

Appendix 2. Sense primers for the site-directed mutagenesis of MITF and OTX2 transcription factor binding sites.

Mutated binding element	Position relative to transcription start site	Wild type binding site	Sense primer sequences (5' to 3')
MITF site (mM-box)	-104 to -99	<u>CATGTG</u>	gaaaaagtcagt <u>GGATCC</u> cctttcagaggatg
MITF site (mE-box)	-12 to -7	<u>CATGTG</u>	aggtctcagccaaga <u>GGATCC</u> ataatcactgtagta
OTX2 site (m1)	-361 to -356	<u>TAATCT</u>	ctacctaaaccca <u>GaTCT</u> tttaataacttctctaac
OTX2 site (m2)	-354 to -349	<u>TAATAC</u>	ccttaaaccataatctt <u>GGatC</u> cttctctaaactttct
OTX2 site (m3)	-226 to -221	<u>TAATCT</u>	cattattattac <u>GGatc</u> Ctattgttaaatatc
OTX2 site (m1m2)	-361 to -349	<u>TAATCTTTAATAC</u>	accttaaaccca <u>GaTCT</u> tt <u>GGatC</u> cttctctaaactttc
OTX2 site (SNP)	-223 (T/C) SNP rs4547091	<u>TAATCT</u>	cattattattactaa <u>C</u> cttattgttaaatatc

Complementary antisense primers for mutagenesis are not shown. Mutated nucleotides are shown with capital letters and the positions of binding sites are underlined.