

Defense Health Agency (DHA) Clinical Communities Speaker Series

22 April 2021 CCSS: Children and Youth in Transition

S02: Application of Genetics in Clinical Practice

Resource List

"The known unknown: the challenges of genetic variants of uncertain significance in clinical practice" (2017) is an article which provides pathways forward in dealing with the challenges in utilizing genetic testing in a clinical environment. The author noted that the genetic testing technology has been advancing rapidly and completion of the Human Genome Project in 2003 was a great accomplishment, however the ability to analyze and interpret the resulting data has not kept the same pace. Due to the inability to evaluate all results fully there are often variants of uncertain significance (VUSs). VUSs may not have clinical meaning now, however with more data and more time VUSs are regularly reclassified. This article reviews the many benefits to pursuing genetic testing, as well as the challenges that clinicians may face in determining how best to use the genetic information.

The American College of Medical Genetics and Genomics (ACGM) produces "ACT Sheets and Algorithms" (2021) to provide resources for health care providers that can better inform clinical decision making. These ACT Sheets include information on genetic conditions identified in newborn screenings, carrier testing, diagnostic testing, family history and more. These resources describe what options or actions are available to the health care provider in the short term, as well as providing an algorithm that demonstrates the basic steps involved in diagnosing conditions.

"Cancer Prevention and Control: Then and Now" (2018) is a paper which reviews the early detection and prevention of cancer, as well as cancer survivorship. In 1971 the National Cancer Act was signed by President Nixon with the goal of controlling or reducing the risk for cancer. These steps took place before the human genome was sequenced, and before the cause of cancer was understood. It has not been 50 years since the War on Cancer began, and this paper looks at the past steps and measures that have been taken.

The Centers for Disease Control and Prevention (CDC) has a <u>Genomics and Precision Health</u> page that provides weekly updates. They also support their 2021 Genomics and Precision Health Webinar Series which highlights global challenges and trends in precision health. Information ranges from Autism, Genetics and Family History to COVID-19 and Health Equity.

Dr. Hu and Dr. Wilkerson are Uniformed Services University (USU) scientists who were recognized by the American Association for Cancer Research (AACR) for their respective work in cataloging genomic changes associated with specific types of cancer. They were both members of The Cancer Genomic Atlas (TCGA) Team and they both received the AACR 2020 Team Science Award. The article USU Genetics Researchers Receive National Award for Cancer Genome Work (2020) discusses both of their roles at USU and their contributions to their research teams.



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References

American College of Medical Genetics and Genomics (ACMG). (2021). ACT Sheets and Algorithms.

https://www.acmg.net/ACMG/Medical-Genetics-Practice-Resources/ACT_Sheets and Algorithms/ACMG/Medical-Genetics-Practice-Resources/ACT_Sheets and Algorithms.aspx?hkey=9d6bce5a-182e-42a6-84a5-b2d88240c508

Centers for Disease Control and Prevention (CDC). (2021). *Genomics and Precision Health*. https://www.cdc.gov/genomics/update/current.htm

Ganz, P.A. (2018). Cancer prevention and control: Then and now. *NAM Perspectives*. Commentary, National Academy of Medicine, Washington, D.C. https://doi.org/10.31478/201810f

Hoffman-Andrews, L. (2017). The known unknown: the challenges of genetic variants of uncertain significance in clinical practice. *Journal of Law and the Biosciences*, 4(3), 648-657. https://doi.org/10.1093/jlb/lsx038

Holland, S. (2020). USU Genetics Researchers Receive National Award for Cancer Genome Work. *Health.mil*. https://www.health.mil/News/Articles/2020/07/07/USU-Genetics-Researchers-Receive-National-Award-for-Cancer-Genome-Work