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Sequence analysis of PCR products (using proximal PCR primers<sup>a)</sup> to characterise homopolymeric tract and flanking regions. The illustrated tract possessed nine G repeats.

PCR amplification of homopolymeric tract and flanking regions using genomic DNA or 'Round 1' PCR products as template in conjunction with proximal FAM-labelled primers<sup>a</sup>.



Fragment analysis of FAM-labelled PCR products (blue peak) against a GeneScan 500 LIZ size standard (orange peaks) to enable quantitation of homopolymeric tract length.

## Specimen DNA

<sup>1</sup>Round 1<sup>1</sup> PCR amplification of homopolymeric tract and flanking regions using remote, intragenically directed primers<sup>b</sup>.

'Round 2' (nested) PCR amplification of homopolymeric tract and closely flanking regions using 'round 1' PCR product as a template in conjunction with intermediate, intragenically directed primers<sup>6</sup>.



Agarose gel electrophoresis gel to confirm the presence of 'round 2' PCR products.

