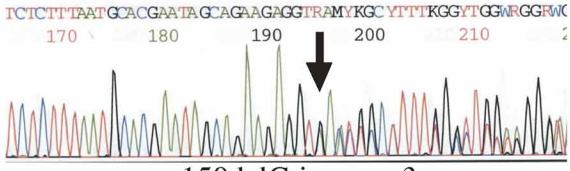
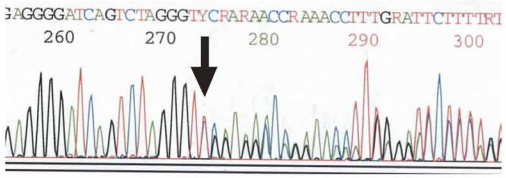
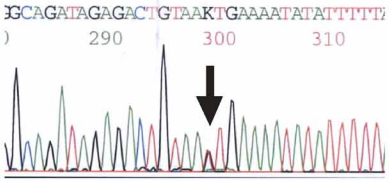


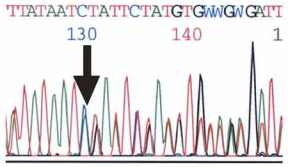
150delG in exon 3
(stop codon 28 amino acids downstream)



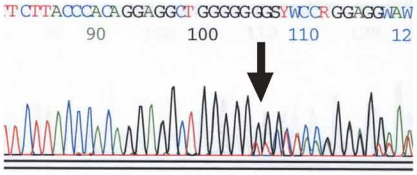
2520delT in exon 10
(stop codon 5 amino acids downstream)



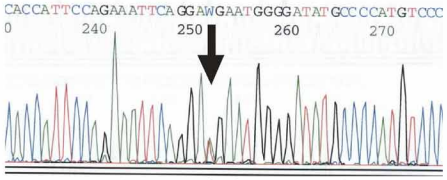
6763+5 G –>T in intron 39
(splice site; seen in 2 affected sibs and not in parents)



3023-3027delTGTCT in exon 10
(stop codon 1 amino acid downstream)



1546-1547insG in exon 10
(stop codon 3 amino acids downstream)



M1K (2G->A) in exon 2
(missense in codon one; seen in 3 affected sibs and not in parents)

Supplementary Figure 2. Mutations in *NIPBL* identified in CdLS individuals. Genomic sequence from all 6 mutations reported are depicted. Arrows indicate site of mutation. The mutations identified included: A *de novo* 276delG in exon 3 creating a stop codon 28 amino acids downstream. A *de novo* 2646delT in exon 10 resulting in a stop codon 5 amino acids downstream. A 3023-3027delTGTCT in exon 10 resulting in a stop codon 1 amino acid downstream (not present in the mother, father not available for testing). A *de novo* 1672 1673insG in exon 10 resulting in a stop codon 3 amino acids downstream. A missense mutation in the first codon, M1K (2G→A) was identified in all 3 affected sibs (who each have a different father), not present in their mother, nor in the 2 fathers on whom samples were available for screening. A splice site mutation (6889+5 G>T) in the intron between exon 39 and 40 seen in the 2 affected siblings and not in either parent.