Supplementary table 1. Primers sequences and thermocycling conditions

	Primers	PCR	Cycling	Ramp Rate
RP-PCR	NBF19-R: 5'-AGCGCCCACAGCAGAGCGGC-3' pGEX3'-$(CGG)_5$: 5'-CCGGGAGCTGCATGTGTCAGAGG$(CGG)_5$-3' Fam-pGEX3': 5-FAM-CCGGGAGCTGCATGTGTCAGAGG-3'	Takara LA taq 1.25units 2X GC buffer II 12.5µl dCTP/dTTP/dATP (2.5mM) 4µl 7 deaza-dGTP (2.5mM) 4µl Primers: 180nmol/µl for NBPF19-R and fam-pGEX3'; 9nmol/µl for pGEX3'-$(CGG)_5$ gDNA 100ng H_2O to 25µl	95°C 5mins [95°C 30s, 98°C 10s, 62°C 30s, 72°C 2mins] X 50 4°C	$2.5°Cs^{-1}$ $2.5°Cs^{-1}$ $-1.5°Cs^{-1}$ $2.5°Cs^{-1}$
PCR for Fragment Sizing	NBPF19-5R2: 5'-TACTCACCATGCGCGGGGGT-3' pGEX3'-NBPF19-6F: 5'-CCGGGAGCTGCATGTGTCAGAGGGCCTGTGCTTCGGAC-3' fam-pGEC3': 5'-FAM-CCGGGAGCTGCATGTGTCAGAGG-3'	Takara LA taq 1.25units 2X GC buffer II 12.5µl dNTP (2.5mM) 4µl Primers: 180nmol/µl each gDNA 100ng H_2O to 25µl	98°C 1min [98°C 10s, 58°C 30s, 68°C 30s] X 35 4°C	n/a

RP-PCR Repeat Primed PCR, 7 deaza-dGTP 7-Deaza-2'deoxyguanosine-5'triphosphates, dATP deoxyadenosine triphosphates, dATP deoxycytodine triphosphates, dTTP deoxythymidine triphosphates, dNTP deoxynucleotide triphosphates, gDNA genomic DNA

Supplementary materials

Supplementary figure 1. (A, C) Representative electropherograms of RP-PCR analysis in a positive control shows a characteristic sawtooth appearance as compared to a patient without the expansion. (B, D) Fragment analysis of a positive control only captured one non-expanded allele (~19 GGC repeats). In contrast, two alleles were captured in a patient in our UK cohort (~15/28 GGC repeats).