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Licenciada en Medicina por la Universidad de Sevilla en 2005. Formación como especialista en Oncología Médica en el Hospital Universitario Virgen Macarena entre 2005 y 2009. Máster en Cuidados Paliativos por la Universidad Pontificia Comillas en 2009. Máster en Ensayos Clínicos por la Universidad de Sevilla en 2010. Experto en Inmunoterapia y Cáncer por la Universidad de Navarra en 2016. Actualmente Oncólogo Médico de HUV Macarena, en la Unidad de Cáncer Genitourinario, Tumores de origen desconocido y Tumores de baja incidencia. Vocal de las Secciones de Cuidados Contínuos y de Prevención y Diagnóstico Precoz de la Sociedad Española de Oncología Médica (SEOM). Miembro de la Sección de Trombosis de SEOM. Secretaria del Grupo Español de Cáncer de Origen Desconocido (GECOD) y Vocal de la Junta Directiva del Grupo Español de Tumores Huérfanos e Infrecuentes (GETHI).

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Introduction

Identification of "agnostic" genetic drivers in cancer is forseen as a major step forward in precision medicine. Unfortunately, "off label" use of targeted therapies is not widely available and many oncogenic alteration do not present the same behaviour accross all tumor types. We aimed to analyze the real impact on patients management of the implementation of a systematic screening of genetic alterations in centers of the Spanish
Group for Rare Cancer (GETHI).

Methods

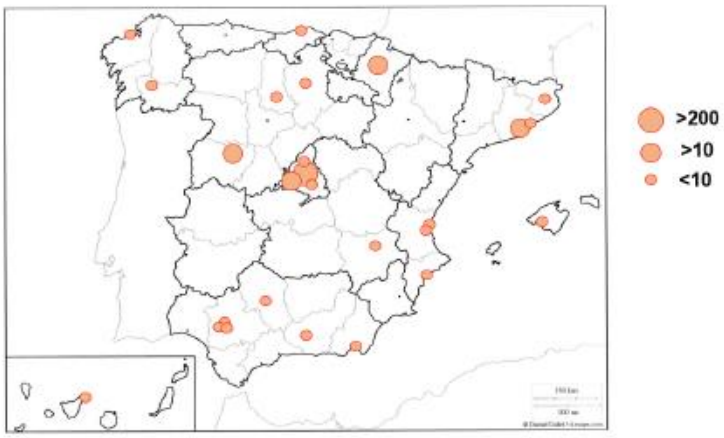
We designed an observational, prospective and multicenter study to molecularly characterize any adult patient with advanced cancer. Clinical data regarding treatment administered and outcome, were collected from patiens identifies as harboring drugable alterations.

Genes analyzed in Archer Fusion Plex Solid Tumor Panel

AKT3	ARHGAP26	ALK	AXL	BRD3	PRKCA
BRAF	BRD4	EGFR	ERG	ESR1	ROS1
ETV1	ETV4	ETV5	ETV6	EWSR1	TFEB
FGFR1	FGFR2	FGFR3	FGR	INSR	PRKCB
MAML2	MAST1	MAST2	MET	MSMB	RSPO2
MUSK	MYB	NOTCH1	NOTCH2	NRG1	THADA
NTRK1	NTRK2	NTRK3	NUMBL	NUTM1	RAF1
PDGFRA	PDGFRB	PIK3CA	PKN1	PPARG	RSPO3
TMPRSS2	TFE3	TERT	RELA	RET	

Results

Hospitals involved in the study. 341 samples collected from 26 National Hospitals (36 MDs) distributed all over the country



Epidemiology. Tumor types and number of cases.

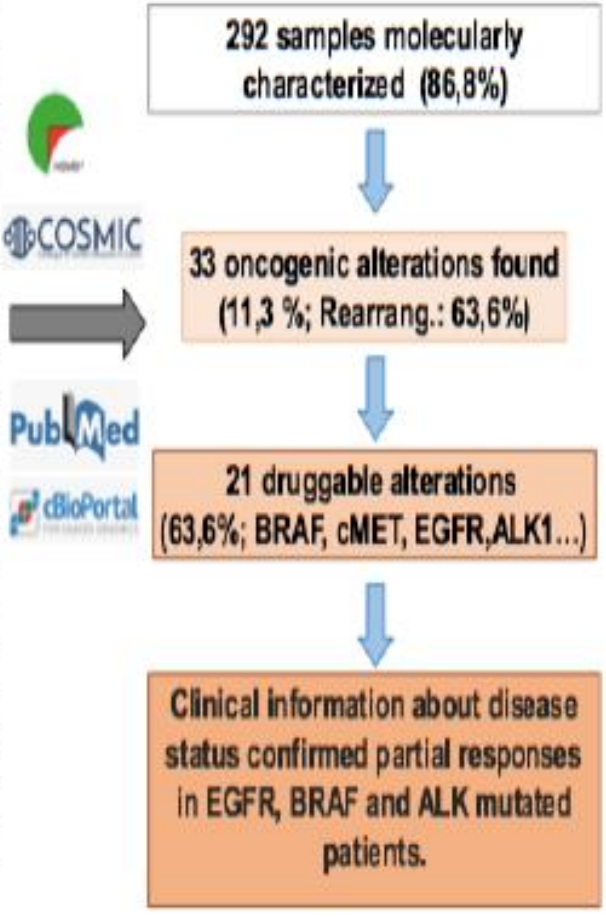
TYPE OF TUMOR (number of cases)

OVARY	52	RECTUM/SIGMA ADENOCARC	6	LYMPHOMA/LEUKEMIA/MYELOMA	3
GLIOBLASTOMA	45	GLIOMA	6	TESTIS	2
LUNG	27	OLOGODENDROGLIOMA	6	BRAIN	2
PROSTATE	18	SALIVARY GLAND	6	MEDULOBLASTOMA	2
RENAL	17	GATRIC CANCER	6	NEUROBLASTOMA	2
BREAST	13	CHOLANGIOCARCINOMA	6	PHARYNX	2
BLADDER	13	THYME	6	GALLBLADDER	2
COLON	10	MESOTELIOMA	5	LARYNX	2
UNKOWN ORIGIN	9	SARCOMA	5	VAGINA	1
MELANOMA	8	BONE	4	PENIS	1
PANCREATIC CANCER	7	CERVIX	3	PARAGANGLIOMA	1
ENDOMETRIUM	7	LIVER	3	ESOPHAGUS	1
ASTROCITOMA	7	THYROID CANCER	3	AMYGDALA ADENOCARC	1
HEAD AND NECK	1	PITUITATY GLAND	1	PARATID GLAND TUMOR	1

Figure 3: Molecular findings

TUMOR	MOLECULAR FINDINGS
Breast cancer	ESR1 c.1613A>G; p.D538G
	ESR1-NFAT5 fusion
Cholangiocarcinoma	BRAF c.1397G>T; p.G466V
	FGFR2-RBM20 fusion
	HOOK1-**RET** fusion
Collecting duct carc	MET p.T992I
Estesioneuroblastoma	TFE3-ABCA7 fusion
Ewing Sarcoma	EWSR1-**ERG** fusión
Gastric cancer	CLDN18-ARHGAP26 fusión
Glioblastoma	EGFR pA289V (3)
	EGFR pG598V (2)
	EGFR deletion exon 2-7
	BRAF deletion
	CAPZA2-**MET** fusion
	SRPK2-**MET** fusion
HG Glial tumor	PTPRZ1-**MET** fusion
Lung carcinoma	MET exon 14 deletion
	CD74-**ROS1** fusion
	EML4-**ALK** fusion
Oligodendroglioma	SYN2-PPARG fusion
	FGFR3-TACC3 fusion
Prostate	TMPRSS2-**ERG** fusion (5)
	UNC5D-NRG1 fusion
	SND1-**BRAF** fusion
Rectum adenoc.	TMPRSS2-ANKRD36 fusion

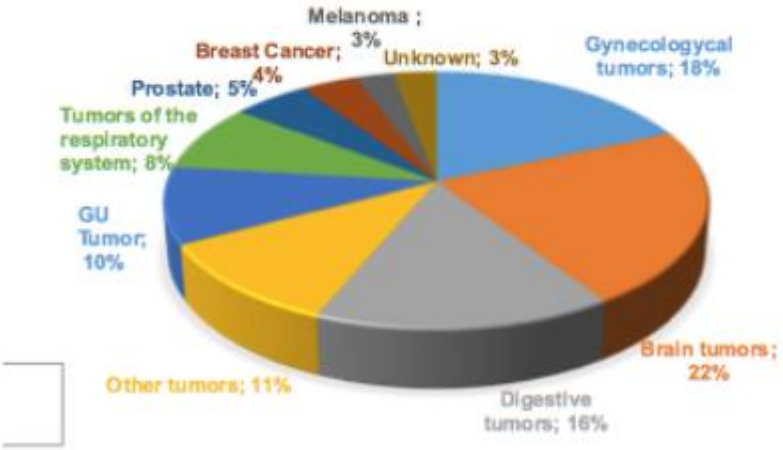
*: Number of patients harbouring indicated in brackets;
** Oncogenic drivers labelled in bold



AGE	
<18	1%
19-40	11%
41-65	56%
>65	33%

FEMALE	52%
MALE	48%

TISSUE SAMPLE	
PRIMARY TUMOR	82%
METASTATIC TUMOR	18%



Conclusions

Though only few cases harboring druggable alterations got specific treatment, 50% achieved a meaningful benefit. A wide access to molecular screening and targeted drugs could improve the outcome of cancer patients.