

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

DIAGNÓSTICO MOLECULAR

COLABORA
Johnson&Johnson

ORGANIZA

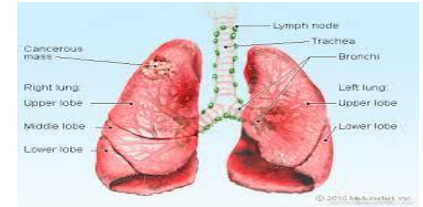
A
Junta de Andalucía
Consejería de Salud y Familias



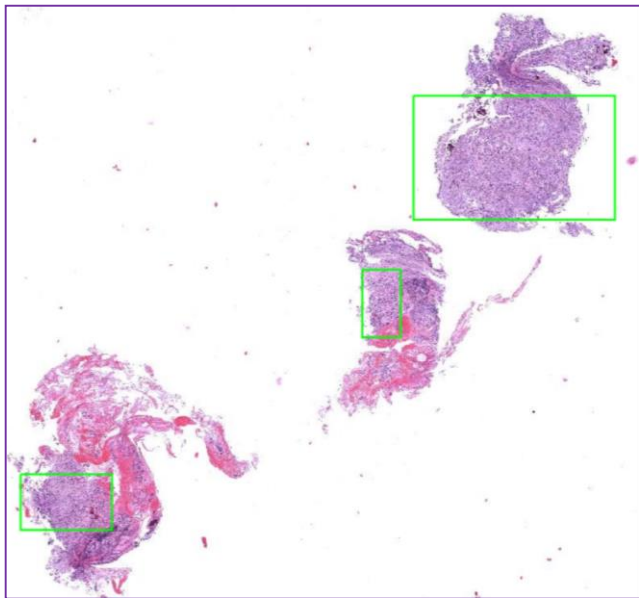
Caso 4

- Mujer de 58 años
- Diagnosticada en Abril 2018 de adenocarcinoma de pulmón

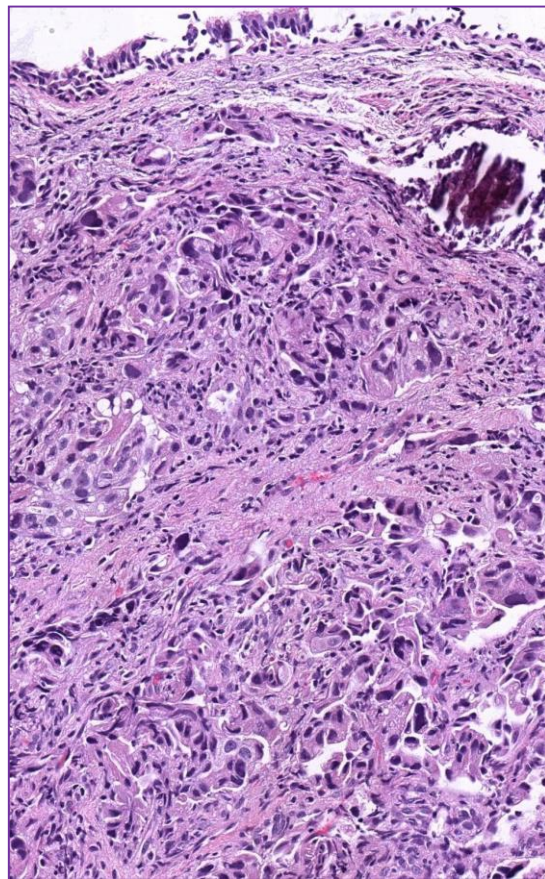
Lung cancer



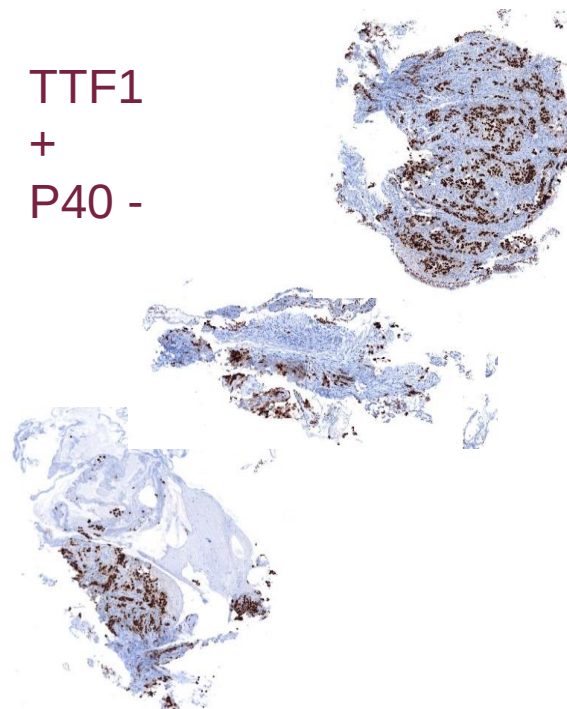
Caso 4



NSLSC, probable
adenocarcinoma después de IHQ



TTF1
+
P40 -



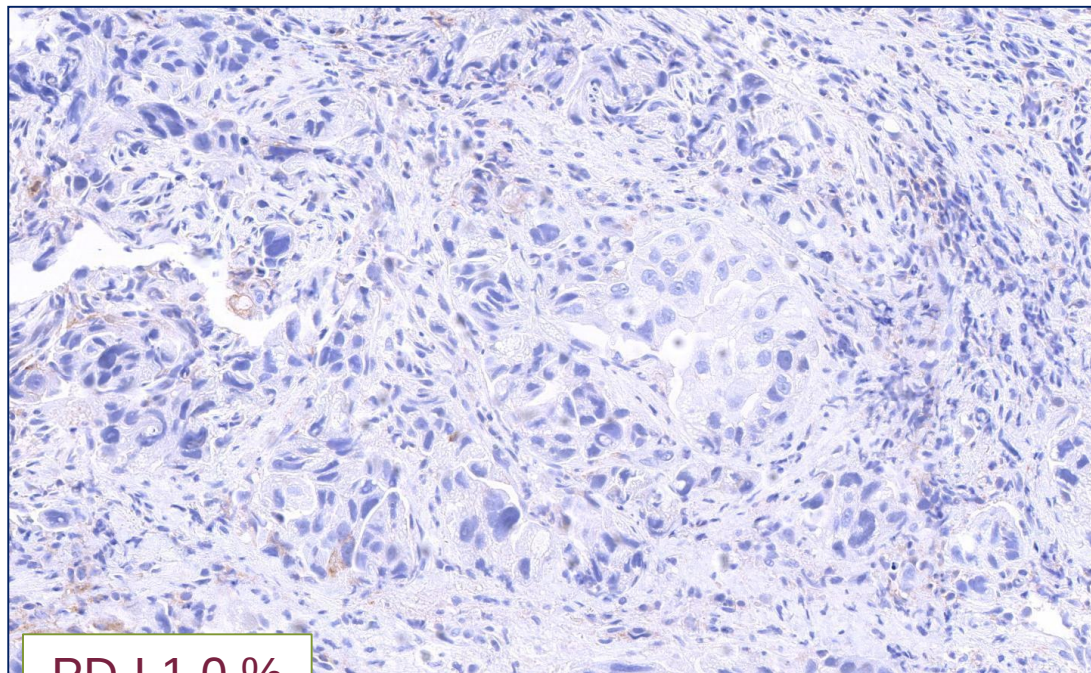
Porcentaje células tumorales:
30 %

Caso 4

PD-L1
FISH ALK
FISH ROS1
FISH MET
ONCOMINE



NEG



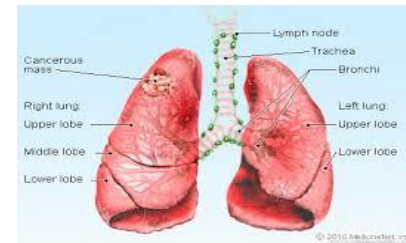
PD-L1 0 %

IHQ for PD-L1 (Ab 28-8, Dako, automatized Link 48 platform)
Resultado : NEGATIVO (0 % células tumorales)

Caso 4

- Mujer de 58 años
- Diagnosticada en Abril 2018 de adenocarcinoma de pulmón
- Sin biomarcadores positivos para terapia dirigida(EGFR-ALK-ROS1)
- Expresión PDL1 : 0%
- Quimioterapia Estandar

Lung cancer



Caso 4

- Mujer de 58 años
- Diagnosticada en Abril 2018 de adenocarcinoma de pulmón
- Quimioterapia estandar
- Progresion clínica en Agosto 2018



Screening NGS en plasma (biopsia líquida)

Oncomine Lung Cell free Total Nucleic Acid Assay (ADN+ARN circulante)

Caso 4

Detecta SNVs, indels + FUSIONS

Assay	Configuration	SNVs	CNVs	TP53 coverage	Fusion (RNA)	Exon Skipping (RNA)
Lung cfTNA	DNA + RNA	36 amplicons	10 (target CNVs) + 12 (ref CNVs)		49 assays	3 assays

	Hotspot Genes	Full-Length Genes	Copy Number Genes	Gene Fusions	Exon Skipping
Lung cfTNA	ALK BRAF EGFR ERBB2 KRAS MAP2K1 MET NRAS PIK3CA ROS1 TP53	-	MET	ALK RET ROS1	MET

Oncomine Lung cell-free Total Nucleic Acid Assay Result

Ion Reporter

Hi, Beatriz Bellosillo 2.7 TB/15 TB Help Sign Out

Home

Samples

Analyses

Workflows

Overview

Launch

My Variants

Analysis Visualization

cDNA IRGV

Summary

SNV / Indel

CNV

Fusion

Analysis Name : Sample Name/Barcode Id

51902_v1_c1147_2017-11-27-13-35-551 : 51902_v1/IonCodeTag_0105

Gene	MAF %
TP53	4.738
TP53	0.058

20 items per page

Fusion

Variant (exons)

Mol Cov.
Mutant

QC Test

KIF5B-
RET.K15R12.COSF1232

16



1 - 1 of 1 items

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Generate Report

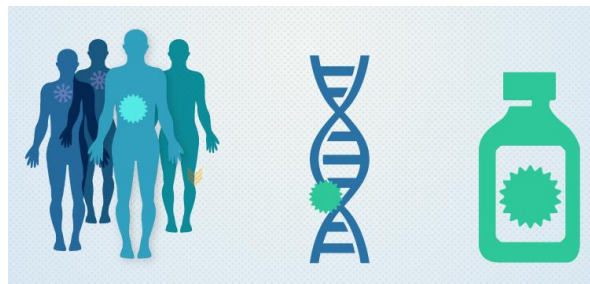
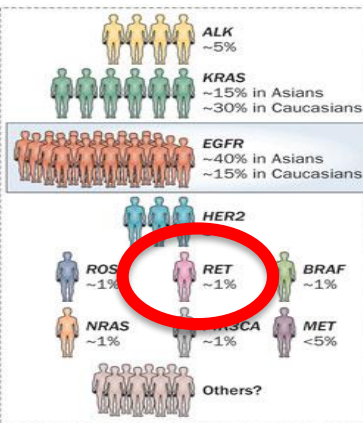
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[Selected Analyses](#)

Variant (exons)	Mol Cov. Mutant	QC Test
KIF5B-RET.K15R12.COSF1232	16	✓

1 - 1 of 1 items

Heterogeneity in patients with adenocarcinoma of the lung according to driver oncogenes



Caso 4



Oncomine solid tumor CE-IVD

Ion Reporter Hi, Beatriz Bellosillo 2.7 TB/15 TB Help Sign Out

Home Samples Analyses Workflows

Overview Launch My Variants

IR Chg • Ion Reporter 5.6

Analysis Visualization

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To learn more about reviewing your results, visit the help guide. Selected Analyses

ctDNA IRGV

Summary SNV / Indel CNV Fusion

Analysis Name : Sample Name/Barcode Id	SNV / Indel				CNV				Fusion		
	Gene	MAF %	AA Chg	QC Test (LOD) %	Gene	Gain / Loss	CNV Ratio	QC Test	Variant (exons)	Mol Cov. Mutant	QC Test
51902_v1_c1147_2017-11-27-13-35-551 : 51902_v1/IonCodeTag_0105	TP53	4.7387	p.R273C	0.1	none detected			✓	KIF5B-RET(15R12.C08F1232)	16	✓
	TP53	0.0588	p.Y220C	0.05							

1 - 1 of 1 items

10p11.22 10q11.21 Chr. 10

KIF5B RET

FISHprobe®

Analysis Results

Analysis Name: 52069_RNA_v1_c864_2017-12-14-13-32-106 Fusion Sample QC: PASS [TotalMappedFusionPanelReads>5000] Fusion Overall
Total Mapped Fusion Panel Reads: 201562

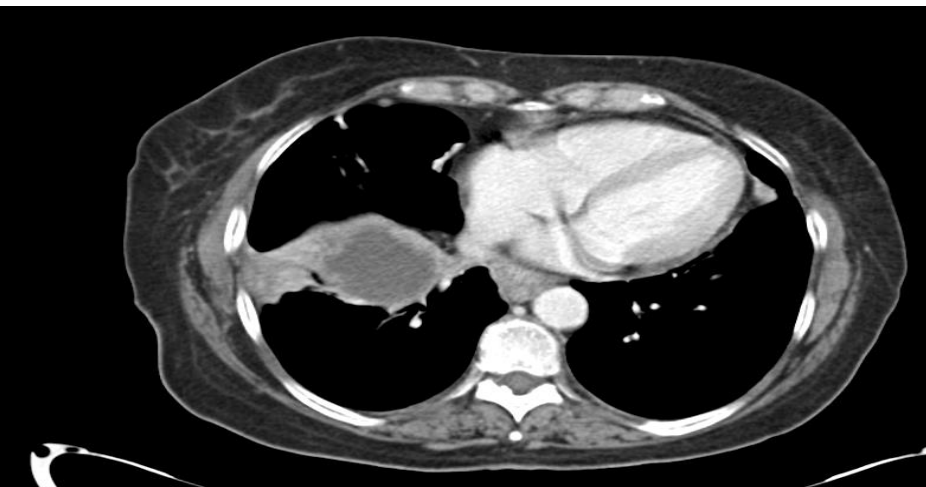
Fusions

Classification	Locus	Type	Gene(s)/Exon(s)	Read Counts	Oncomine Variant Class	Oncomine
Unclassified	chr10:32317355 - chr10:43612031	FUSION	KIF5B(15) - RET(12)	755	Fusion	Gain-of-fur

1 - 1 of 1 items

Caso 4

Basal



C5D1



LOXO 292
120 mg BID

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

Gracias por su atención

www.IAVANTE.es

