難病・遺伝医学セミナー

日時：2018 年 6 月 21 日（木） 18 時～
場所：順天堂大学 10 号館 1 階 105 カンファレンスルーム

講演 1：Color Genomics Speaker

Recent advancements in next generation sequencing (NGS) based multi-gene testing panels in hereditary cancer clinical diagnosis

講師紹介

Lily Servais, *MS, LCGC*
Senior genetic counselor
Color Genomics

座長：岡崎康司（難病の診断と治療研究センター）
司会：新井正美（ゲノム診療センター）

Lily Servais, MS, LCGC is a senior genetic counselor at Color Genomics. She has been practicing as a genetic counselor in the San Francisco Bay Area since 2010 and specializes in hereditary cancer. Before joining Color she was the head genetic counselor of the El Camino Hospital's Genomic Medicine Institute. She received her bachelor’s degree in genetics and biology from the University of Wisconsin-Madison and her masters in genetic counseling from California State University-Stanislaus. She has had research published in Cancer, Journal of Community Genetics and ASCO Annual Meeting Proceedings. She currently resides in Oakland, California.

Abstract

- **Introduction to traditional practices on hereditary cancer testing**

Approximately 5–10% of all cancers are associated with hereditary cancer syndromes, the majority of which are inherited in an autosomal–dominant manner with high–to–moderate penetrance. As such, current recommendations for genetic testing of cancer susceptibility genes are primarily based on family and personal history of cancer. However, because of phenotypic variability, age–related penetrance, and gender–specific cancer risks, some carriers may be missed.

- **New broad–base cancer screening approach**

Clinical utility studies to date have largely focused on these high–risk and affected populations, and thus, the clinical utility of genetic testing in a broader population has yet to be fully defined. To address this, we developed and validated a 30–gene NGS panel for hereditary breast, ovarian, uterine/endometrial, colorectal, melanoma, pancreatic, prostate, and stomach cancer that was offered through a low cost, easy access delivery model.

- **Updates on recent multi–population cancer screening results including patients from Chinese and other Asian populations**

Here, we report the results of individuals who received this genetic test, providing data on the frequency and spectrum of pathogenic variants, including associations of high frequency alleles and cancer phenotypes both in the US population and Asian population.

- **Potential impacts on current recommendations for hereditary cancer genetic testing and screening**

We also evaluate the results with respect to the current recommendations for genetic testing and screening for hereditary cancer risk provided by the National Comprehensive Cancer Network (NCCN).

講演 2：OneOme Speaker

Personalizing medication management - insight into the clinical benefits of implementing pharmacogenomic testing into clinical practice

講師紹介

Ryan Gregg, *PhD*
Medical Science Liaison
OneOme

座長：岡﨑康司（難病の診断と治療研究センター）
司会：新井正美（ゲノム診療センター）

Ryan Gregg, PhD., is a Medical Science Liaison at OneOme, serving as OneOme's field medical resource for the United States, and internationally. He obtained a Bachelors of Science degree with Honors in Forensic Chemistry from the University of Mississippi, and a Ph.D. in Pharmacology from the Center for Substance Abuse and Research at Temple University School of Medicine in Philadelphia, where his thesis work focused on pre-clinical drug discovery and development for the treatment of substance use disorders.

Ryan's clinical experience includes academic and pharmaceutical research in psychiatry, hematology, and oncology. His two primary responsibilities at OneOme are clinician education and training around medication management with pharmacogenomic testing, and implementing ongoing clinical trials investigating RightMed pharmacogenomic testing in oncology and pain management as part of OneOme's Medical Affairs & Clinical Trials teams.

Abstract

- **Introduction to the basic principles of pharmacogenomics**

Selecting the right medication and dose to treat a patient's condition is among the most important decisions made by physicians, yet many of the most commonly prescribed medications do not work effectively in certain populations of patients. Pharmacogenomics is the science underlying the impact of specific genetic variants on the metabolism and mechanism of action of different medications, and using pharmacogenomic testing, physicians can now gain further insight into which medications may pose additional risk of toxicity, lack of efficacy, or risk of side effects.

- **Current pharmacogenomic applications in cardiology, psychiatry and oncology practices**

Highlighting medications that are impacted by pharmacogenomic variants in cardiology, psychiatry, and oncology, as well as citing supporting clinical evidence

- **Clinical case studies**

Presenting patient cases where pharmacogenomic testing may be beneficial in optimizing medication selection and dosing

- **Supporting physicians to adopt pharmacogenomics at point of prescriptions**

Discussing the various services available to physicians interested in performing OneOme's RightMed pharmacogenomic testing, including the use of OneOme's unique physician web portal coupled with Rainbow Genomics' real-time support.

主催：難病の診断と治療研究センター／ゲノム診療センター
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