

GeneWell社 遺伝子検査標準サンプル Spec Sheet

商品名： OncoSight 800 gDNA Reference Standard
 型式： IB-GW-OGTM800
 品番： 89-8610-65
 形態： gDNA
 濃度： 30ng/μL、1μg/tube
 Buffer： Tris-EDTA (10mM Tris-HCl, 1mM EDTA), pH 8.0
 保存温度： 2-8°C
 使用期限： 18 months

変異リスト

Number	Variant Type	Mutation	CDS Mutation	Expected Allelic Frequency(%) / Copy Number	COSMIC ID	Chromosome Number	Genomosome Position
1	SNV	EGFR L858R	2573T>G	1%	COSM6224	7	55191822
2	SNV	KRAS A146T	c.436G>A	1%	COSM19404	12	25225628
3	SNV	NRAS Q61K	c.181C>A	1%	COSM580	1	114713909
4	SNV	EGFR T790M	2369C>T	2%	COSM6240	7	55187178
5	Deletion	EGFR ΔE746-A750	c.2235_2249del15	2%	COSM6223	7	55174772-55174786
6	Deletion	FLT3 ΔI836	2506_2508delATC	2%	COSM797	13	28018500-28018502
7	SNV	KIT D816V	2447A>T	2%	COSM1314	4	54733155
8	SNV	KRAS G12D	c.35G>A	2%	COSM1314	4	54733155
9	Insertion	EGFR V769_D770 insASV	c.2307_2308insGCCAGCGTG	3%	COSM12376	7	55181317-55181318
10	SNV	EGFR G719S	c.2155G>A	4%	COSM6252	7	55174014
11	SNV	KRAS G13D	c.38G>A	4%	COSM532	12	25245347
12	Fusion	EML4-ALK Fusion	ML4:r.1_929_ALK:r.4080_6220 EML4{ENST00000318522} ; ALK{ENST00000389048}	5%	COSF734	2,2	42472827, 29446394
13	Fusion	CD74-ROS1 Fusion	CD74:r.1_627_ROS1:r.5757_7368 CD74{ENST00000009530} ; ROS1{NM_002944}	6%	COSF1201	6,5	117700222, 149782188
14	SNV	BRAF V600E	c.1799T>A	7%	COSM476	7	140753336
15	SNV	PIK3CA H1047R	c.3140A>G	7%	COSM775	7	140753336
16	CNV	ERBB2 Amplification	NA	5copies	NA	17	39687914-39730426