

Appendix 7. SNP (rs 4547091) in OTX2 binding element 3 (-226 to -221) decrease the activity of the human tyrosinase promoter dramatically in the presence of OTX2 and MITF-A. Luc-reporter construct containing the tyrosinase promoter fragments Enh./-2525, Enh./-2525SNP, -462, -462SNP or -462m3 were co-transfected into D407 RPE cells with MITF-A, OTX2, MITF-A/OTX2 factors or with empty expression vector (pCR3). Normalized luciferase activities are relative to tyrosinase promoter construct -152 (=1) co-transfected with empty expression vector pCR3 (not shown). Data are means \pm SEM from two independent transfections performed in triplicate.

