

Supplementary

Table S1 The primer information for Sanger sequencing to verify mutations

Patient	Variant	Primer length (bp)	Product size (bp)	T _m (°C)	Primer sequence
1	SEMA3E: Exon9,c.967C>T, p.P323S	23	522	58.1	F:AGTGTCTGGTTCCTTCCTATTGT
		22			R:CACAGGAGGATTCTGGGTA
2	FGFR1:Exon15,c.1981C>T, p.R661X	20	421	55.5	F:GGGACATTCACCACATCGAC
		20			R:GCCTTTCAACATCTGGAGCA
3	KISS1R: Exon5,c.991C>T, p.R331X	20	496	57.3	F:AGCCTTTCGTCTAACCACCT
		18			R:TCAGAGAGGGGCGTTGTC
4	SOX10: Exon4,c.1287delG,p.S431Rfs*69	19	622	56.6	F:CCAAAGCCCAGGTGAAGAC
		20			R:GGCAGTGAGCCAGACAGAAA
5	ANOS1: Exon6,c.844delC,p.R282Vfs*26	20	417	55.5	F:TGTTCCCTGACAGCTCTCTT
		20			R:GCAAAGCCACCTGTTGACTA
6	ANOS1: Exon13,c.1886_1887insCTACTCT, p.R631Sfs*36	20	478	58.1	F:CTAGGCTCTCTGTGCATTGC
		20			R:AAGAAAGGGAGAAAAGCGGG
7	FGFR1:Exon16, c.2117_2118insT, p.E705Gfs*16	20	696	58.7	F:TGCTTTCCTCGGACTGAG
		20			R:GTA
8	FGFR1:Exon17, c.2239_2255delTTCAAGCAGCTGGTGA, p.F745Rfs*45	20	673	60.6	F:ATTTGCAGGTGGTCTTTTCGG
		18			R:CAGTCAGCGGCGTTTGAG
9	CHD7: Exon29, c.5695_5698delGGCC, c.5700_5707delACTTT ACT, p.G1899R,Q1900_W1903del	19	617	56.6	F:AAATGAGGGCACTGAGATG
		20			R:TTGTCCACCTGTTAGAAAGA
10	CHD7:Exon25, c.5355G>T, p.W1785C	21	667	55.5	F:AGCACTGGTGTCTGAGATGA
		20			R:CACTGGTTGGCAGAGTTAGC
11	PROKR2: Exon2, c.667T>G, p.Y223D,c.892C>T, p.R298C	20	862	57.3	F:TGAGGATCACTGTGCCACT
		20			R:TTGATGGTGGGTGATGCTCT
12	KISS1R: Exon2, c.309C>A, p.Y103X	20	549	57.3	F:CGGAGGCGTAATGTTGAGA
		18			R:GTGCATCTCCCTCCGTCC

T_m, Melting Temperature.