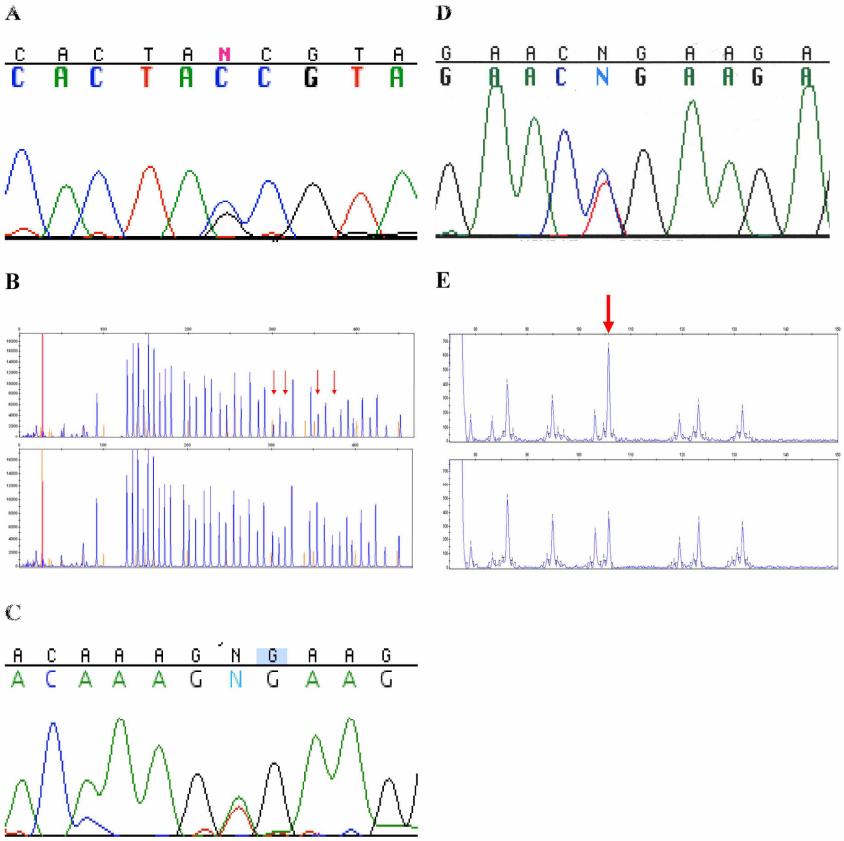
Figure S1: Identification of previously unpublished mutations in the cohort. (A) Sequence analysis of exon 2 of *GLI3* in patient OX1746 reveals the mutation 366C>G, Y122X. (B) MLPA chromatogram from patient OX3448 (upper panel) and normal control (lower panel). Peaks from probes hybridising to *GLI3* exons 10, 11, 13, and 14 are marked with red arrows (note: a probe for *GLI3* exon 12 was not available in this earlier version of probe set P179). Comparison with the equivalent peaks in the control shows a reduction in peak height, indicating a heterozygous deletion of these exons. (C) Sequence analysis of *TBX5* exon 4 in patient OX2084 reveals the mutation 266T>A, V89E. (D) Sequence analysis of *HOXD13* exon 2 in patient OX3015 reveals the mutation 995C>T, R319X. (E) MLPA chromatogram from patient OX3424 (upper panel) and normal control (lower panel). Peak from the *ZRS* probe is marked with a red arrow. Comparison with the equivalent peak in the control shows an increase in peak height, consistent with a *ZRS* triplication.

Figure S1


A
C A C T A N C G T A
C A C T A C C G T A
B
C
A C A A A G N G A A G
A C A A A G N G A A G
D
G A A C N G A A G A
G A A C N G A A G A
E