

# Percorsi di Oncologia di Precisione:

Appropriatezza diagnostica e  
Molecular Tumor Board

**30 GENNAIO 2026**  
**MILANO**

INNSiDE by Meliá Milano Torre Galfa  
Via Gustavo Fara, 41

## AIROLDI MARIO, MD – DISCLOSURES

CONSULTING OR ADVISORY BOARD: Astra Zeneca, Novartis, Bristol MS, Daiichi S.

TRAVEL EXPENSES: Gilead, Novartis, Roche

**AIROLDI MARIO**

Coordinatore Area Ospedaliera Rete Oncologica del Piemonte e Valle d'Aosta  
Coordinatore MTB Regionale



rete  
oncologica  
Piemonte | Valle d'Aosta

**Molecular Tumor Board**

# CONSUNTIVO ATTIVITA'

- **PRIMA SEDUTA : 14.12.23**
- **NUMERO SEDUTE : 48**
- **CASI PRESENTATI DAI GIC AL MTB : 145**
- **CASI DISCUSSI MTB : 130 (89%)**
- **CASI TESTATI = 99/130 (76%)**
- **CASI DRUGGABLE = 61/99 (61%)**

# NGS (99 TEST)

• SOMATICO	93
• BIOPSIA LIQUIDA	6
• <u>FondationOne CDx</u>	95
• <u>FondationOne HEME</u>	4
• TURNAROUND TIME	13 gg ( <u>median</u> ) (12-17)

# CONSUNTIVO DRUGGABLE

- **N. CASI : 61**
- **SESSO : 42 F (68%) / 19 M (32%)**
- **ETA' : mediana 55 (23-80)**
- **PS: 0 = 25 (40 %), 1 = 31 (51 %) , 2 = 5 (9 %)**
- **MEDIANA TERAPIE MEDICHE = 2 (0-9)**

# CONSUNTIVO DRUGGABLE : SEDI

• FOCUS IGNOTO	= 10
• PANCREAS	= 7
• MAMMELLA	= 6
• GH. SALIVARI	= 5
• SARCOMI	= 4
• OVAIO	= 4
• ANO	= 3
• FEGATO	= 3
• TESTA COLLO	= 3
• PLEURA/POLMONE	= 3
• ENDOMETRIO	= 2
• PROSTATA	= 2
• ALTRO	= 9

# MUTAZIONI DRUGGABLE

- PI3KCA/PTEN = 12 ( 19 %) (4 GH SALIVARI)
- HTMB = 12 (19 %) (4 FOCUS IGNOTO)
- BRCA 1-2 + HRD = 9 (14 %) (3 MAMMELLA germ neg)
  
- CDKN2A = 15 ( 24 %) (4 PANCREAS)

# PAZ IN TERAPIA

- PI3K = 10/12 (PI3K / mTOR INIB)
- HTMB = 11/12 (ANTI PD1)
- BRCA 1-2/ HRD + = 8/9 (PARP INIBITORI)
  
- CDKN2A = 8/15 (INIBITORI CICLINE)

**ABOUT THE TEST** FoundationOne®CDx is a next-generation sequencing (NGS) based assay that identifies genomic findings within hundreds of cancer-related genes.

**PATIENT**  
DISEASE Peritoneum mesothelioma  
NAME 03-2024-00122906, IT  
DATE OF BIRTH 02 July 1950  
SEX Male  
MEDICAL RECORD # MTB-029

**PHYSICIAN**  
ORDERING PHYSICIAN AIROLDI, MARIO  
MEDICAL FACILITY RETE ONCO PIEMONTE VALLE D'AOSTA  
ADDITIONAL RECIPIENT None  
MEDICAL FACILITY ID 336652  
PATHOLOGIST Not Provided

**SPECIMEN**  
SPECIMEN SITE Abdomen  
SPECIMEN ID 24-C-00762 A1  
SPECIMEN TYPE Block  
DATE OF COLLECTION 19 February 2024  
SPECIMEN RECEIVED 27 December 2024

### Genomic Signatures

**HRD signature** - HRDsig Positive

**Microsatellite status** - MS-Stable

**Tumor Mutational Burden** - 5 Muts/Mb

### Gene Alterations

*For a complete list of the genes assayed, please refer to the Appendix.*

**CDKN2A** loss

**MTAP** loss

**NF2** splice site 448-2A>T

**CDKN2B** loss

### Report Highlights

- Targeted therapies with potential clinical benefit approved in another tumor type: Niraparib (p. 8), Olaparib (p. 8), Rucaparib (p. 9), Talazoparib (p. 9)
- Evidence-matched clinical trial options based on this patient's genomic findings: (p. 10)

## GENOMIC SIGNATURES

HRD signature - HRDsig Positive

10 Trials [see p. 10](#)

Microsatellite status - MS-Stable

Tumor Mutational Burden - 5 Muts/Mb

## GENE ALTERATIONS

**CDKN2A** - loss

10 Trials [see p. 12](#)

**MTAP** - loss

6 Trials [see p. 14](#)

**NF2** - splice site 448-2A>T

3 Trials [see p. 15](#)

### THERAPIES WITH CLINICAL RELEVANCE (IN PATIENT'S TUMOR TYPE)

none

No therapies or clinical trials. See Genomic Signatures section

No therapies or clinical trials. See Genomic Signatures section

### THERAPIES WITH CLINICAL RELEVANCE (IN OTHER TUMOR TYPE)

Niraparib

Olaparib

Rucaparib

Talazoparib

### THERAPIES WITH CLINICAL RELEVANCE (IN PATIENT'S TUMOR TYPE)

none

none

none

### THERAPIES WITH CLINICAL RELEVANCE (IN OTHER TUMOR TYPE)

none

none

none

# CARCINOMA TIMICO

## PATHOLOGIST COMMENTS

*Douglas A. Mata, MD, MPH 11-Nov-2024*

A KIT exon 11 V560del mutation was detected. This particular mutation has been recurrently identified in thymic carcinomas, gastrointestinal stromal tumors, and mucosal melanomas, and less commonly has been reported in non-small-cell lung carcinomas. Clinicopathologic correlation is advised.

### GENOMIC SIGNATURES

HRD signature - HRDsig Negative

Microsatellite status - MS-Stable

Tumor Mutational Burden - 2 Muts/Mb

### GENE ALTERATIONS

**KIT** - exon 11 deletion (V560del)

10 Trials see p. 2

### THERAPY AND CLINICAL TRIAL IMPLICATIONS

No therapies or clinical trials. See Genomic Signatures section
No therapies or clinical trials. See Genomic Signatures section
No therapies or clinical trials. See Genomic Signatures section

THERAPIES WITH CLINICAL RELEVANCE (IN PATIENT'S TUMOR TYPE)	THERAPIES WITH CLINICAL RELEVANCE (IN OTHER TUMOR TYPE)
none	Dasatinib
	Imatinib
	Nilotinib
	Ponatinib
	Sorafenib
	Sunitinib

**ABOUT THE TEST** FoundationOne®CDx is a next-generation sequencing (NGS) based assay that identifies genomic findings within hundreds of cancer-related genes.

## PATIENT

DISEASE Soft tissue fibromatosis  
NAME 03-2024-00104923, IT  
DATE OF BIRTH 05 April 1986  
SEX Male  
MEDICAL RECORD # MTB-004

## PHYSICIAN

ORDERING PHYSICIAN AIROLDI, MARIO  
MEDICAL FACILITY OSPEDALE S G BATTISTA MOLINETTE - REPARTO  
ONCOLOGIA MEDICA 2  
ADDITIONAL RECIPIENT None  
MEDICAL FACILITY ID 333537  
PATHOLOGIST Not Provided

## SPECIMEN

SPECIMEN SITE Abdominal wall  
SPECIMEN ID I 3058 2023 A2  
SPECIMEN TYPE Block  
DATE OF COLLECTION 06 March 2023  
SPECIMEN RECEIVED 01 March 2024

## Genomic Signatures

Microsatellite status - MS-Stable  
Tumor Mutational Burden - 1 Muts/Mb

## Gene Alterations

*For a complete list of the genes assayed, please refer to the Appendix.*

**APCE422fs\*32, D610fs\*14, E1464fs\*8**

## Report Highlights

- Variants with diagnostic implications that may indicate a specific cancer type: **APC D610fs\*14, E1464fs\*8, E422fs\*32 (p. 3)**
- Evidence-matched clinical trial options based on this patient's genomic findings: (p. 4)

**GENE****APC****ALTERATION**

E422fs\*32, D610fs\*14, E1464fs\*8

**RATIONALE**

Based on preclinical and limited clinical data, APC inactivation may be associated with sensitivity to CBP/beta-catenin interaction inhibitors.

**NCT04008797****PHASE 1**

A Study of E7386 in Combination With Other Anticancer Drug in Participants With Solid Tumor

**TARGETS**

CBP, Beta-catenin, FGFRs, RET, PDGFRA, VEGFRs, KIT

**LOCATIONS:** Lyon (France), Pessac (France), Paris (France), Amiens (France), Texas, Seodaemun (Korea, Republic of), Jongno-gu (Korea, Republic of), Seoul (Korea, Republic of), Songpa-gu (Korea, Republic of)**NCT03264664****PHASE 1**

Study of E7386 in Participants With Selected Advanced Neoplasms

**TARGETS**

CBP, Beta-catenin

**LOCATIONS:** London (United Kingdom), Manchester (United Kingdom), Glasgow (United Kingdom), Minnesota, Florida, Arizona, California**NCT05949099****PHASE 2**

Study of Cryoablation and Nirogacestat for Desmoid Tumor

**TARGETS**

Gamma-secretase

**LOCATIONS:** California

## Genomic Signatures

Microsatellite status - MS-Stable

Tumor Mutational Burden - 2 Muts/Mb

## Gene Alterations

*For a complete list of the genes assayed, please refer to the Appendix.*

**PTPN11 S502L**

### GENE

**PTPN11**

### ALTERATION

S502L

### HGVS VARIANT

NM\_002834.3:c.1505C>T (p.S502L)

### VARIANT CHROMOSOMAL POSITION

chr12:112926885

### VARIANT ALLELE FREQUENCY (% VAF)

34.9%

# PTPN11 – SINDROME NOONAN

RARE IN ADC 0-1.62 %

ENCODE SHP-2

Embrional rhabdomyosarcoma, neuroblastoma,  
juvenile myelomonocytic leukemia

SHP-2 activate RAS-MEK-ERK-PI3K-AKT-  
mTOR, SCR kinase (activity of MEK inhibitors)

**INSERITO TRIAL MEK INIBITORI**