

Supplementary figure 1

A) IGV (Integrative Genomics Viewer) image of RNA sequencing showing expression of variant c.622T>C, p.(*208Glnext*39) on transcript level in *CLDN11* (arrow). B) Sashimi Plot showing alternative splicing (arrow) of *CLDN11* (full-length isoform vs. shorter isoform) but no aberrant splicing. Control has been added for comparison.

