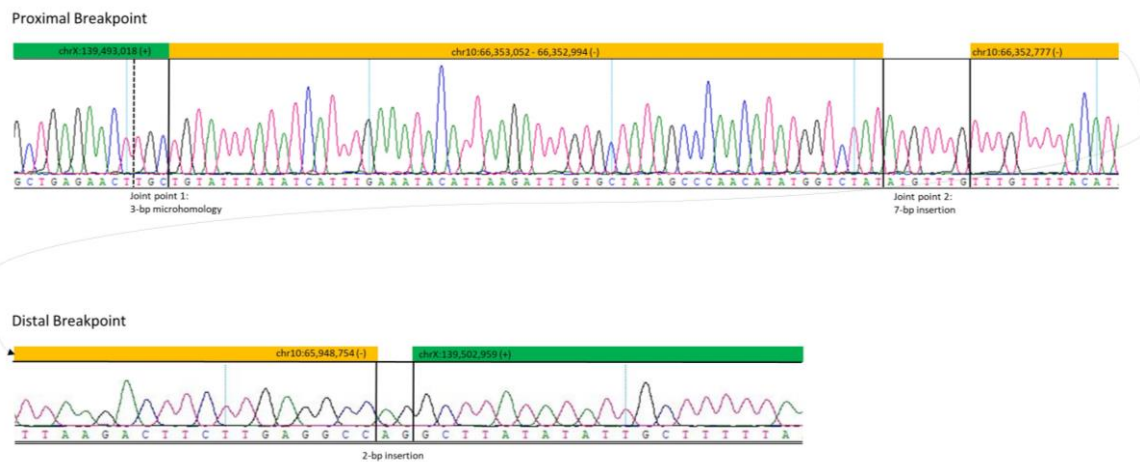
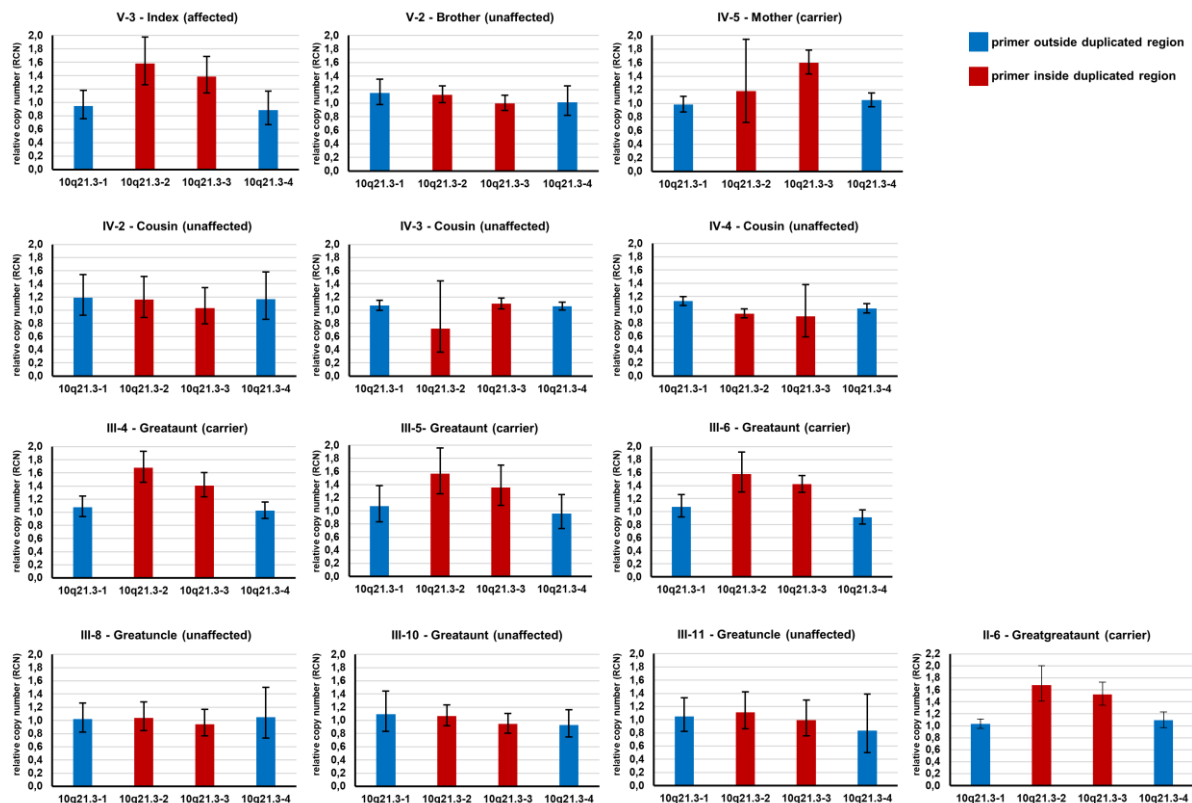


Supplementary Figure 1



Sequence analysis of the proximal and distal breakpoint junctions at base-pair resolution. Between the green marked reference sequence on chromosome X, the inversely inserted duplicated sequence from chromosome 10 is shown in yellow.

Supplementary Figure 2



Validation of the duplication and its segregation by qPCR. For all DNA samples available from the family two primer pairs adjacent (blue) and inside (red) the duplicated region respectively were used to investigate the relative copy number for these regions. By this approach, we confirmed the presence of the duplication in the index patient (V-3) and found the female individuals IV-5, III-4, III-5, III-6 and II-6 to be obligate carriers. RCN, relative copy numbers, Error bars indicate SD.

Supplementary Table 1

<u>breakpoint PCR</u>		
	Forward (5'-3')	Reverse (5'-3')
<i>BP1</i>	GGAGGTGGCAATTTCAAGAG	CCAAGGTAGGCTTCAAGACAAG
<i>BP2</i>	AGTGGGTAGAAGCCGGAGA	AGTGGCTTGTTCACCAG
<u>qPCR</u>		
	Forward (5'-3')	Reverse (5'-3')
<i>10q21.3-1</i>	GGCCTCTACTTGTGAGGTGC	GTAGGCATATCTCCGCCACC
<i>10q21.3-2</i>	ACAGGGCTAGGCAGGTAAGA	TCTAGCCAGGGCCTGTACTT
<i>10q21.3-4</i>	GCCTTGCTGTGGGTAGTAT	AAGGTCTCTCCACTCAGGC
<i>10q21.3-5</i>	CCCAAGGCTTATCTTTGGAGGA	GTTGAGCAAGGAGCTTTCTTCTG