

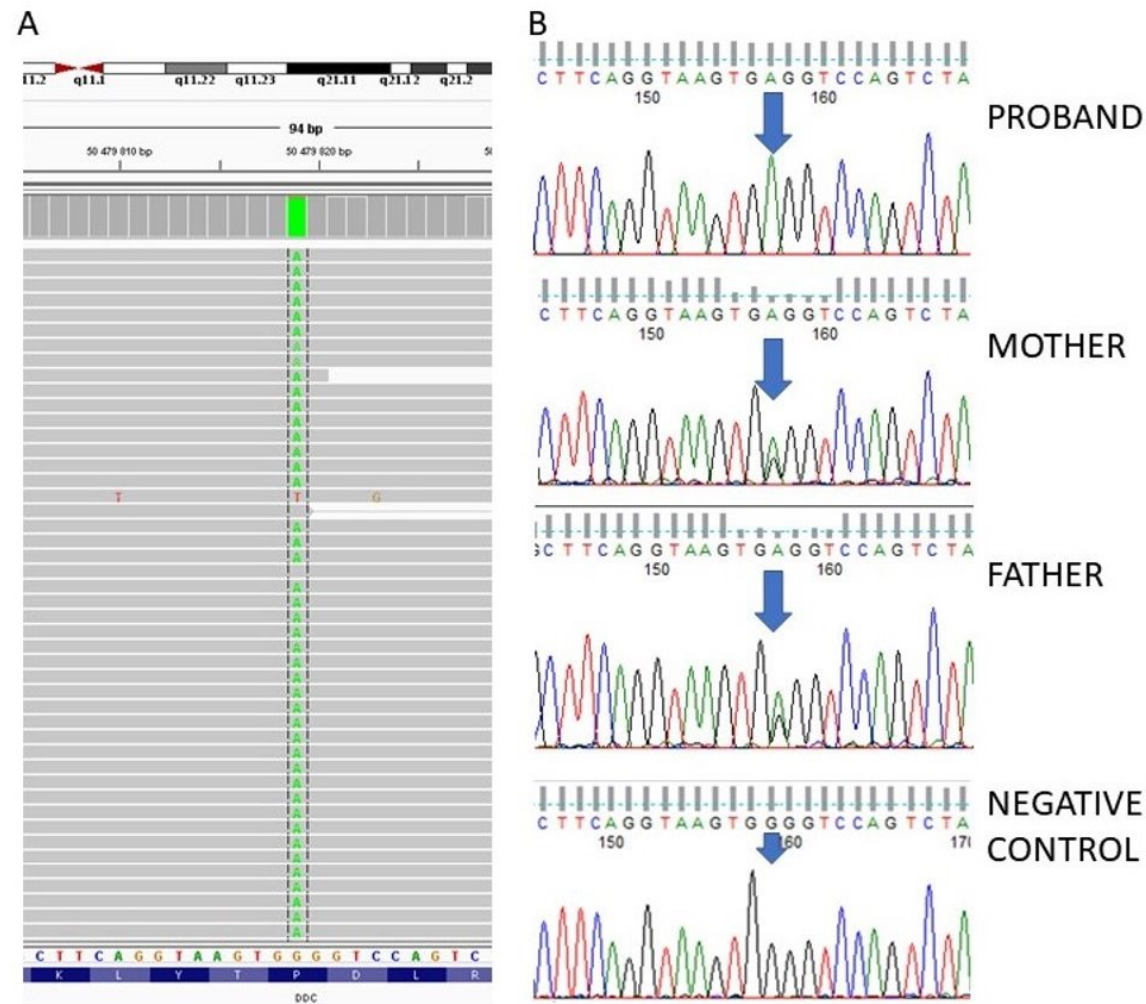


Figure S1- Molecular analysis of the *DDC* gene. A) WES analysis; the view of chromosome 7 region with fragment of *DDC* gene and the variant c.989C>T (p.Pro330L) is shown (the picture generated using IGV, the Integrative Genomic Viewer). Please note that *DDC* gene is in the **reverse** (REV) orientation on the chromosome (3'-5'), therefore G>A substitution is indicated, B) Sanger sequencing of the proband and his parents; the arrow points to the c.989C>T variant, consequently also shown in the reverse orientation.

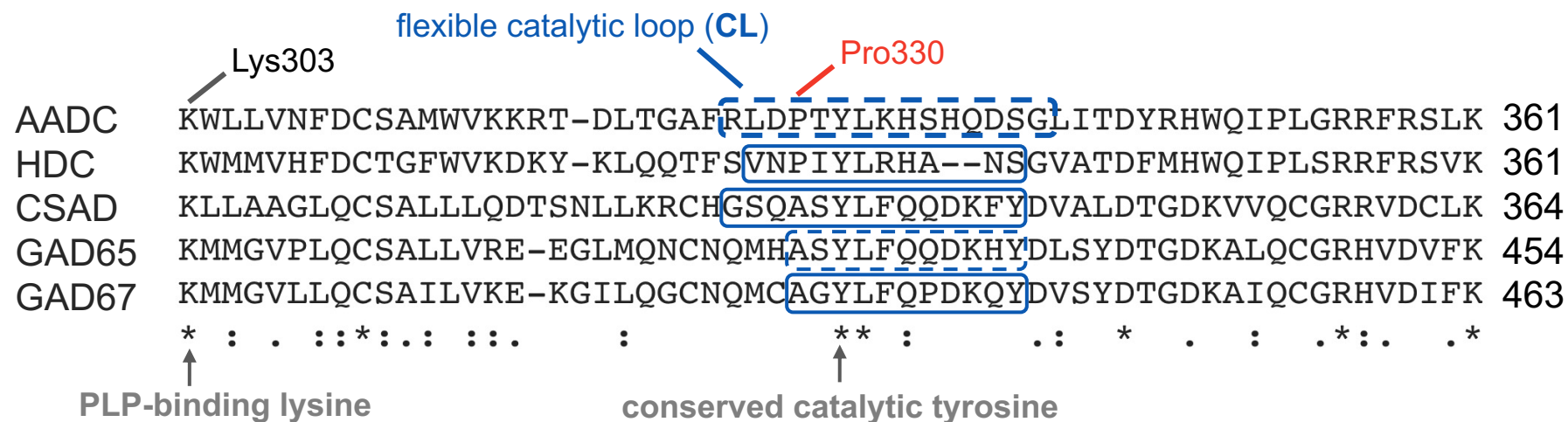


Figure S2. Multiple sequence alignment of the CL of group II PLP-dependent α -decarboxylases. Sequences of hAADC (P20711), hHDC (P19113), hCSAD (Q9Y600), hGAD65 (Q05329), hGAD67 (Q99259) were taken from the UniProt database and aligned with Clustal Omega alignment. Amino acid one letter code is used. Dashes represent insertions and deletions. Invariant positions are indicated with *. Mobile CL is circled with a blue dashed line in correspondence to the part where electron density map is missing in the spatial solved structure. For AADC, conserved PLP-binding Lys303, conserved catalytic Tyr332 and Pro330 are indicated.

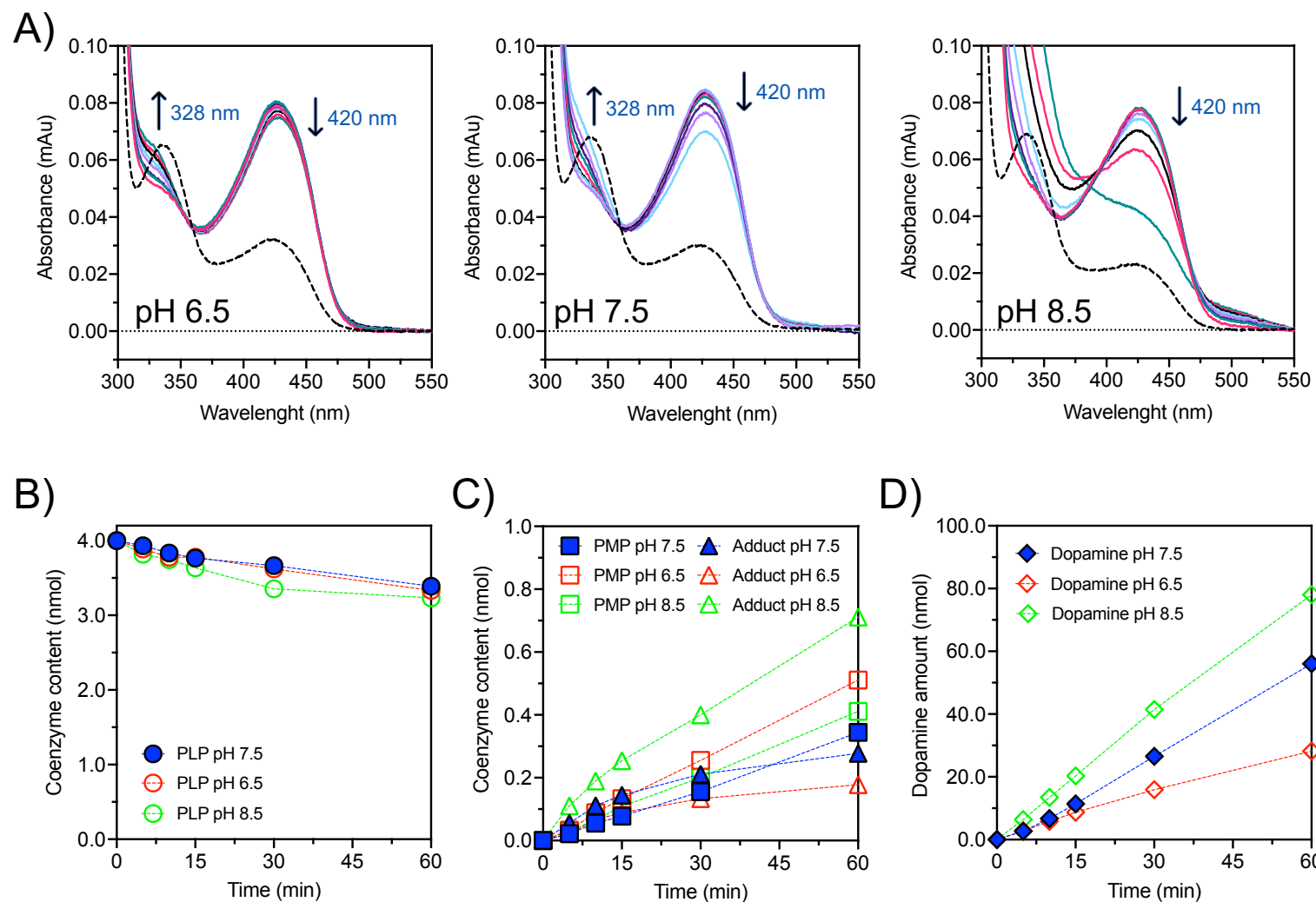


Figure S3. Coenzyme modification during the catalysis of P330L with L-Dopa at different pH values. A) 10 μ M P330L was incubated with 2 mM L-Dopa in 50 mM bis-tris-propane buffer at pH 6.5, 7.5 and 8.5 at 25° C for 1 h and the reaction was analyzed by absorbance changes. At pH 8.5 the oxidation of the L-Dopa is not negligible with time. B) Amount of PLP content followed by HPLC analysis at the indicated pH values, concomitant with C) PMP and PLP-L-Dopa adduct. D) Production of dopamine at the indicated pH values.