



Personalized Healthcare Summit 2013

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MASSACHUSETTS
GENERAL HOSPITAL

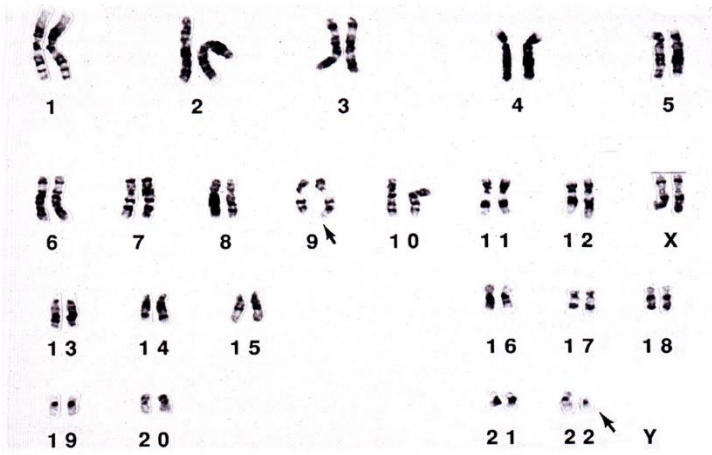
CANCER CENTER



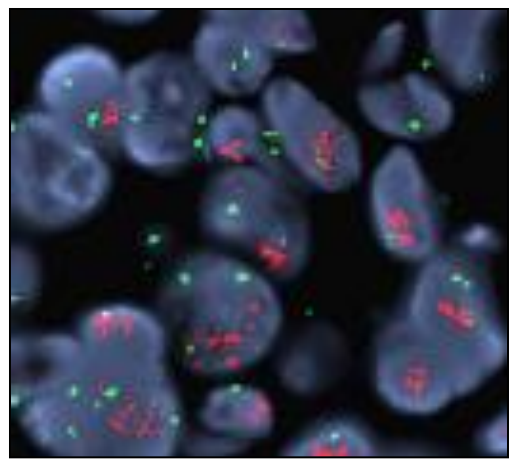
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PATHOLOGY

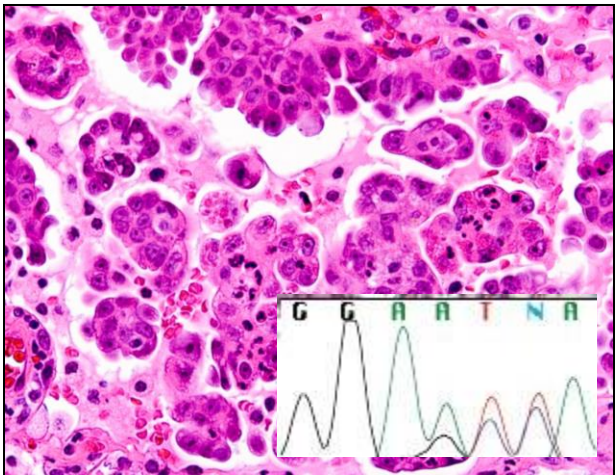
BCR-ABL Imatinib
100% CML



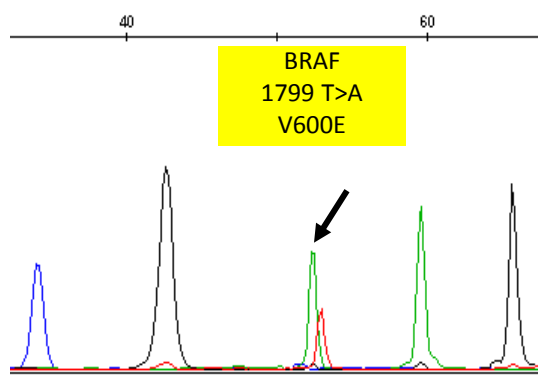
HER2 Trastuzumab
20-30% IDC



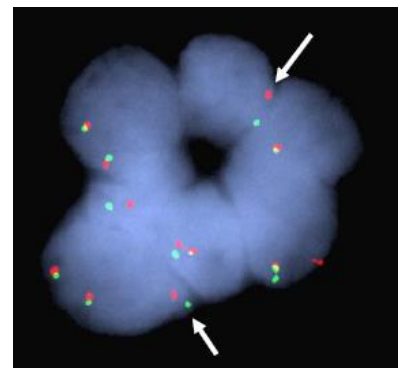
EGFR Erlotinib/ Gefitinib
20% Lung adenocarcinomas



BRAF V600E
Vemurafenib
50-60% Melanoma



ALK Crizotinib
3-5% Lung adenocarcinoma



Crizotinib : Benefits of Real-time Screening

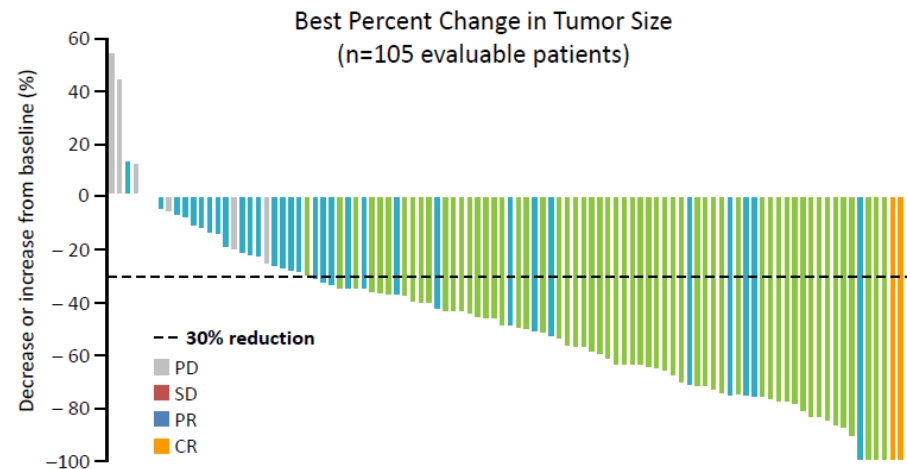
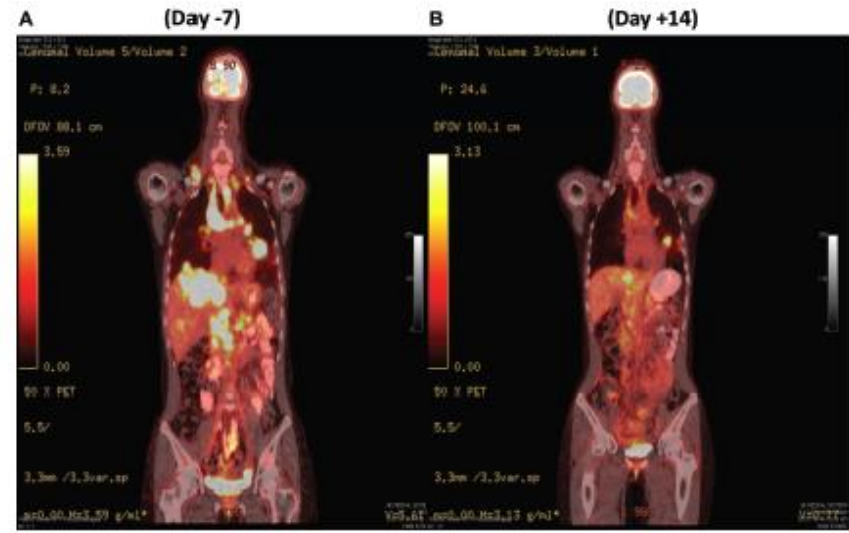
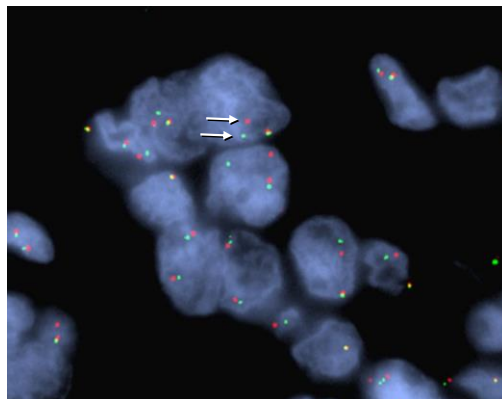
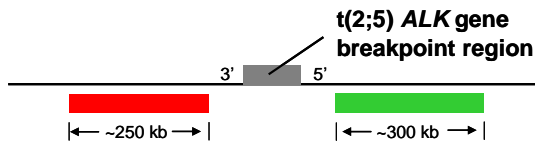
Crizotinib: Potent & selective ATP competitive oral inhibitor of MET, ALK, and ROS1

Vol 448 | 2 August 2007 | doi:10.1038/nature05945 nature

Identification of the transforming *EML4-ALK* fusion gene in non-small-cell lung cancer

Manabu Soda^{1,2}, Young Lim Choi¹, Munehiro Enomoto^{1,2}, Shuji Takada¹, Yoshihiro Yamashita¹, Shunpei Ishikawa³, Shin-ichiro Fujiwara¹, Hideki Watanabe¹, Kentaro Kurashina¹, Hisashi Hatanaka¹, Masashi Bando², Shoji Ohno², Yuichi Ishikawa⁴, Hiroyuki Aburatani^{5,7}, Toshiro Niki³, Yasunori Sohara⁴, Yukihiro Sugiyama² & Hiroyuki Mano^{1,7}

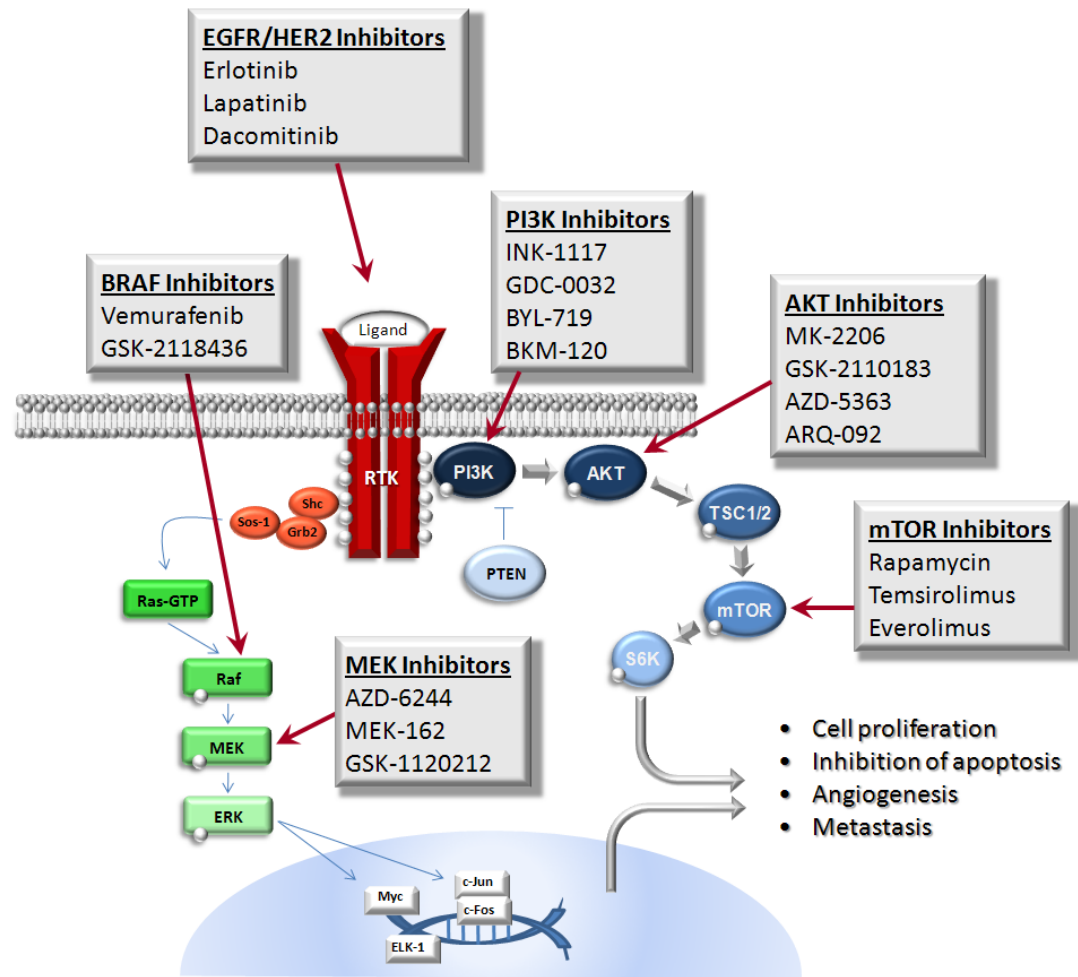
Telomere 2p23 region Centromere



SNAPSHOT Genotyping Assay

24 cancer genes – 160 described mutations

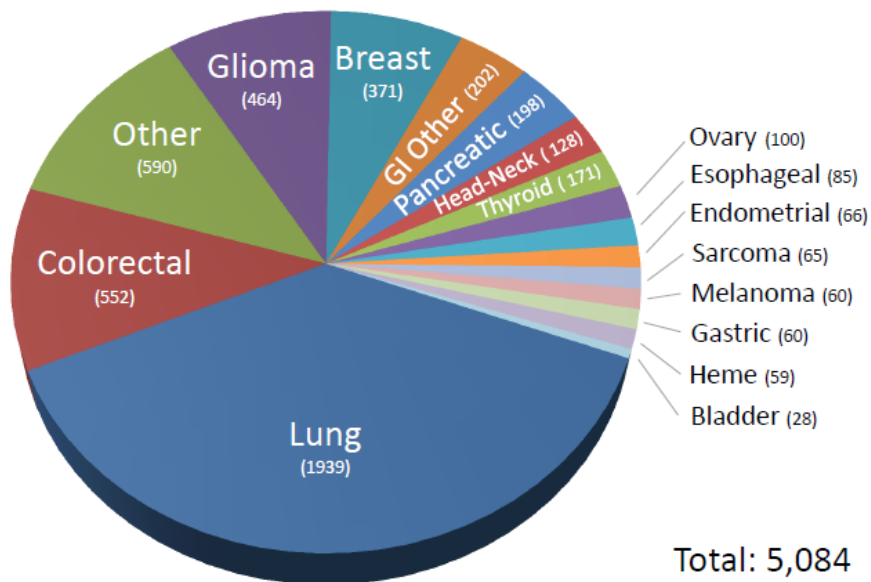
Gene	Amino Acid - cDNA Residue	Gene	Amino Acid - cDNA Residue
AKT1	E17 - 49G	KRAS	G12 - 34G G12 - 35G G13 - 37G G13 - 38G G61 - 181C G61 - 182A G61 - 183A
APC	R1114 - 3340C Q1338 - 4012C R1450 - 4348C T1556fs*3 - 4666_4667insA	MAP2K1	Q56 - 167A K57 - 171G D67 - 199G
BRAF	G466 - 1397G G469 - 1406G L597 - 1789C V600 - 1798G V600 - 1799T	NOTCH1	L1575 - 4724T L1601 - 4802T
CTNNB1	D32 - 94G D32 - 95A S33 - 98C G34 - 101G S37 - 109T S37 - 110C T41 - 121A T41 - 122C S45 - 133T S45 - 134C	NRAS	G12 - 34G G12 - 35G G13 - 37G G13 - 38G Q61 - 181C Q61 - 182A Q61 - 183A
EGFR	G719 - 2155G G719 - 2156G E746_A750del - 2235_2249del15 E746_A750del - 2236_2250del15 T790 - 2369C L858 - 2573T L861 - 2582T Exon 19 deletions Exon 20 insertions	PIK3CA	R88 - 263G E542 - 1624G E545 - 1633G Q546 - 1636C Q546 - 1637A H1047 - 3139C H1047 - 3140A G1049 - 3145G
ERBB2	Exon 20 insertions	PTEN	R130 - 388C R173 - 517C R233 - 697C K267fs*9 - 800delA
IDH1	R132 - 394C R132 - 395G	TP53	R175 - 524G G245 - 733G R248 - 742C R248 - 743G R273 - 817C R273 - 818G R306* - 916C
KIT	D816 - 2447A Exon 11 deletions/insertions		



SNAPSHOT Genotyping Assay

24 cancer genes – 160 described mutations

Number of Tumors Tested



Lung Cancer Profiles

