

琉球大学学術リポジトリ

SIW/SNF遺伝子異常の食道扁平上皮がん発がんの早期における誘発

メタデータ	言語: 出版者: 琉球大学 公開日: 2017-05-11 キーワード (Ja): キーワード (En): Epigenetics, SWI/SNF, mutation, ESCC 作成者: 仲里, 秀次, Nakazato, Hidetsugu メールアドレス: 所属:
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Table 2. Somatic mutations detected in the 92 ESCCs.

Case	Gene	Read coverage	Mutant allele frequency (%)	Nucleotide change	Amino acid change
20	<i>PBRM1</i>	439	70.8	c.239A>G	Asn80Ser
85	<i>ATRX</i>	1191	16.3	c.277G>A	Asp93Asn
89	<i>PBRM1</i>	403	64.3	c.4129G>A	Glu1377Lys
	<i>PBRM1</i>	1427	67.4	c.3883G>C	Glu1295Gln
	<i>PBRM1</i>	567	64.4	c.3778G>C	Asp1260His
94	<i>ARID2</i>	765	10.6	c.1925G>T	Gly642Val
126	<i>ARID1A</i>	669	80	c.2017C>T	Gln673*
127	<i>SMARCA4</i>	504	26	c.2644G>A	Glu882Lys
169	<i>SMARCA4</i>	838	29	c.254C>T	Ser85Leu
176	<i>SMARCAL1</i> [#]	3384	58	c.1129G>C	Glu377Gln
	<i>SMARCC1</i> [#]	3795	47.8	c.3095G>A	Arg1032His

Termination codon is shown by *. Novel mutations in ESCCs are marked by #.