

SOMATIC CANCER

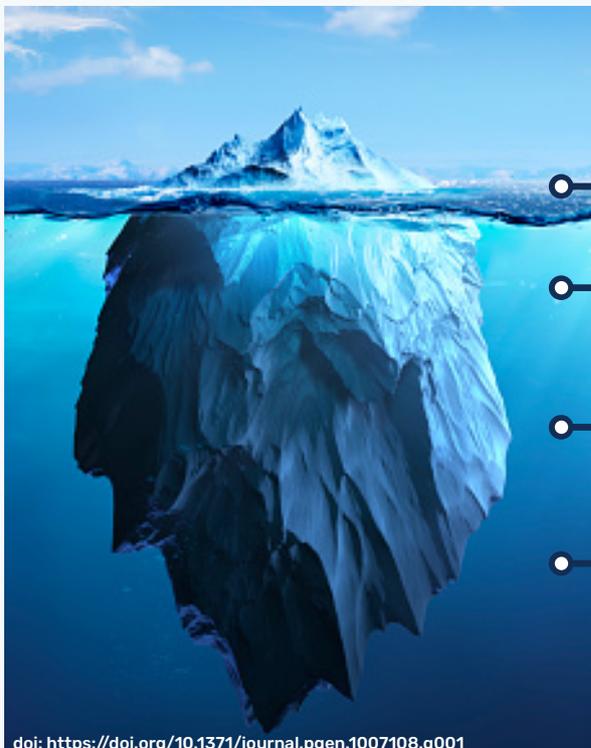
Diagnosis by NGS Cancer Panels

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

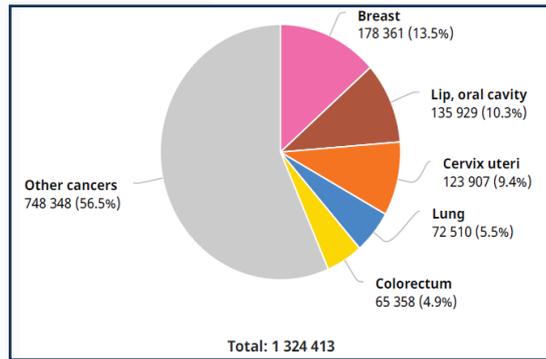


doi: <https://doi.org/10.1371/journal.pgen.1007108.g001>

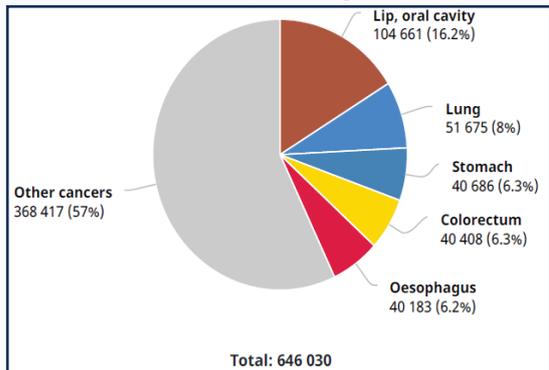
Mutation Type	Prevalence	Limit of Detection
Small CNVs and rearrangements	3% of older individuals	10% Sanger Seq.
SNV and Small in/dels in selectable genes (large clones)	10% of older individuals	2-10% Standard NGS
SNV and Small in/dels in selectable genes (small clones)	>95% of older individuals	0.001-0.1% 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

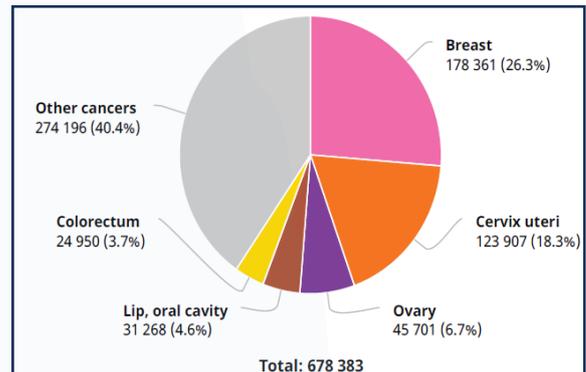
Both Sexes, All Ages



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