



Supplementary Figure S1. Schematic representation of the predicted structure of EGF-L21 domain of the EYS gene. The EGF-L21 domain consists of six cysteine residues, which forms paired disulfide bonds under normal conditions. Mutations altering a cysteine residue into another amino acid might result in unpaired cysteine, thus forcing it to bind other residues, resulting in protein misfolding. The 3D structure of the EGF-L21 (left) was predicted by swissmodel.expasy.com and rendered using YASARA software. The EGF-L21 illustration (right) was created using Biorender.