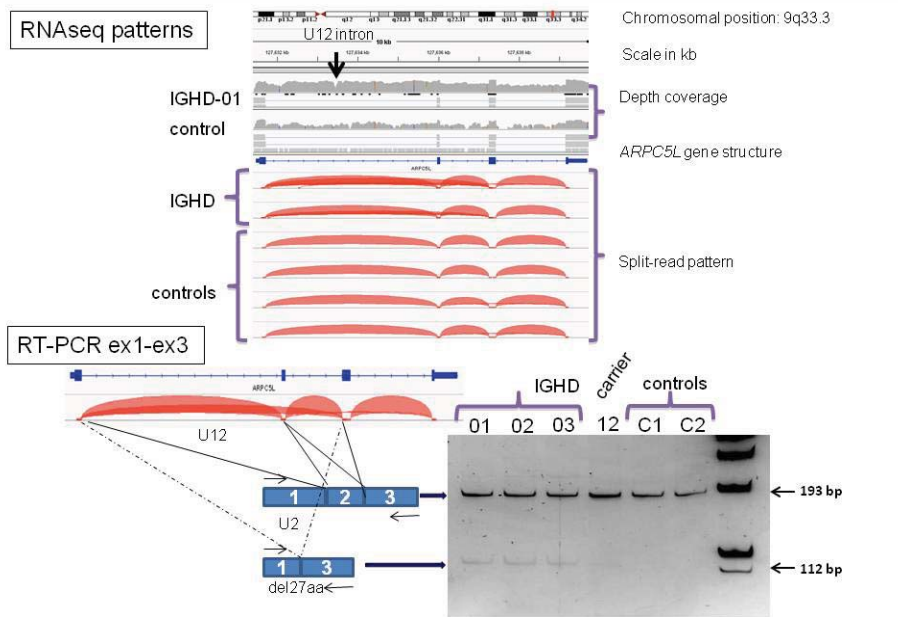
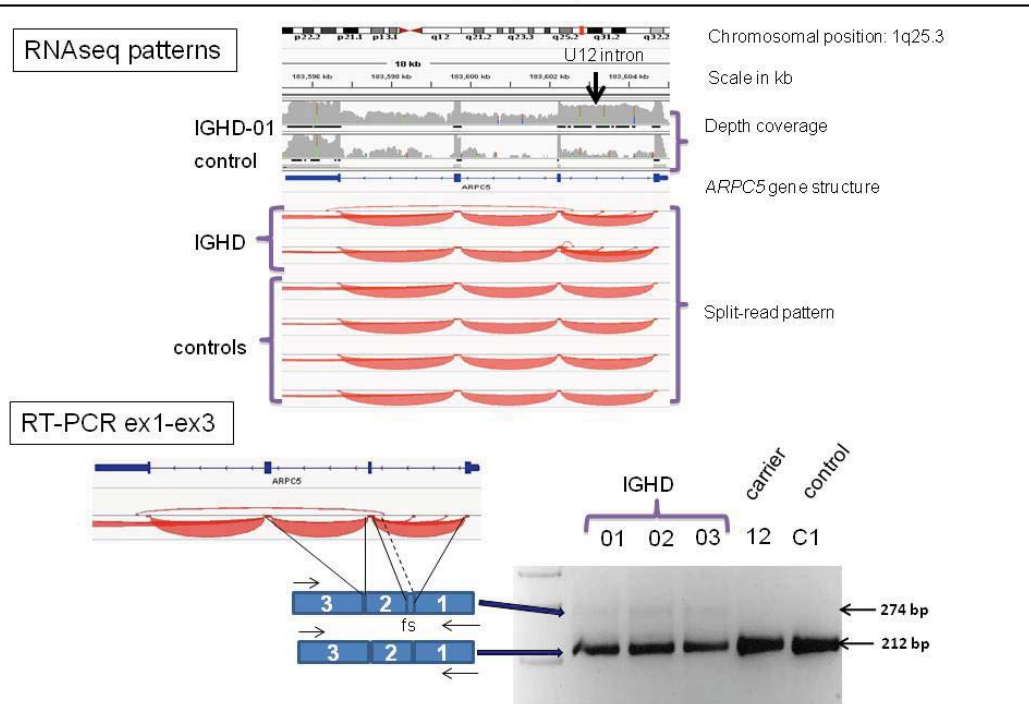


ARPC5L actin-related protein 2/3 complex, subunit 5-like



Supporting figure S2: Transcription profiles of the *ARPC5L* gene. Poor U12-type splicing with the use of alternative (cryptic) U2-type splicing and exon skipping is only seen in patients, being absent in controls. Intron retention is also higher in patients, but affects both U12 and U2-type introns.

ARPC5 actin-related protein 2/3 complex, subunit 5



Supporting figure S3: Transcription profiles of the *ARPC5* gene. Increased U12-type intron retention is seen in patients with otherwise similar U12-type splicing. An alternative U2-type cryptic splicing within intron 1 is very weakly seen in patients, but absent in controls.