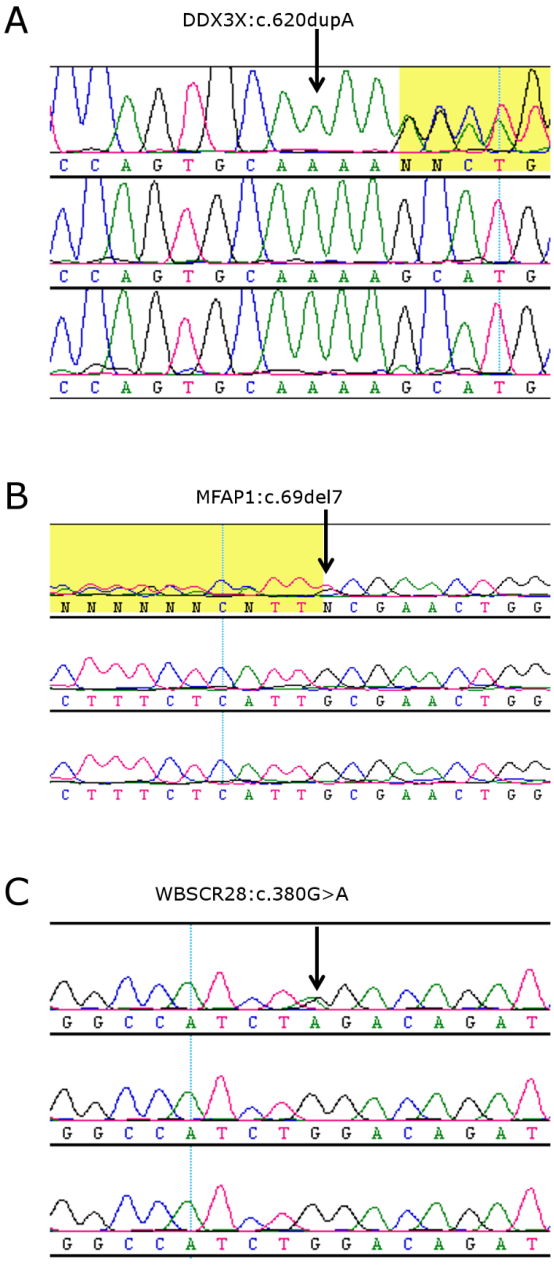
Supplementary figure S1. LoF *de novo* mutations identified in *DDX3X*, *MFAP1* and *WBSCR28* in 3 trios affected with NTDs. Chromatograms of the 3 trios (from top to bottom: proband, mother, father) showing the c.620dupA *DDX3X* frameshift variant (panel A), the c.69del7 *MFAP1* frameshift variant (panel B) and the c.380G>A *WBSCR28* nonsense variant (panel C).



Supplementary figure S2. *De novo* mutations identified in orthologues of genes associated with NTDs in mouse models. Chromatograms of the 2 trios (from top to bottom: proband, mother, father) showing the c.1171C>T *GRHL3* variant and (panel A) and the c.4475G>A *PTPRS* variant (panel C). Multi-species alignment of the amino acids surrounding the p.R391C variant in GRHL3 (panel B) and the p.R1492Q variant in PTPRS (panel D).