

Figure S13: A) UCSC screen capture of *POLR1B* **splice region.** Figure shows the human homologous sequence of the splice-region site including the alignment of 8 mammalian, and three non-mammalian species (chicken, frog, and zebrafish). The track '100 Vert. Cons' shows the basewise PhyloP sequence conservation scores. The *POLR1B* affected splice-donor position is indicated in the black box. The GTRAG site affected by the LA1 splice-region mutation is strongly conserved in vertebrates in all 11 species. **B) Exon skipping of** *POLR1B* **exon 14 in one LA1 carrier animal**. Figure shows the Ensembl-predicted *POLR1B* transcript and the RNA-seq alignment from one carrier animal from exon 13 to exon 15. The location of the mutation is indicated with a red cross.