

# SOMATIC CANCER

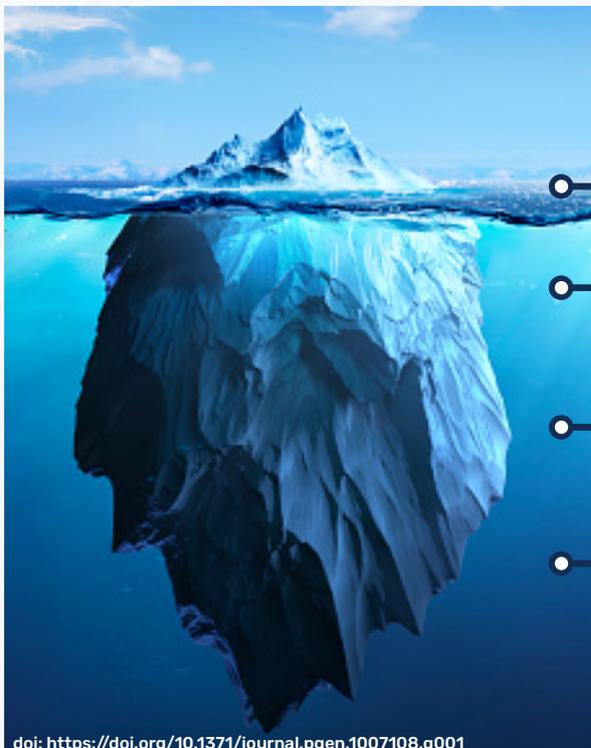
## Diagnosis by NGS Cancer Panels

### What are Acquired or Somatic mutations?

- Most common cause of cancers are caused by acquired mutations
- These mutations cannot be inherited from parent to child
- Do not occur in reproductive cells (egg or sperm cells)
- Are much more common than inherited mutation
- An individual with a germline mutation may also develop a somatic mutation
- Some common carcinogens that cause pathogenic variants include tobacco use, ultraviolet light or radiation, viruses, chemical exposures, and aging.



## NGS Reveals the Complete Iceberg of Cancer Mutations

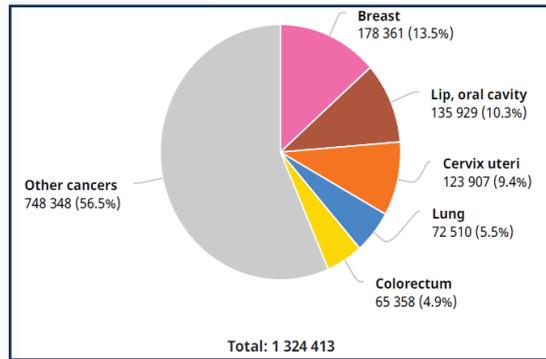


doi: <https://doi.org/10.1371/journal.pgen.1007108.g001>

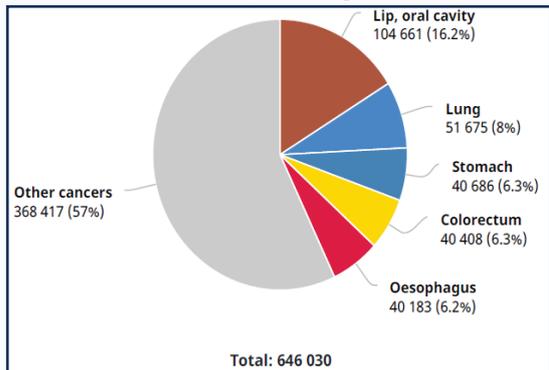
Mutation Type	Prevalence	Limit of Detection
Small CNVs and rearrangements	3% of older individuals	10% Sanger Seq.
SNV and Small in/dels in selectable genes (large clones)	10% of older individuals	2-10% Standard NGS
SNV and Small in/dels in selectable genes (small clones)	>95% of older individuals	0.001-0.1% Error-corrected NGS
SNV and Small in/dels in across genome (late arising or unselected event)	All cells in all people	Single genome Single-cell Seq. & Error-Corrected NGS

# Number of New Cancer Cases in India in 2020

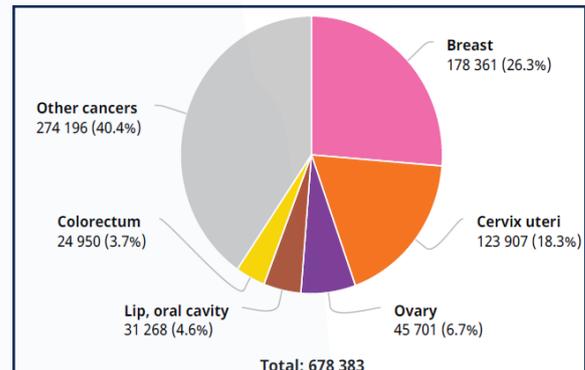
Both Sexes, All Ages



Males, All Ages



Females, All Ages



## Advantages of Next-generation sequencing (NGS) in Cancer Diagnosis

- NGS determines the sequence of DNA or RNA to study genetic variation associated with diseases or other biological phenomena.
- NGS allows clinicians to test multiple genes of a cancer simultaneously, thus, eliminates the need of multiple tests to identify the causative mutation and new markers that may offer additional treatment options
- NGS offers advantages in accuracy, sensitivity, and speed of diagnosis of mutations making significant impact on the treatment
- When standard cancer treatments don't work, or if doctors can't determine where a patient's cancer originated, genomic sequencing can help pinpoint mutations in a tumor that might be matched with medicines targeting those specific alterations

### Ion AmpliSeq Cancer Hotspot Panel v2

Hotspot regions, including ~2,800 COSMIC mutations of 50 oncogenes and tumor suppressor genes, with wide coverage of the KRAS, BRAF and EGFR genes

### Ion AmpliSeq™ Comprehensive/v3 Cancer Panel

16,000 primer pairs covering 409 genes

### Ion AmpliSeq BRCA1 and BRCA2 Panel

Targeted research panel investigating somatic and germline variants in BRCA1 and BRCA2

### AmpliSeq for Childhood Cancer Panel

Targeted panel for investigating 203 genes associated with cancer in children and young adults