

ACS Cancer Conference 2025

March 12-14 | Phoenix, AZ

Basics of Integrating Genetics and Genomics into Multidisciplinary Care

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ACS Cancer Programs
American College of Surgeons

No disclosures

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Objectives

- Understand germline vs somatic mutations
- Give practical applications in cancer care
- Explain precision oncology

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Genetic questions for the cancer clinician

- Did you inherit a mutation?
 - Germline mutation
 - Blood or saliva test
- Are there acquired mutations?
 - Somatic mutation
 - Cancer tissue or blood (liquid biopsy)

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Genetics vs Genomics

- Genetics
 - Understanding the structure and function of a single gene and is usually used in the context of inherited gene variants
- Genomics
 - The study of the complete set of genes (the genome), of the way genes work, interact with each other and with the environment and is mainly used in the context of somatic cancer mutations

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Hereditary cancer syndromes

- Hereditary Breast and Ovarian Cancer
- Cowden Syndrome
- Li-Fraumeni Syndrome
- Von Hippel-Lindau
- Lynch Syndrome (Hereditary non-polyposis colon cancer)
- Familial Adenomatous Polyposis
- Multiple Endocrine Neoplasia

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Implication of inherited mutations

- Assessment of future cancer risk
- Surgical decision making
- Risk reducing (prophylactic) surgery
- Adjuvant treatment options
- Screening for other associated cancers
- Testing of family members

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Genetic testing

- Multigene panel testing has shown that pathogenic germline mutations are common
 - 13-17% of cancer patients may harbor a pathogenic or likely pathogenic mutation when universal germline testing is performed
- NCCN endorses universal genetic testing in ovarian, pancreas, advanced prostate and metastatic breast cancers
- PARP inhibitors are FDA approved for advanced ovarian cancer patients with germline *BRCA 1/2* mutations
 - Additional drugs have gained approval for other pathogenic germline alterations (ovarian, prostate, astrocytoma)

JAMA 318 : 825 - 835, 2017

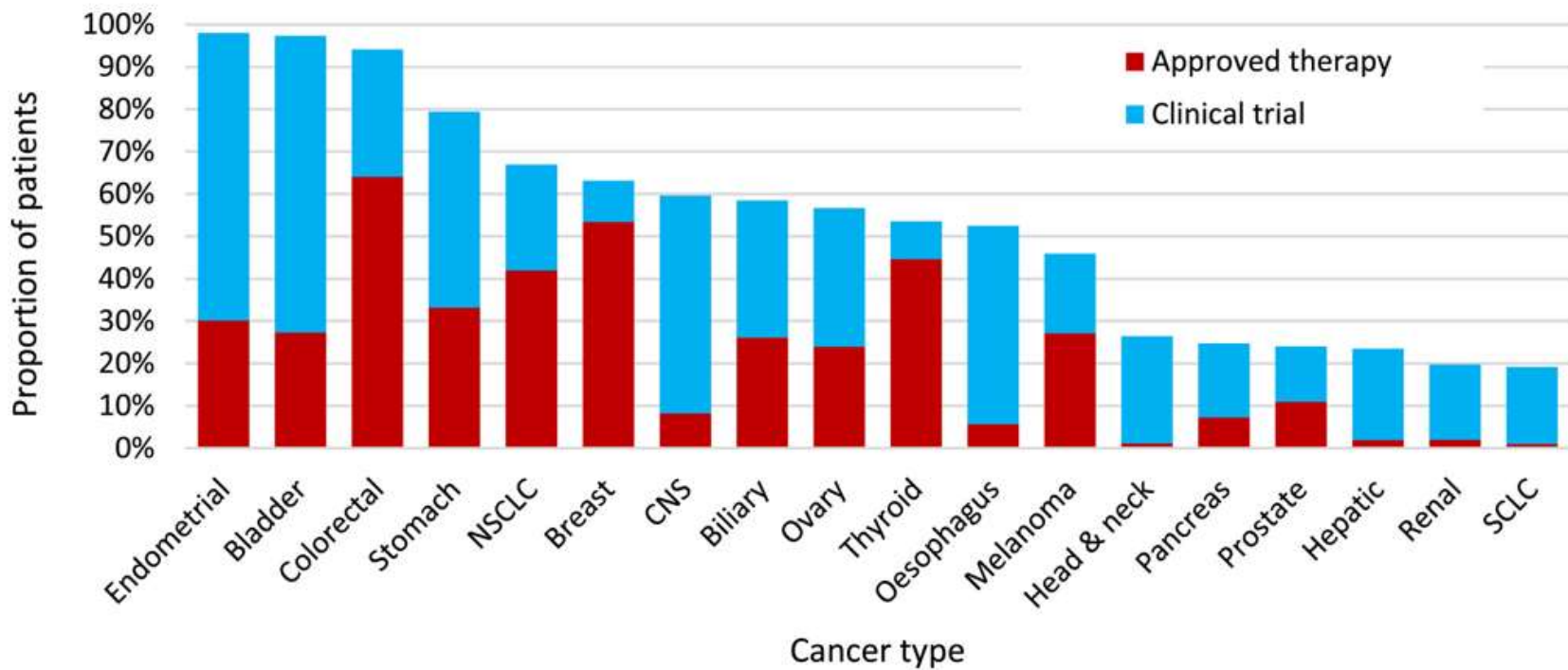
JAMA Oncol. 2021 Feb 1;7(2):230-237

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Precision Oncology

- Molecular characterization of tumors
 - Scientific use into a valuable clinical tool
- Advances in next-generation sequencing (NGS) technologies
- Novel diagnostic and therapeutic modalities
 - E.g. Tamoxifen, Trastuzumab in breast cancer
 - 115 new approvals for novel biomarker associated cancer therapies
- Interplay between modern diagnostics and molecularly driven therapeutics

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Proportion of patients with advanced cancer, by cancer type, who are potentially eligible for a biomarker-associated therapy or a biomarker-directed clinical trial

CA Cancer J Clin. 2024; 74(3): 264-285

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NGS recommendations

- NGS testing is now recommended by ASCO and ESMO in some cancer types and/or advanced cancers
- ASCO
 - Metastatic or advanced solid tumor patients with adequate performance status where genomic biomarker linked therapies are approved
- ESMO
 - Routine NGS in NSCLC, cholangiocarcinoma, prostate and ovarian cancers
 - Tumor mutational burden in well-and moderately differentiated neuroendocrine tumors, cervical, salivary, thyroid and vulvar cancers

J Clin Oncol. 2022 Apr 10;40(11):1231-1258
Ann Oncol. 2020 Nov;31(11):1491-1505

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Take Aways

- Patient genetic and genomic data can influence multidisciplinary cancer care
- Rapidly evolving information with respect to prognostication, treatment decisions and novel targeted therapeutics
- Disease site specific implications

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Thank you

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Disease Site Panel

- Thyroid - Jennifer Rosen, MD, FACS, Washington, DC
- Lung - Timothy Mullett, MD, MBA, FACS, Lexington, KY
- Colon - Nell Maloney Patel, MD, New Brunswick, NJ
- Breast – Katharine Yao, MD, FACS, Evanston, IL

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