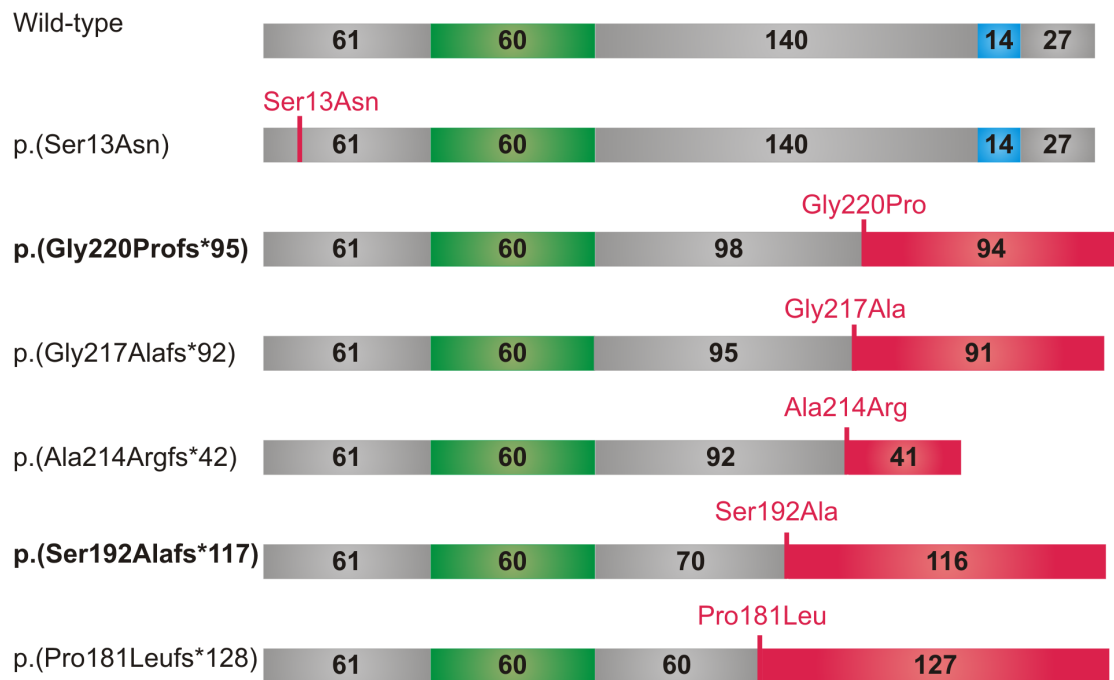


Additional file 2



Additional Figure 1. Schematic representation of all reported mutated PITX3 proteins

The top diagram represents the wild-type PITX3 protein. The green box displays the homeodomain of 60 amino acids and the OAR (named after *otp*, *aristaless* and *rax*) domain of 14 amino acids is displayed by a blue box. The recurrent p.(Gly220Profs*95) and novel p.(Ser192Alafs*117) mutations are indicated in bold. The positions of the mutations are indicated with a red line and a red box displays the resulting aberrant protein segments.