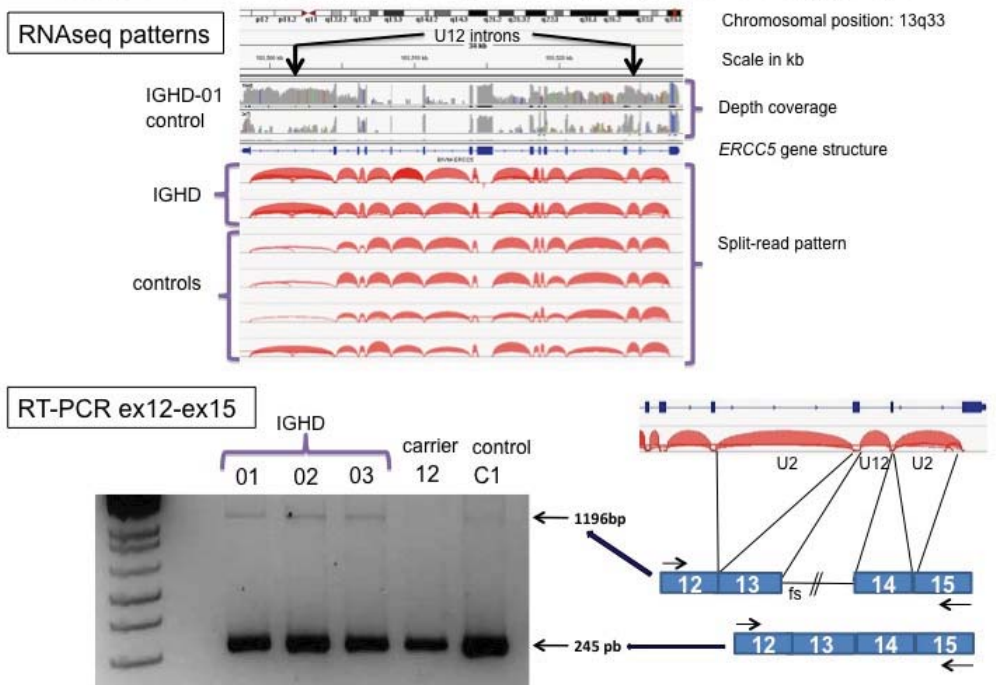
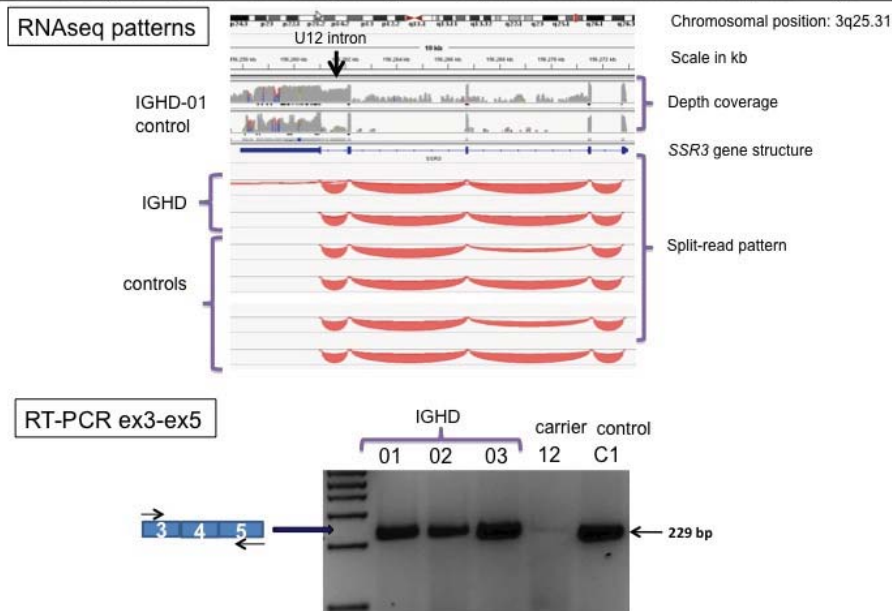


ERCC5 excision repair cross-complementing repair deficiency, group 5



Supporting figure S6: Transcription profiles of the *ERCC5* gene. Increased U12-type intron retention is seen in patients, and also detected but weaker in controls. No alternative transcripts are detected in the studied interval.

SSR3 signal sequence receptor gamma (translocon-associated protein gamma)



Supporting figure S7: Transcription profiles of the *SSR3* gene. Increased U12-type intron retention is seen in patients by RNAseq, although a single transcript with no differences between cases and controls is detected by RT-PCR.