

Figure 2s. Genomic region of *SLC7A9* rearranged in patients with the 5 kb deletion and the 5 kb duplication. We have detected two novel large mutations in two Spanish families: a 5kb deletion (c.1224+4166_c.1399+119del) and a 5 kb duplication (c.1224+4166_c.1399+119dup) both affecting exon 12, originated by the crossing over of two 195 bp sequences (Repeat 1 and 2), separated by 4778 bp, which differ by 1 nucleotide. Both mutations have been confirmed by standard PCR method using primers which specifically amplify the mutated allele in both cases. Primers sequences of F11, F13, R10d and R13 are listed in Materials and Methods.

