Supplementary Materials: mRNA Quantification of *NIPBL* Isoforms A and B in Adult and Fetal Human Tissues, and a Potentially Pathological Variant Affecting Only Isoform A in Two Patients with Cornelia de Lange Syndrome

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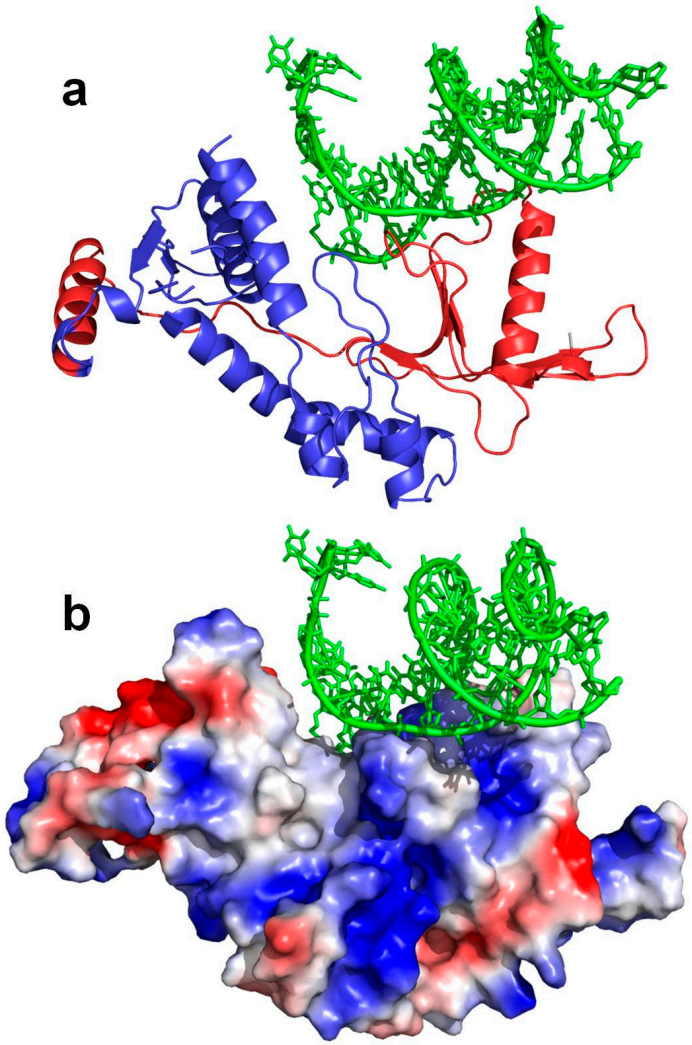

Figure S1. Structural model for the C-terminal domain (residues 2541-2804) of NIPBL isoform A, as obtained from the RaptorX server (http://raptorx.uchicago.edu/). (**a**) 3D model showing secondary structure elements. Blue: residues 2541-2682, shared by both isoforms A and B. Red: residues 2683-2804, present in isoform A and absent in isoform B. Green: putative position of DNA molecule, according to the structure template (Protein Data Bank entry = 3kd1E). Note that most protein-DNA contacts are located in the protein domain absent in NIPBL isoform B. (**b**) Model surface, colored according to electrostatic charge (blue: positive, red: negative), showing positive patches in contact to the negatively charged DNA molecule.