**Supplement 2: Identification of *RFC1* repeat expansions in exome and genome datasets**

Exome sequencing was performed on genomic DNA from patients P2.1 and P10. Exonic regions were enriched with a SureSelect Human All Exon Kit V6 (P2.1) or V7 (P10) (Agilent technologies, Santa Clara, California). Generated libraries were sequenced as 2x125/2x100 bp paired-end reads on a HiSeq2500 / NovaSeq 6000 system (Illumina, San Diego, California) to an average 109.8X /141.5X coverage of the target region. 71.7 % and 74.5 % of the generated reads mapped to the target regions.

Libraries for genome sequencing were generated from genomic DNA of patient P9 with a PCR-free protocol (Illumina, San Diego, California) for subsequent sequencing as 2x150 bp paired-end reads on a NovaSeq 6000 system to an average 48.2X coverage.

Generated sequences were analyzed using the megSAP pipeline (https://github.com/imgag/megSAP). Routine diagnostic data analysis failed to identify disease-causal point mutations or copy number variants in established disease genes previously associated with the patients’ disease phenotypes.