXPANDED	17, 74 or 75	17, 75
NA20245	NORMAL	15, 15	15, 15
NA20246	NORMAL	15, 24	15, 24
NA20247	INTERMEDIATE	15, 29	15, 29
NA20248	REDUCED PENETRANCE	17, 36	17, 36
NA20249	REDUCED PENETRANCE	22, 39	22, 39
NA20250	EXPANDED	15, 40	15, 40
NA20251	EXPANDED	39, 50	39, 50
NA20252	EXPANDED	22, 65 or 66	22, 63°, 66
NA20253	EXPANDED	22, 101	22, 100, 128'
SRM 2393 A	INTERMEDIATE	15, 29	15, 29
SRM 2393 B	REDUCED PENETRANCE	17, 36	17, 36
SRM 2393 C	EXPANDED	15, 40	15, 40
SRM 2393 D	EXPANDED	35, 45	35, 45
SRM 2393 E	EXPANDED	39, 50	39, 50
SRM 2393 F	EXPANDED	17, 75	17, 75
	*Additional min	or peak consistently identi	ified with Asuragen assay

Figure 3. 100% Concordance (±1 repeat) observed with previously characterized samples, as reported by Kalman et al. (2007)

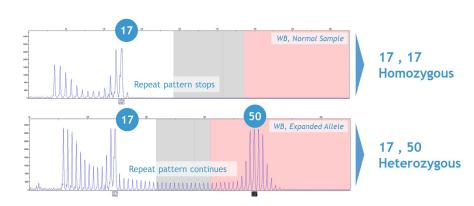


Figure 4. Clear resolution of zygosity.



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