

Table S1 Gene mutations of Case1 and Case2 using whole exome sequencing

Case1	Mutation site	Allele frequency	Mutation type	Case2	Mutation site	Allele frequency	Mutation type
Type II (indicate potential clinical significance)							
IDH2 p.R172S	c.516G>T	19.44%	missense mutation	IDH2 p.R172W	c.516G>T	18.87%	missense mutation
PIK3CA p.H1047R	c.3140A>G	25.36%	missense mutation	PIK3CA p.H1047R	c.3140A>G	16.67%	missense mutation
SOX 9 p.Q401*	c.1201C>T	2.11%	nonsense mutation	KMT2D p.Q2816*	c.8446C>T	1.38%	nonsense mutation
				TP53 p.P151S	c.451C>T	0.60%	missense mutation
				FGFR3 p.G380R	c.1138G>A	0.51%	missense mutation
				SPEN p.R844*	c.2530C>T	2.44%	nonsense mutation
				ELAC2 p.Q611*	c.1831C>T	1.46%	nonsense mutation
				FGFR2 p.M538I	c.1614G>A	1.07%	missense mutation
				ADGRA2 p.W281*	c.843G>A	1.06%	nonsense mutation
				SMARCA4	c.4170+1G>A	1.04%	shear mutation
				ERBB2 p.R487W	c.1459C>T	0.81%	missense mutation
Type III (unknown clinical significance)							
KDM5C p.E439D	c.1317G>T	15.13%	missense mutation	PIK3CG p.D946N	c.2836G>A	3.59%	missense mutation
BRAF	c.627T>C	15%	synonymous mutation	NCOA2 p.G1306R	c.3916G>A	3.18%	missense mutation
ERCC3 p.K528R	c.1583A>G	4.92%	missense mutation				

Table S2 Clinical history, immunohistochemistry, gene mutation and follow-up information of the patients

Case#	Author[ref.]	Clinical History			Immunohistochemistry								Gene Mutation		Follow-up
		Sex/age	Size	Treatment	TTF-1	Calretinin	GATA-3	SOX-10	CK5/6	ER	PR	Her-2	IDH2	PIK3CA	
1-5	Eusebi <i>et al.</i> (1)	F/58	1.2	W	-	ND	ND	ND	ND	-	-	ND	ND	ND	A&W 26
		F/70	1.3	M	-	ND	ND	ND	ND	-	-	ND	ND	ND	A&W 54
		F/57	1.6	W	-	ND	ND	ND	ND	-	-	ND	ND	ND	A&W 28
		F/74	2	W	-	ND	ND	ND	ND	-	-	ND	ND	ND	A&W 108
		F/56	0.8	UK	ND	ND	ND	ND	ND	ND	ND	ND	ND	ND	ND
6	Cameselle-Teijeiro <i>et al.</i> (4)	F/64	4.1	M+AL+ C/XRT/HT	-	ND	ND	ND	ND	+	+	-	ND	ND	Alive with bone metastasis
7-10	Tosi <i>et al.</i> (5)	F/80	2.5	Q	ND	ND	ND	ND	ND	-	-	ND	ND	ND	A&W 3
		F/45	5	Q	ND	ND	ND	ND	ND	+	+	ND	ND	ND	A&W 5
		F/61	2	Q	ND	ND	ND	ND	ND	+	-	ND	ND	ND	A&W 8
		F/47	2.3	Q	ND	ND	ND	ND	ND	+	+	ND	ND	ND	A&W 10
11	Chang <i>et al.</i> (6)	F/66	1.1	S+SLN	-	ND	ND	ND	ND	+	-	-	ND	ND	A&W 12
12	Masood <i>et al.</i> (2)	F/57	3.7	M	-	ND	ND	ND	ND	+	+	-	ND	ND	UK
13	Colella <i>et al.</i> (7)	F/79	3	M+AL	-	ND	ND	ND	ND	-	-	-	ND	ND	A&W 18
14-26	Chiang <i>et al.</i> (3)	F/68	0.9	UK	-	ND	ND	ND	+	-	-	ND	-	+	UK
		F/52	0.8	UK/XRT	-	ND	ND	ND	+	-	-	-	+	+	A&W 77
		F/63	1.2	UK	ND	ND	ND	ND	+	+	+	ND	+	+	UK
		F/79	UK	UK	-	ND	ND	ND	+	-	-	ND	+	+	UK
		F/64	1.8	UK	-	ND	ND	ND	+	-	-	-	+	+	A&W 31
		F/51	0.8	UK	-	ND	ND	ND	+	+	+	-	-	+	A&W 30
		F/64	1.4	UK/XRT	-	ND	ND	ND	+	+	-	-	+	-	A&W 29
		F/58	0.6	UK	-	ND	ND	ND	+	-	-	-	+	+	UK
		F/66	0.9	UK/C	-	ND	ND	ND	+	-	-	-	+	+	A&W 20
		F/65	1.5	UK/XRT/C	-	ND	ND	ND	+	+	-	-	+	+	A&W 37
		F/70	1.3	UK	-	ND	ND	ND	+	+	-	-	-	+	A&W 12
		F/65	1.2	UK	-	ND	ND	ND	+	-	-	-	+	-	UK
		F/65	0.9	UK	-	ND	ND	ND	+	-	-	-	+	+	UK
27-39	Foschini <i>et al.</i> (9)	F/58	1.2	W+SLN	-	ND	+/-	ND	+	-	-	-	+	-	Recurrence 60
		F/80	2.5	W	-	ND	+/-	ND	+	-	-	-	ND	ND	A&W 120
		F/61	2	W+SLN	-	ND	-	ND	-	+	-	-	ND	ND	A&W 132
		F/62	1	W	-	ND	ND	ND	ND	-	-	-	ND	ND	A&W 96
		F/51	2	W	-	ND	ND	ND	-	-	-	-	ND	ND	UK
		F/58	0.8	W+SLN	-	ND	ND	ND	ND	+	+	-	ND	ND	A&W 24
		F/61	0.6	W	-	ND	ND	ND	+	-	-	-	ND	ND	A&W 124
		F/50	0.8	W/XRT/C	-	ND	+/-	ND	-	-	-	-	ND	ND	A&W 84
		F/59	2.5	W	-	ND	-	ND	+	-	-	-	+	+	A&W 76
		F/48	2.2	W	-	ND	ND	ND	+	-	-	-	ND	ND	A&W 24
		F/85	1.5	W	-	ND	ND	ND	+	-	-	-	+	+	UK
		F/64	2	W+ AL	-	ND	+/-	ND	ND	-	-	-	ND	ND	A&W 36
		F/77	1.2	W	-	ND	+/-	ND	+	+	+	-	+	+	A&W 24
40-42	Bhargava <i>et al.</i> (8)	F/65	0.9	UK	ND	ND	ND	ND	+	-	-	ND	+	-	A&W 19
		F/77	1.7	UK	-	ND	ND	ND	+	+	+	ND	-	-	UK
		F/48	1.2	UK+NAC	ND	ND	ND	ND	+	+	ND	ND	+	+	A&W 19
43	Gai <i>et al.</i> (11)	F/55	UK	M+SLN	-	ND	+	ND	+	-	-	-	ND	ND	UK
44-46	Lozada <i>et al.</i> (12)	F/60	1.1	UK+SLN	ND	ND	ND	ND	+	-	-	-	+	+	UK
		F/60	2.1	UK	ND	ND	ND	ND	+	-	-	-	+	+	UK
		F/67	0.6	UK	ND	ND	ND	ND	+	-	-	-	+	-	UK
47-54	Zhong <i>et al.</i> (13)	F/63	0.6	UK	ND	ND	ND	ND	ND	-	-	-	+	+	UK
		F/63	1	UK	-	ND	ND	ND	ND	-	-	-	+	-	UK
		F/79	1.6	UK	-	ND	ND	ND	+	ND	ND	ND	+	+	UK
		F/69	1.2	UK	ND	ND	ND	ND	ND	+	-	+	+	+	UK
		F/69	1.8	UK	-	ND	+	ND	ND	-	-	-	+	+	UK
		F/71	1.7	UK	-	ND	+	ND	ND	-	-	-	+	+	UK
		F/74	0.8	UK	ND	ND	ND	ND	ND	-	-	ND	ND	ND	UK
F/76	0.7	UK	ND	ND	ND	ND	ND	+	+	ND	+	+	UK		
55-63	Alsadoun <i>et al.</i> (10)	F/63	1.6	S	ND	+	ND	ND	-	+	+	-	-	ND	UK
		F/70	1.1	S	ND	+	ND	ND	-	-	-	-	+	ND	UK
		F/72	1.0	S	ND	+	ND	ND	-	+	-	-	+	ND	UK
		F/64	1.5	S	ND	+	ND	ND	-	-	-	-	+	ND	UK
		F/71	0.9	S	ND	+	ND	ND	-	-	-	-	+	ND	UK
		F/52	4.0	S	ND	+	ND	ND	-	+	+	-	+	ND	UK
		F/69	1.0	S+SLN	ND	+	ND	ND	+	-	-	-	+	ND	A&W 53
F/57	1.2	S	ND	+	ND	ND	-	+	-	-	+	ND	UK		
F/75	3.0	S	ND	+	ND	ND	-	+	+	-	-	ND	UK		
64	Haefliger <i>et al.</i> (14)	F/60	0.8	UK+SLN	-	ND	ND	ND	+	-	-	-	+	+	A&W 8
65	Jassim <i>et al.</i> (15)	F/40	5.5	M+AL	-	+	+	ND	+	+	+	-	+	ND	A&W 6
66-74	Pareja <i>et al.</i> (16)	F/67	1	UK	ND	+	ND	ND	+	-	ND	ND	+	-	UK
		F/59	1.5	Partial M/XRT	ND	+	ND	ND	ND	+	ND	ND	+	-	UK
		F/64	1.2	Partial M/XRT	ND	+	ND	ND	+	+	ND	ND	+	-	UK
		F/70	1.3	UK	ND	ND	ND	ND	+	-	ND	ND	+	+	UK
		F/67	1.7	UK	ND	+	ND	ND	+	-	ND	ND	+	+	UK
		F/60	2.6	UK	ND	+	ND	ND	+	+	ND	ND	+	+	UK
		F/47	1.3	UK	ND	+	ND	ND	+	-	ND	ND	+	-	UK
		F/80	0.6	UK	ND	+	ND	ND	+	+	ND	ND	+	-	UK
F/46	0.6	UK	ND	+	ND	ND	+	-	ND	ND	+	-	UK		
75	Zhang <i>et al.</i> (17)	F/45	1.0	W+SLN +C/XRT	-	ND	+	ND	+	-	-	-	+	+	A&W 12
Present 76-77	Wei <i>et al.</i>	F/72	2.2	M+SLN	+/-	+	+/-	+	+	-	-	-	+	+	A&W 6
		F/70	1.6	M	-	+	+	+	+	+	-	-	-	+	+
Sum		F/40-80 (64)	0.6-5.5 (1.4)	W 17/43(39.5%); S 10/43(23.3%); Q 4/43(9.3%); M 6/43(14%)	- 43/44; +/- 1/44	+ 19/19	+/- 6/14; + 6/14; - 2/14	+ 2/2	+ 42/53	TN33 33/50(66%)	ER 33/50(66%)	PR 33/50(66%)	Her-2 33/50(66%)	IDH2 46/52(88.5%); PIK3CA 30/42(71.4%); Both 26/42(61.9%)	3-132 (29) Local recurrence1, Bone metastasis 1

F, female; UK, unknown; AL, axillary lymph node dissection; W, wide excision; Q, quadrantectomy; S, segmental resection; M, mastectomy; SLN, sentinel lymph node excision; C, chemotherapy; XRT, irradiation; HT, hormonal treatment; ER, estrogen receptor; PR, progesterone receptor; AR, androgen receptor; HER-2, human epidermal growth factor receptor 2; CK, cytokeratin; TTF-1, thyroid transcription factor-1; +/-, focal positive; +, positive; -, negative; ND, not done; A&W, alive and well.