

PROGRAMA ANDALUZ DE FORMACIÓN EN
MEDICINA PERSONALIZADA Y DE PRECISIÓN (PANMEP)

DIAGNÓSTICO MOLECULAR

COLABORA
Johnson & Johnson

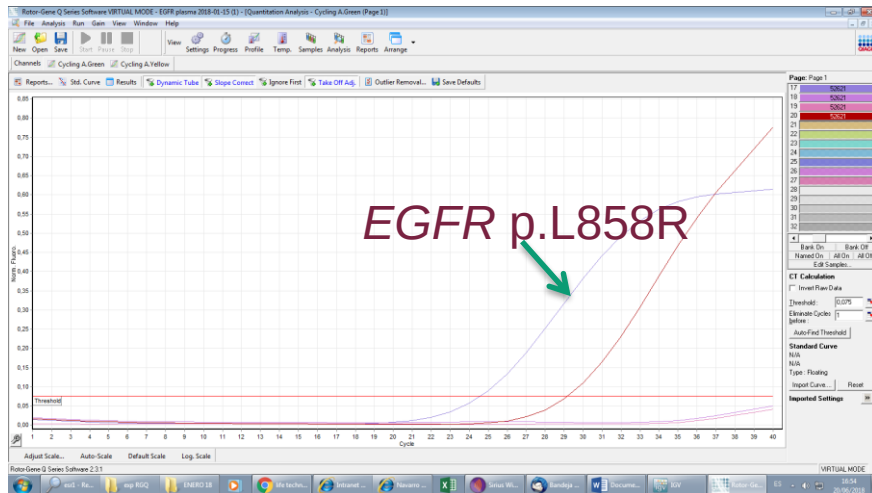
ORGANIZA

A
Junta de Andalucía
Consejería de Salud y Familias

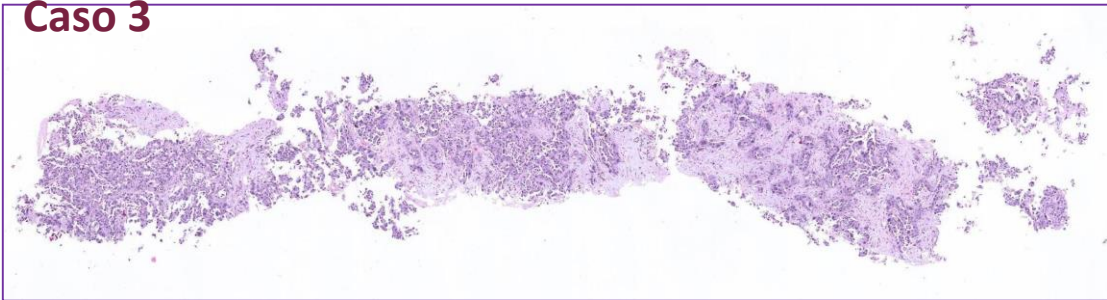


Caso 3

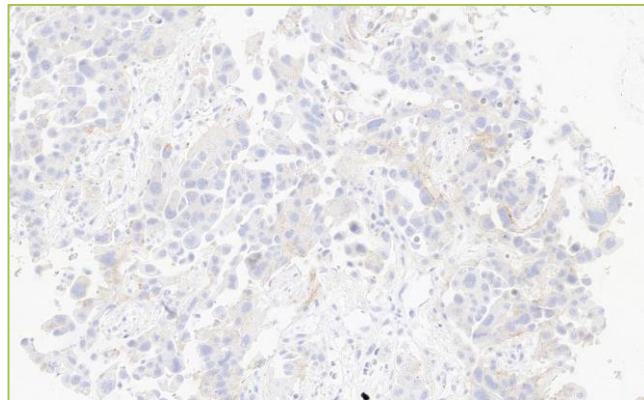
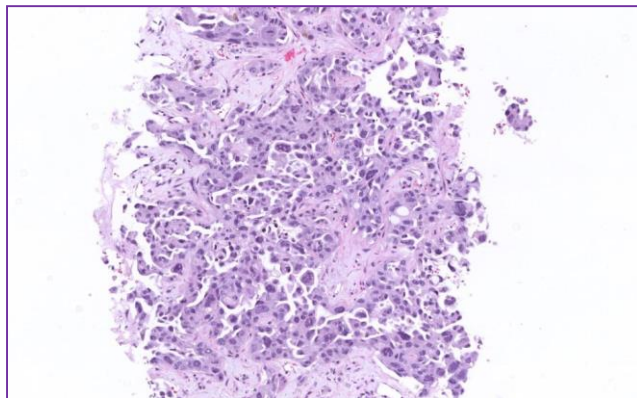
- Hombre 59 años
- Enero 2018 : UCI - disnea
- TAC: lesion 7cm lobulo pulmonar superior derecho - se realiza biopsia
- Biopsia líquida



Caso 3



PD-L1
FISH ALK
FISH ROS1
FISH MET
EGFR
ONCOMINE



PD-L1 40 % TC (28-8
clone)

% células tumorales para estudios
moleculares : 90 %.

Caso 3

OncoPrint™ Solid Tumour DNA Kit (Thermo Fisher)

Analysis Results Download Visualize Selected Variants

Analysis Name

Summary Functional Population Ontologies Pharmacogenomics Somatic QC

Search Go Preferences

		Classification	Type	Locus	Genes	Amino Acid Change	Coding	Coverage	% Frequency
☐	☐	Unclassified	SNV	chr17:7578457	TP53	p.Arg158Leu	c.473G>T	3987	AG=70.86, GG=0.00, TG=0.00, C=0.00
☐	☐	Unclassified	SNV	chr7:55259515	EGFR	p.Leu858Arg	c.2573T>G	3984	GG=37.37, GT=0.00

OncoPrint™ DNA Focus Assay (Thermo Fisher)

Analysis Results Download Visualize Selected Variants

Analysis Name: 52653_v2_c1061_2018-05-16-11-55-726 MAPD: 0.337

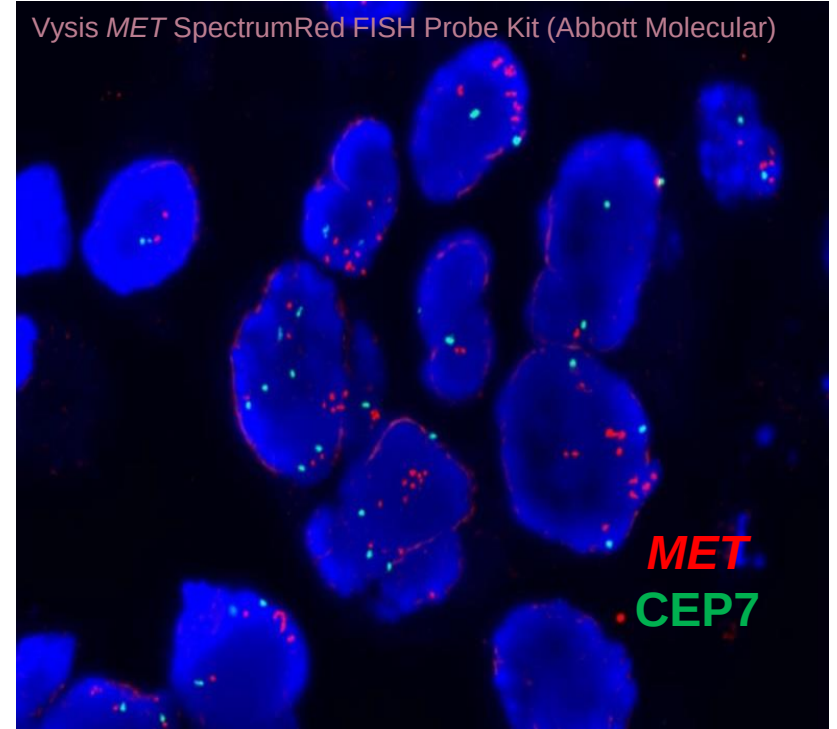
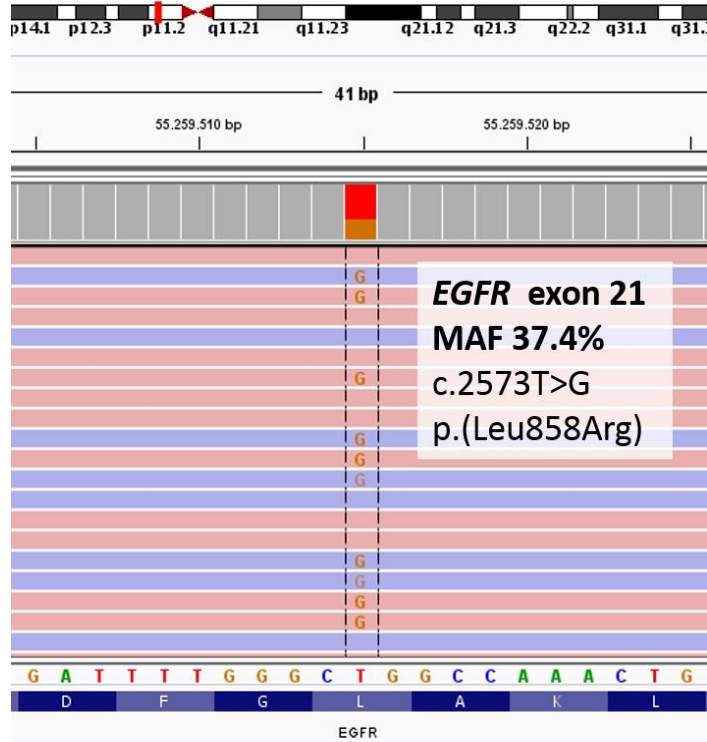
Summary **OncoPrint** Functional Population Ontologies Pharmacogenomics Somatic QC

Search Go Preferences

		Locus	OncoPrint Variant Class	OncoPrint Gene Class	Genes	Amino Acid Change	Copy Number
☐	☐	chr7:55259514	Hotspot	Gain-of-function	EGFR	p.Leu858Arg	
☐	☐	chr7:116313479	Amplification	Gain-of-function	MET		5.67

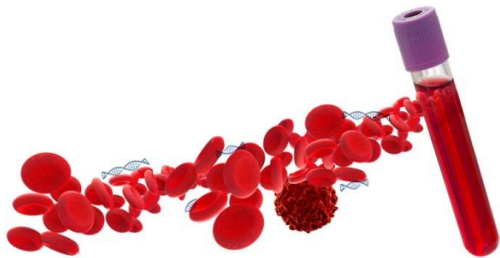
Caso 3

Biopsia



Caso 3

SAMPLE ID 18M355	ANALYSIS START DATE Sep 12, 2018 13:26:06 UTC
ANALYSIS WORKFLOW DefaultExpandedPanelWorkflow	PANEL Expanded
ADAPTER SA 1	FILTERSET DefaultFilterSet*
ISOLATED DNA MASS 50.00 ng	SAMPLE VOLUME 4.50 mL



Results Summary

SNVS (3)				
Gene	Variant	Variant Description	Allele Fraction	No. of Mutant Molecules per mL
TP53	p.Arg158Leu p.Arg147Leu	Missense variant Missense variant	3.61%	133
EGFR	p.Leu858Arg	Missense variant	3.02%	111
BRAF	p.Leu597Gln	Missense variant	0.05%	1.95

INDELS (0)

CNV AMPLIFICATIONS (1)

Gene
MET

/CENTRO DE SIMULACIÓN **CLÍNICA AVANZADA** **IAVANTE**

Gracias por su atención

www.IAVANTE.es

