



## Genomic profiling in patients with bone metastases

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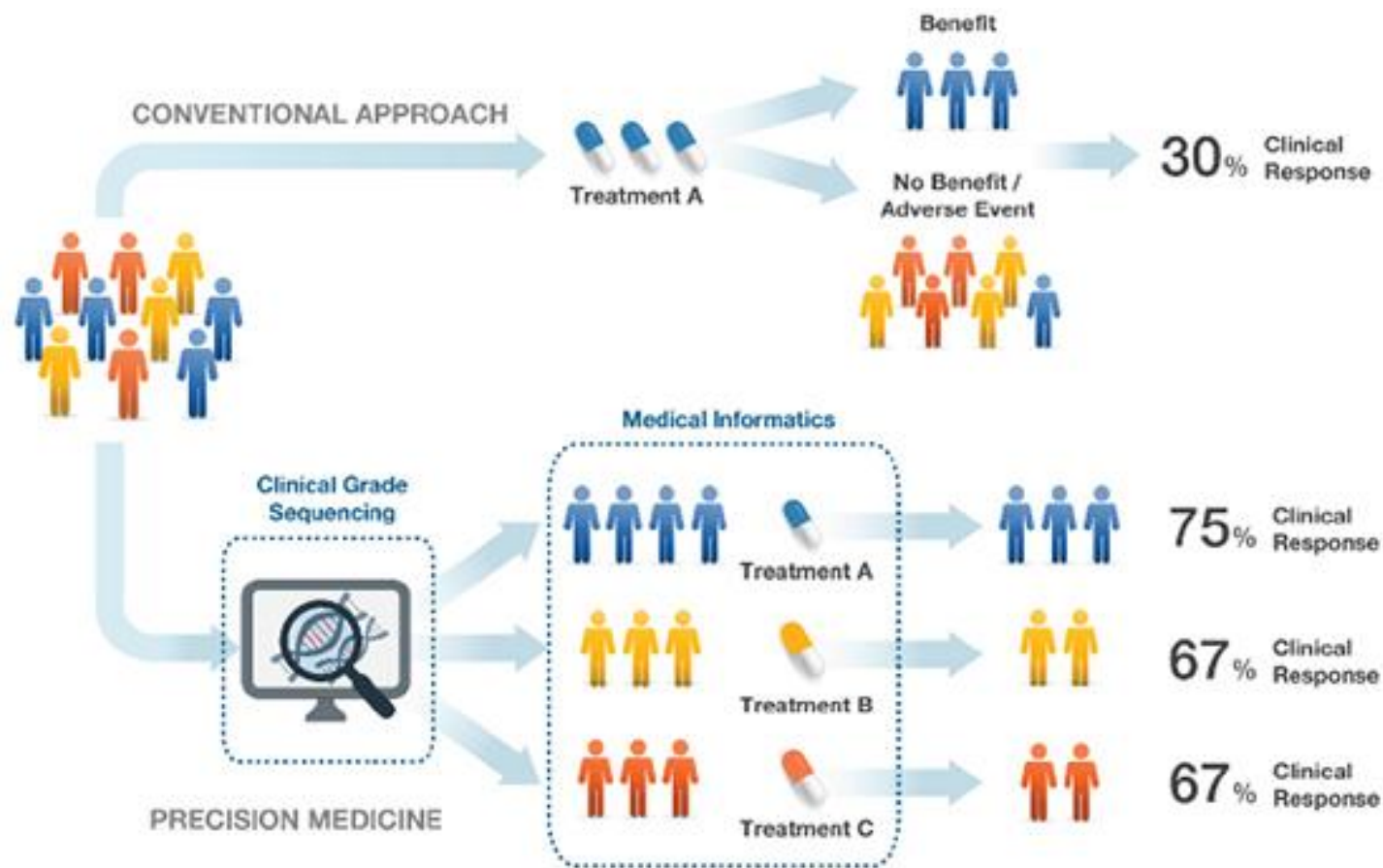


Figure 1. Precision diagnostics stratifies patients according to their molecular signature

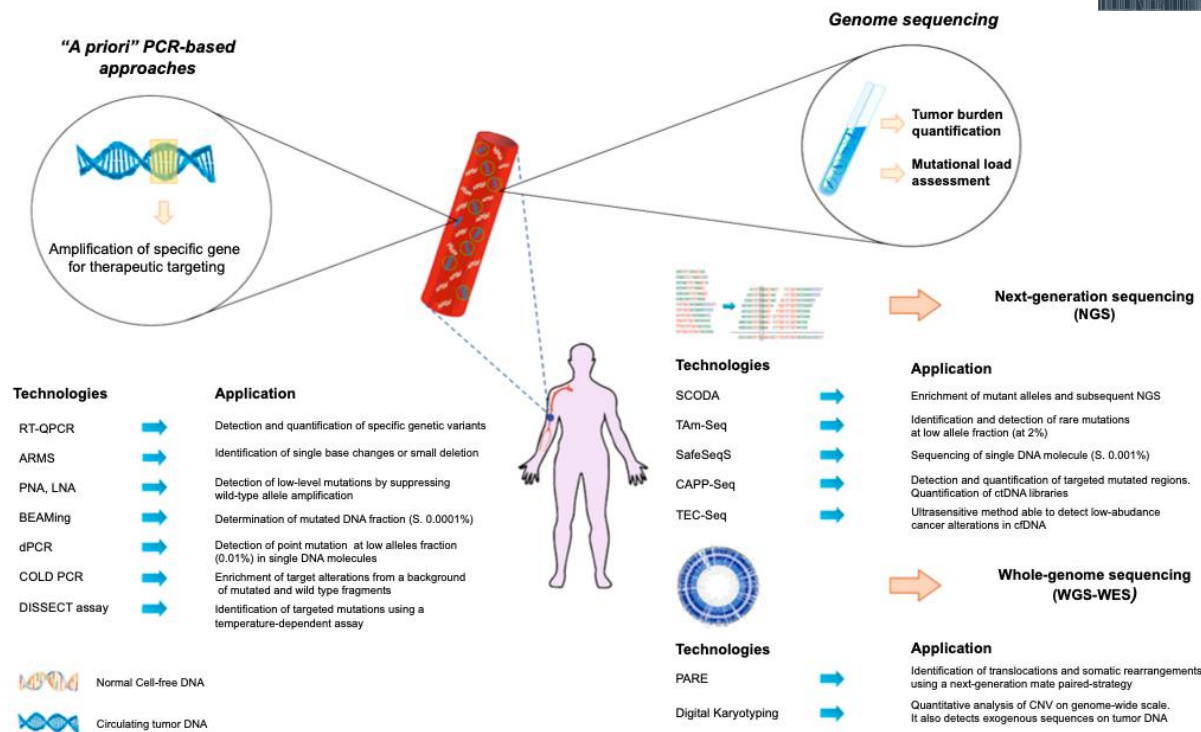
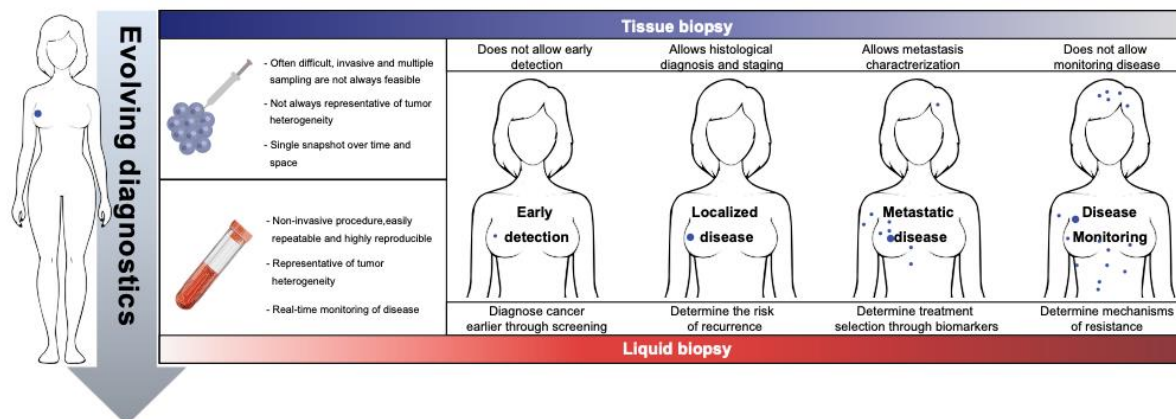
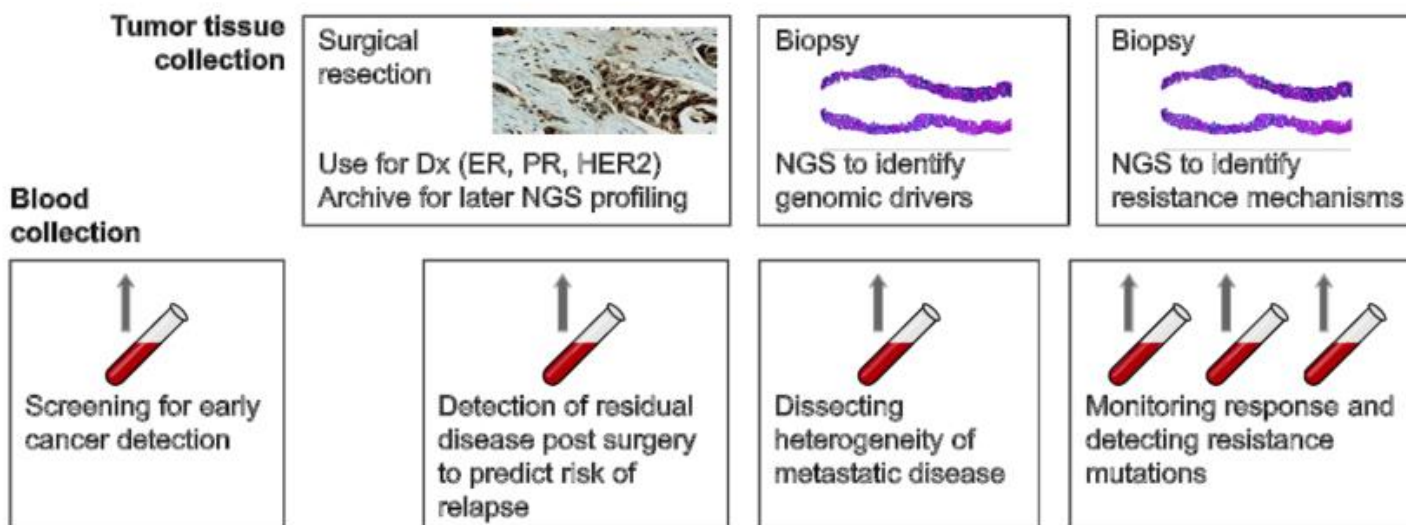
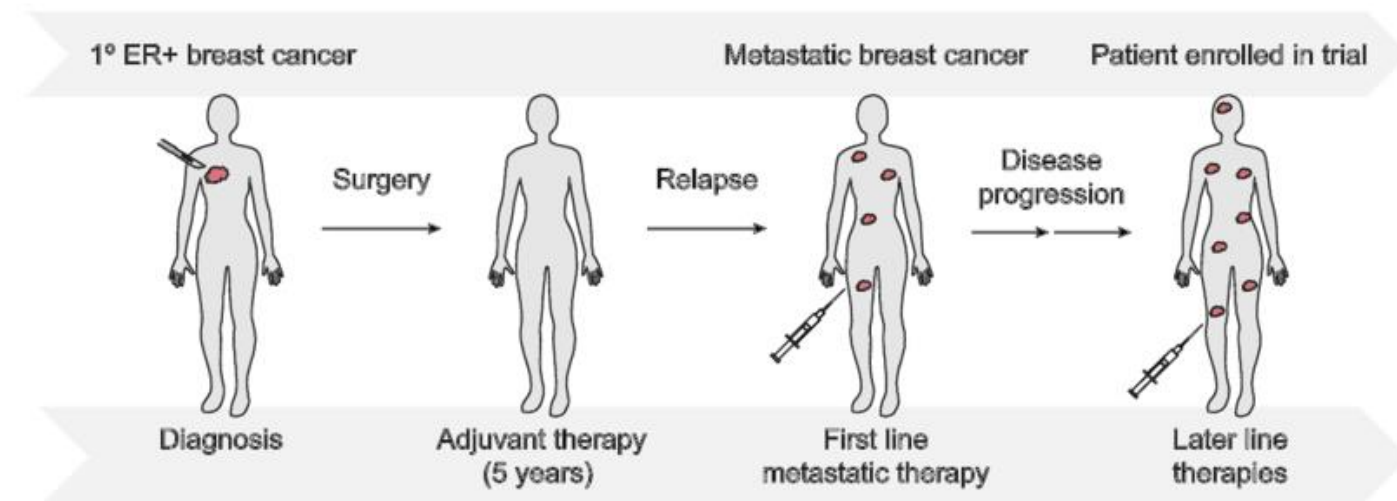
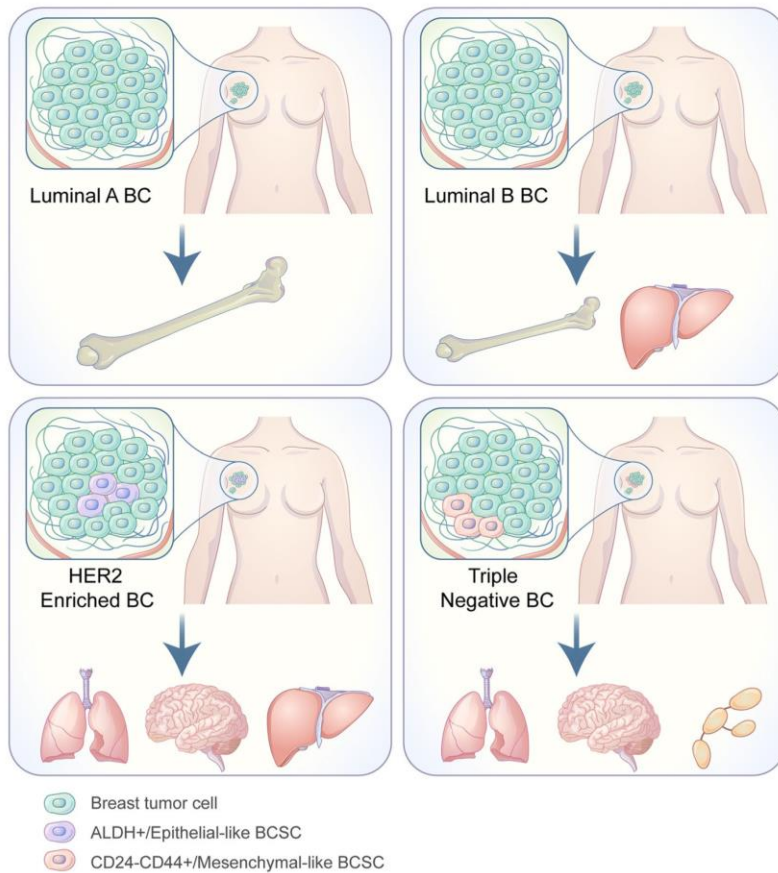


Fig. 1. Genome sequencing vs "a priori" techniques: comparison and application summary.

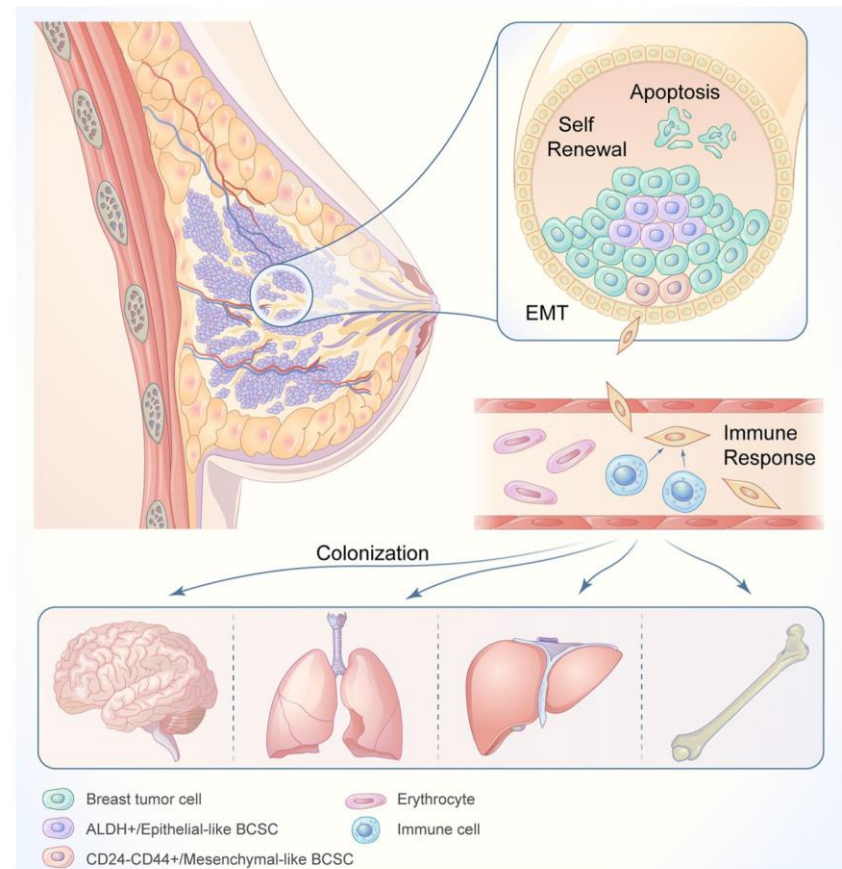


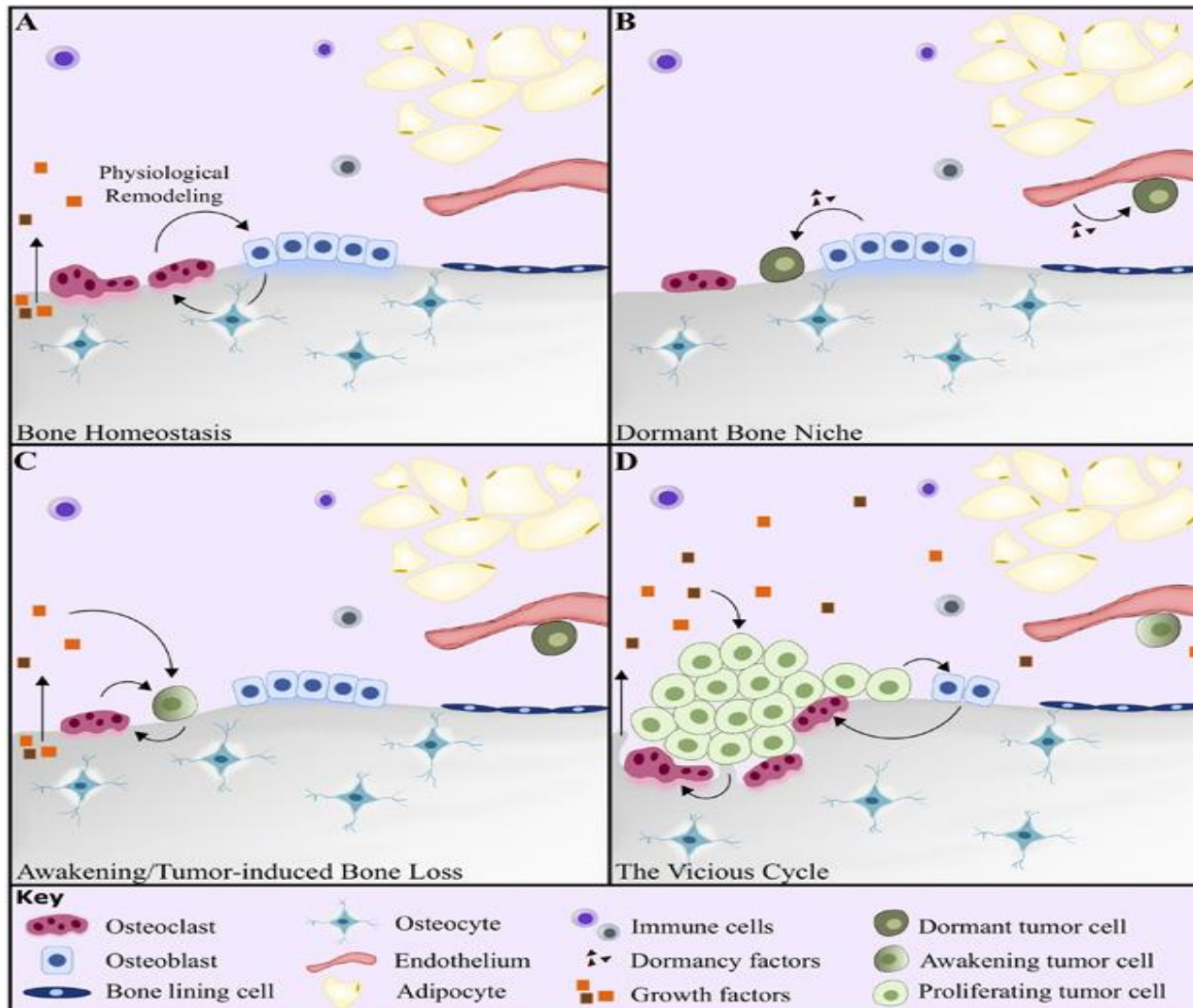






Bone is the most common location for BC metastases.  
Bone metastases occur in 50-70% of MBC.

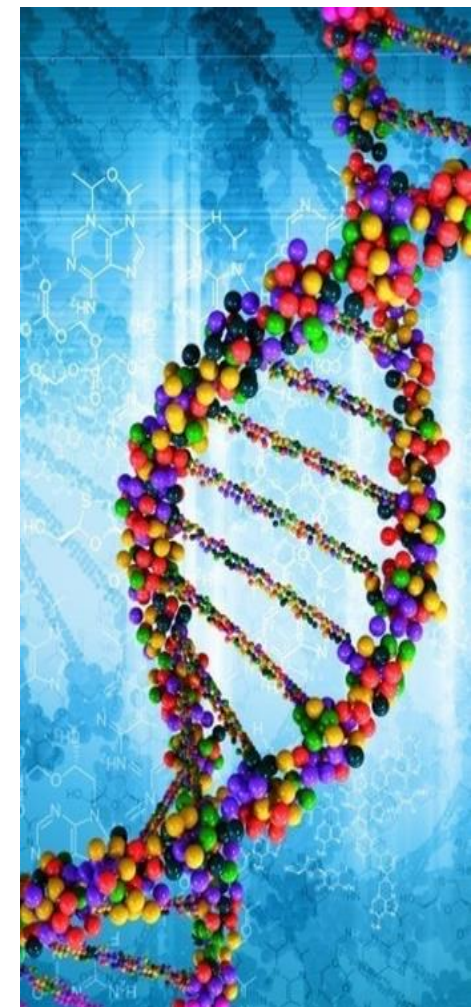




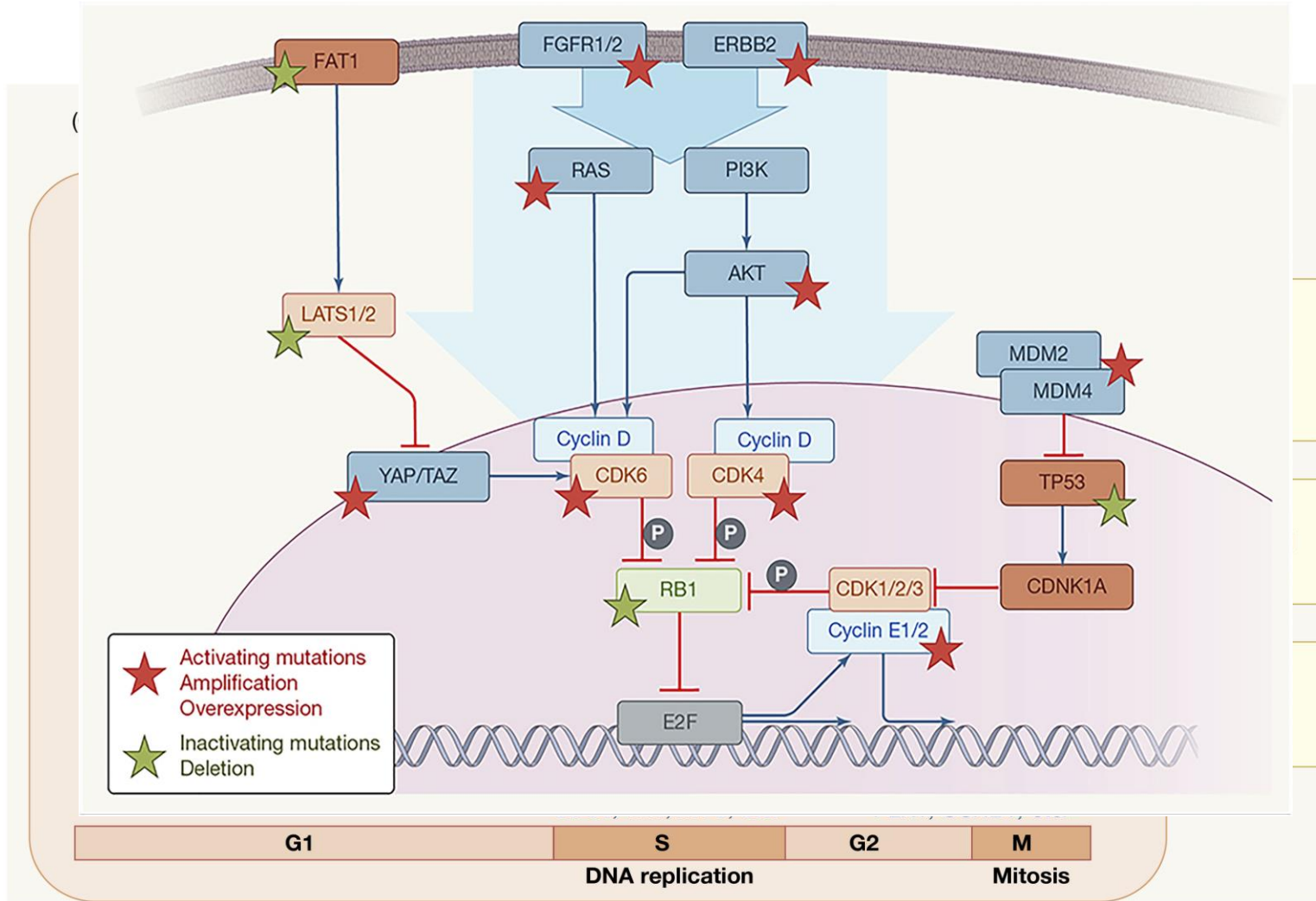
Primary Breast Cancer	Bone	Lung	Liver	Brain
<i>PIK3CA</i>				PAM50
<i>TP53</i>				VEGF-A
<b><i>ERBB2</i></b>				IL-8
<i>FGFR1</i>	<i>TFF1</i>	<i>MMP1</i>	<i>MAPK</i>	<i>ATAD2 DERL1</i>
<i>CCND</i>	<i>TFF3</i>	<i>MMP2</i>	<i>NFκB</i>	<i>NEK2A</i>
<i>MUC16</i>	<i>AGR2</i>	<i>CXCL1</i>	<i>VEGF</i>	<i>ATM</i>
<i>AHNAK2</i>	<i>NAT1</i>	<i>PTGS2</i>	<i>ESR1</i>	<i>CRYAB</i>
<i>SYNE1</i>	<i>CR1P1</i>	<i>ID1</i>	<b><i>AKT1</i></b>	<i>HSPB2</i>
<i>KMT2C</i>	<i>CXCR4</i>	<i>VCAM1</i>	<b><i>ERBB2</i></b>	<i>ANGPT1</i>
<i>GATA3</i>	<i>CCR7</i>	<i>EREG</i>	<i>FGFR4</i>	<i>KDR</i>
<b><i>AKT1</i></b>	<i>SNAI1</i>	<i>SPARC</i>	<i>MS APOBEC</i>	<i>ITGAM</i>
<i>PTEN</i>	<i>IL11</i>	<i>IL13RA2</i>	cytidine	<i>PIK3CG</i>
<i>INPP4B</i>	<i>CTGF</i>	<i>AGR2</i>	deaminases	<i>TEK</i>
<i>PPP2R2A</i>	<i>CXCR4</i>	<i>KLF5</i>	<i>PPFIA1</i>	<i>BRAF</i>
<i>MTAP</i>	<i>MMP-1</i>	<i>Loxl2</i>		<i>BCL2</i>
<i>MAP2K4</i>				<i>AURKA</i>
<i>MNX1</i>				<i>AURKB</i>
				<i>FOXM1</i>

TFF1 TFF3 AGR2 NAT1 CR1P1

CXCR4 IL11

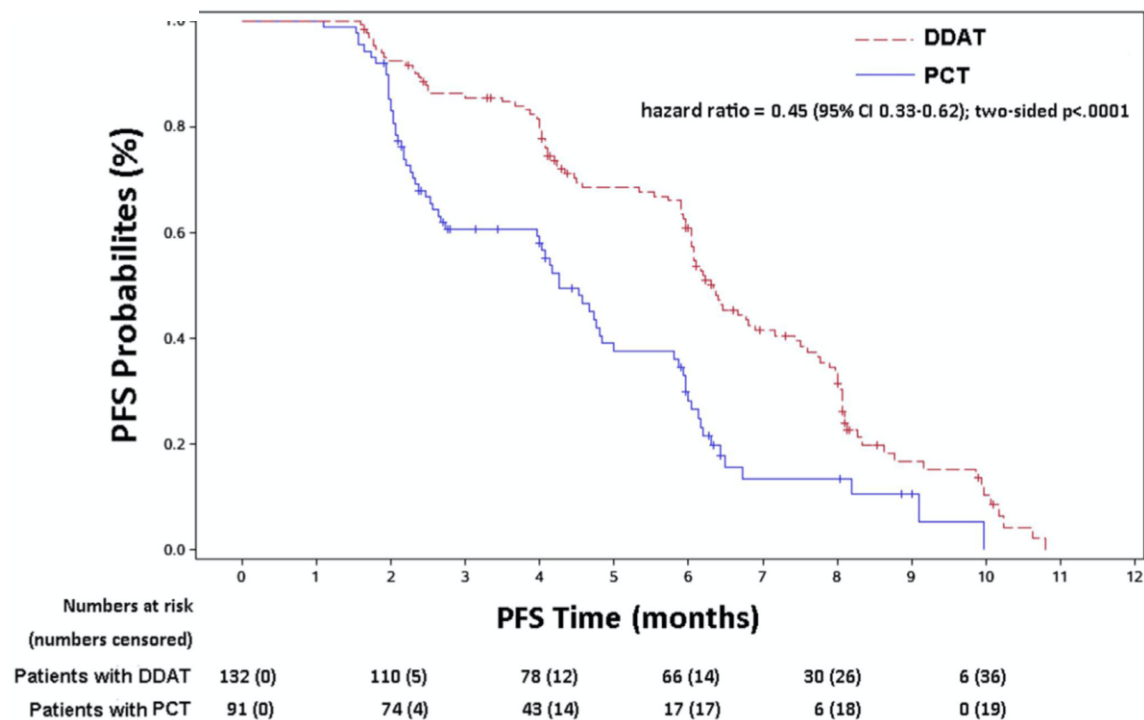




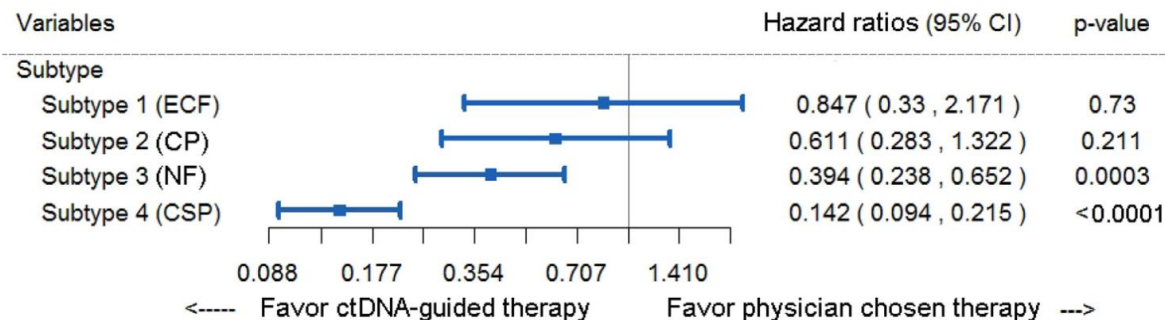




## Subtyping of metastatic breast cancer based on plasma circulating tumor DNA alterations: An observational, multicentre platform study



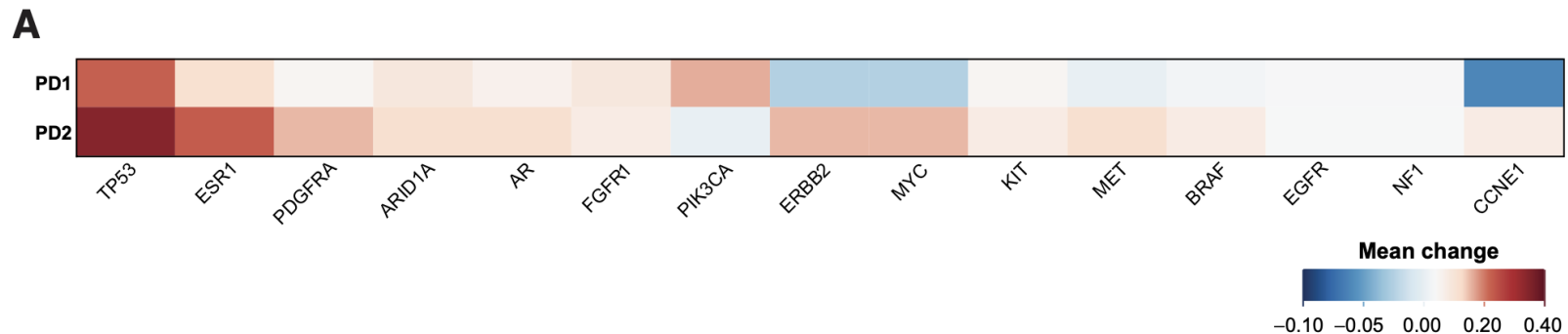
Druggable ctDNA alteration-guided treatment can significantly improve the PFS of patients with ctDNA alterations.



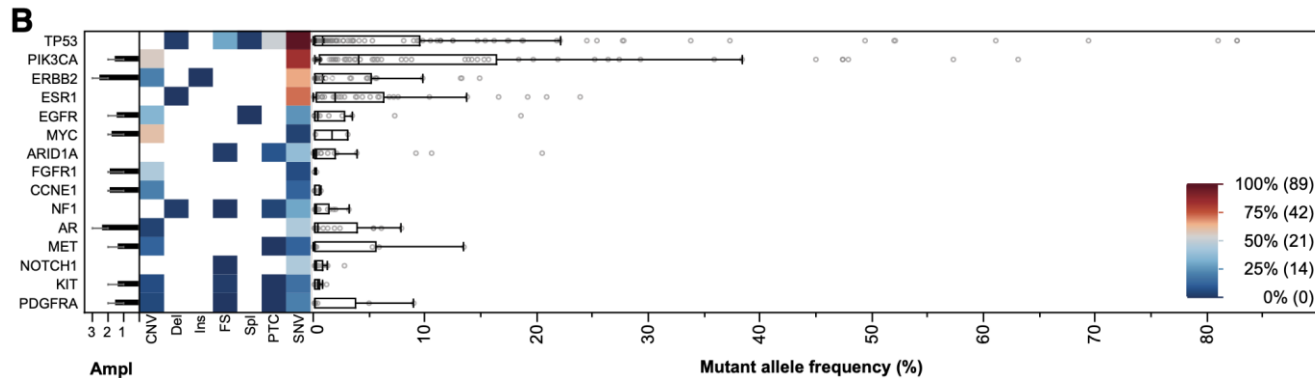
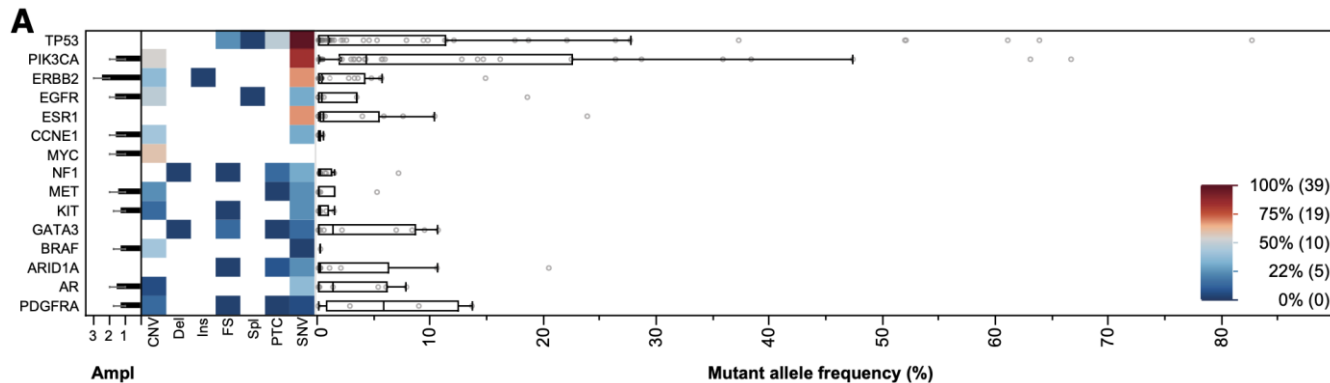
Tumor genomic evolution in metastatic breast cancer poses a significant treatment challenge at times of progression. Circulating tumor DNA (ctDNA) has emerged as a promising, noninvasive tool to detect alterations in peripheral blood over time



Potential of ctDNA as a tool for monitoring tumor genetic evolution and detecting resistance alterations in the setting of progressive disease.



## The Use of Serial Circulating Tumor DNA to Detect Resistance Alterations in Progressive Metastatic Breast Cancer



Circulating tumor DNA (ctDNA) is a promising tool for non-invasive monitoring of genomic alteration.

Analyzed serial ctDNA to characterize genomic evolution in progressive MBC.

## **Patients with Endocrine-Resistant Metastatic Breast Cancer with only BONE METASTASES.**

**Identify genomic biomarker associated with progression disease.**

Identify druggable ctDNA alteration guided treatment





## INCLUSION CRITERIA

- Age  $\geq 18$
- HR+ MBC
- HER2- MBC
- No prior treatment for MBC
- ONLY bone metastases
- Treatment with Denosumab – Zoledronic Acid



## EXCLUSION CRITERIA

- Age  $< 18$
- HR -MBC
- HER2+ MBC
- Prior treatment for MBC
- Visceral and No-visceral metastases

# Patients with Endocrine-Resistant Metastatic Breast Cancer with only BONE METASTASES.



## T0 – Baseline

Anamnesis, physical examination,  
Informed Consent, Blood sample  
collection.

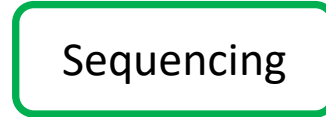
START first- line with iCDK4/6



Informative  
Blood sample  
with NGS



Sequencing



## T1 – Progressione Disease



## Materials and Methods

Peripheral blood samples before starting treatment with iCDK4/6 is collected.

Response to therapy was evaluated through routine staging with CT or PET-CT.

Progression is defined as evidence of clinical or radiographic progression resulting in change of treatment.







*“Quando curi una malattia puoi vincere o perdere. Quando ti prendi cura di una persona vinci sempre.”*

Patch Adams





## **DIRITTO ALLA MEDICINA PERSONALIZZATA**

**trattamento  
personalizzato**

**qualità  
della vita**

**accesso alle  
cure appropriate**

**privacy e  
rispetto degli  
aspetti psicologici**

**innovazione  
diagnostica e  
terapeutica**

**informazione  
e orientamento**

**partecipazione e  
scelta delle cure**

**umanizzazione  
delle cure**

**equità  
delle cure**

**tempestività  
delle cure**

**formazione**