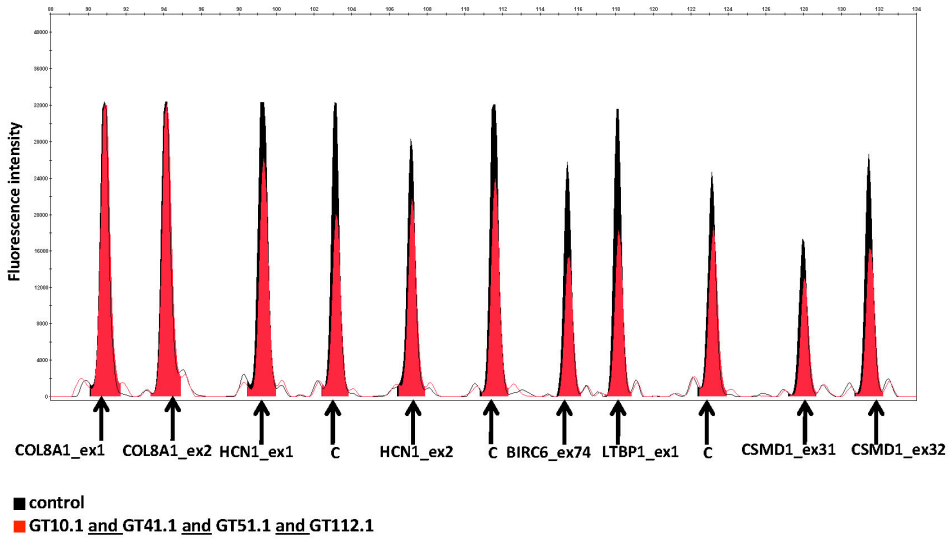
Figure S5: CNV calls using Multiplex Ligation-dependent Probe Amplification (MLPA)

Figure S5-1: Validation of the SNP-based CNV calls in *COL8A1* and *NRXN1* by MLPA



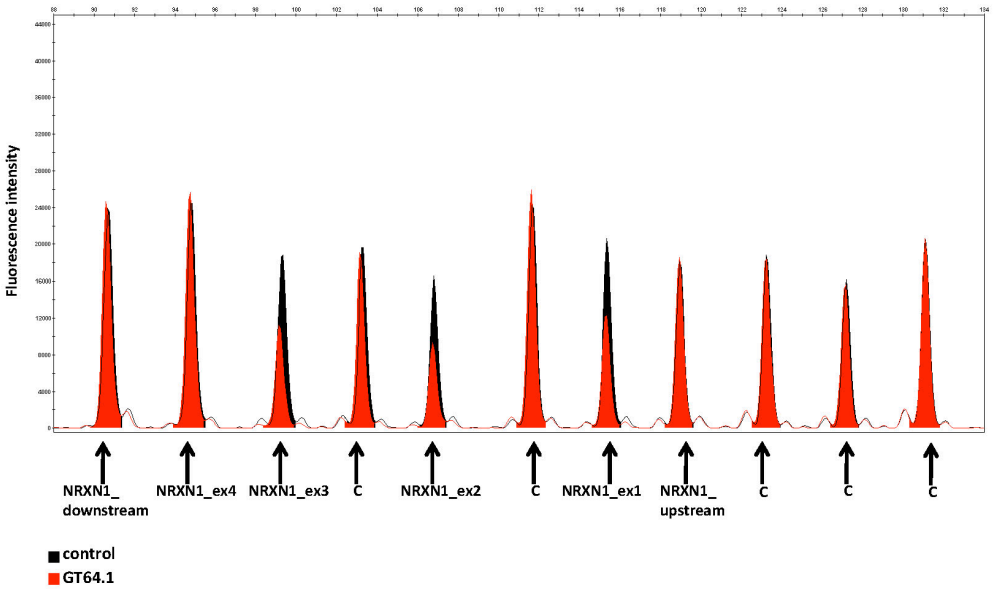
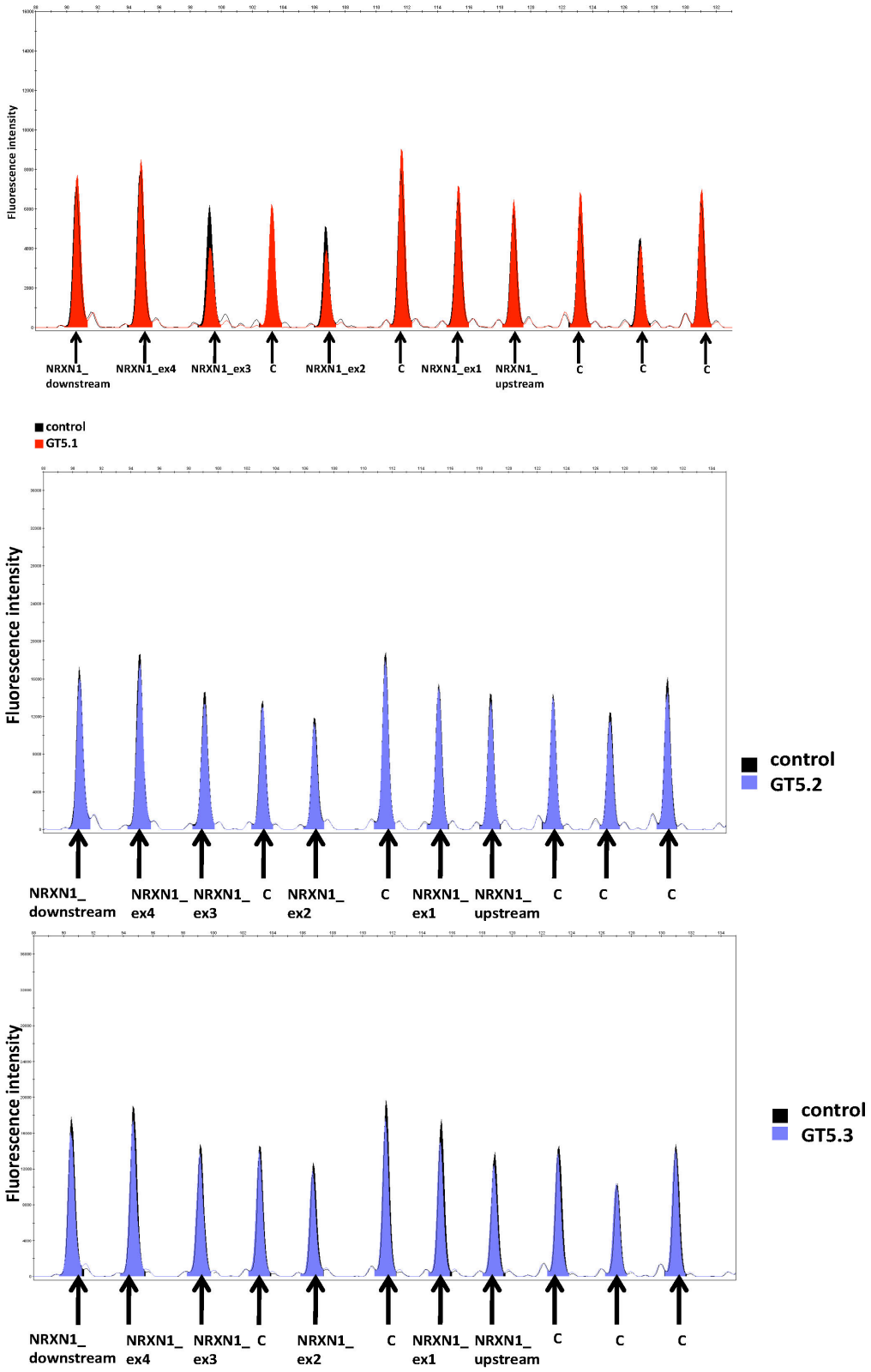
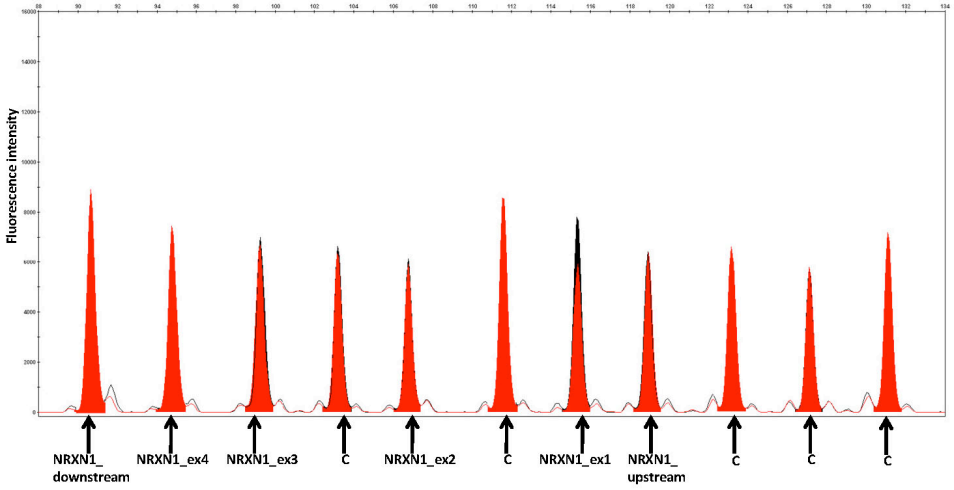


Figure S5-2: Detection of *de novo* deletions in *NRNX1* (exons 2 and 3) in TS cases.

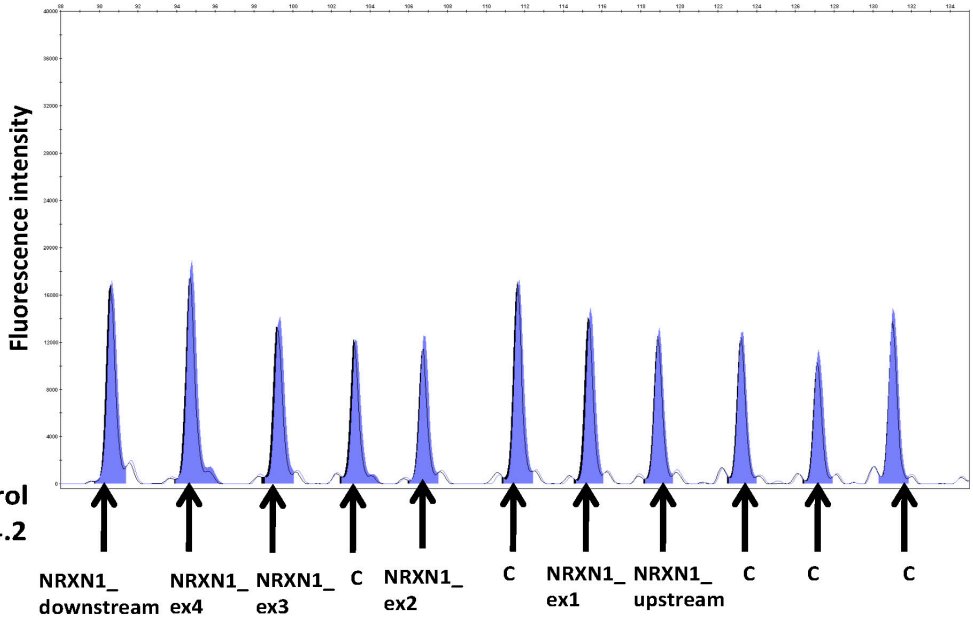
A. Trio 5 (GT5.1=case, GT5.2=parent, GT5.3=parent)



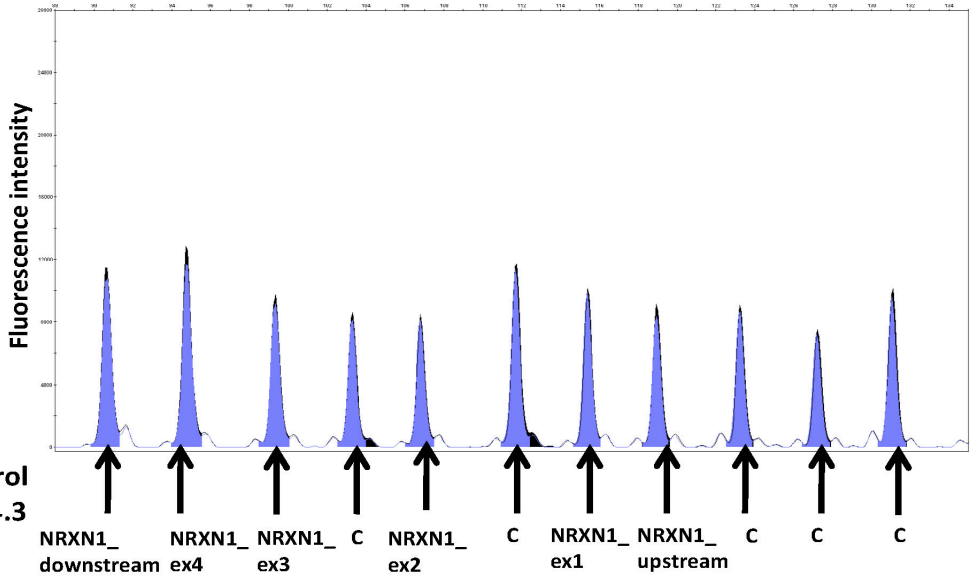
B. Trio 34 (GT34.1=case, GT34.2=parent, GT34.3=parent)



control
GT34.1



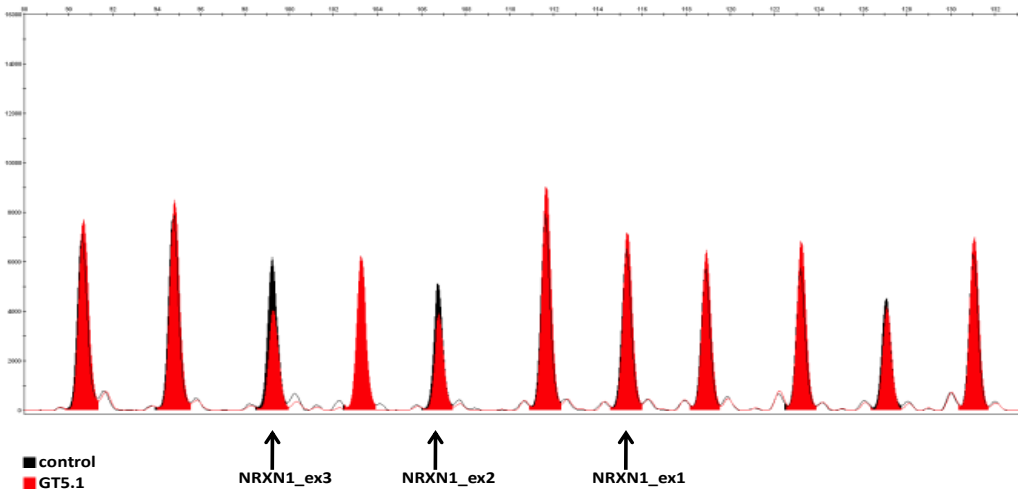
control
GT34.2



control
GT34.3

Figure S5-3: Two additional TS cases (GT5.1 and GT34.1) with deletions involving either exon 1, 2 or 3 of *NRXN1* detected by MLPA.

GT5.1 – heterozygous deletion of NRXN1 exons 2 and 3



GT34.1 – heterozygous deletion of NRXN1 exon 1

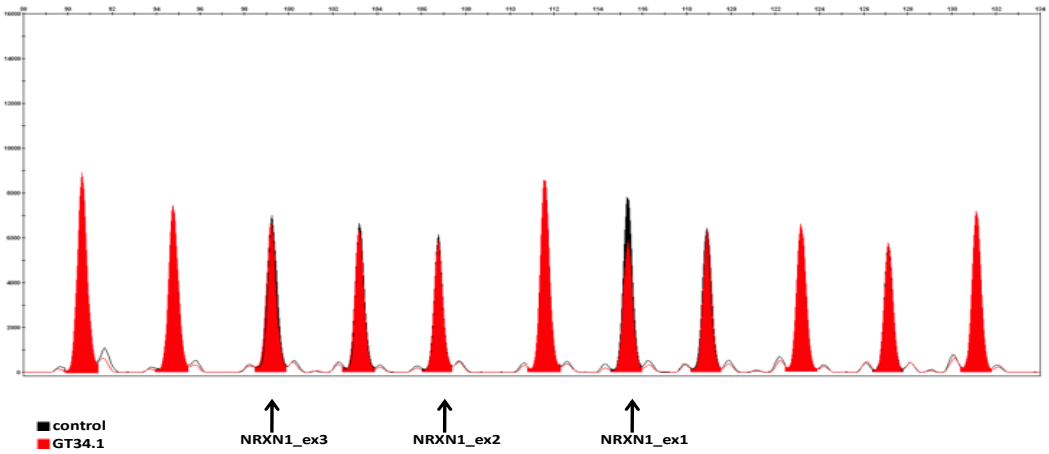
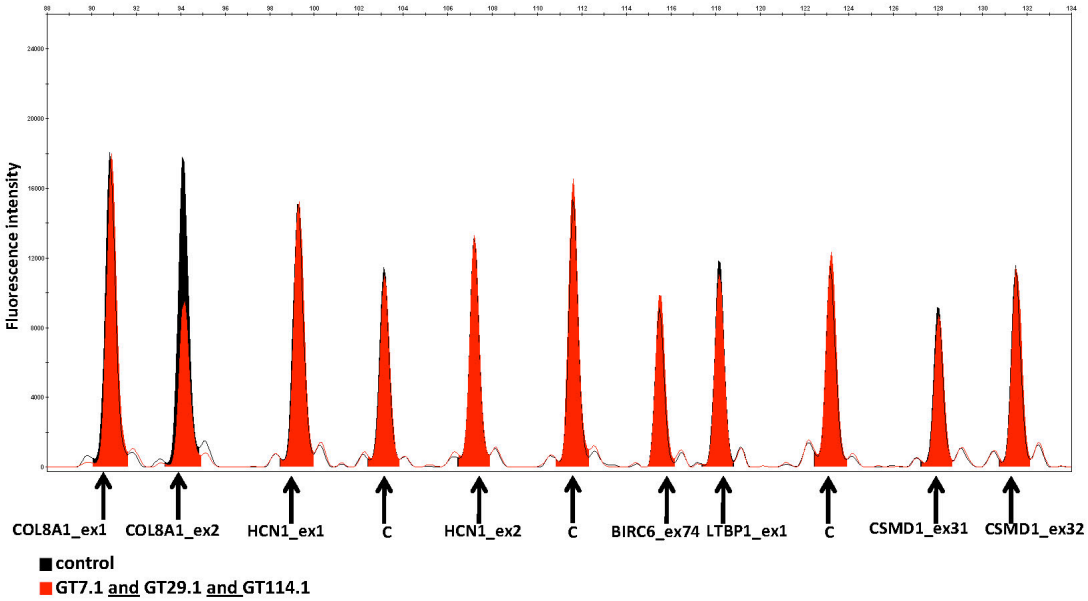



Figure S5-4: Three additional TS cases (GT7.1, GT29.1 and GT114.1) with deletion of exon 2 of *COL8A1* detected by MLPA.