

October 21, 2015



OPKO Health's GeneDx business Presenting at National Society of Genetic Counselors Annual Meeting

Focus is on Genetic Diagnosis of Rare Disease

PITTSBURGH--(BUSINESS WIRE)-- OPKO Health, Inc. (NYSE:OPK) announced today that GeneDx, a business unit of OPKO will be presenting at the National Society of Genetic Counselors (NSGC) 34th Annual Education Conference in Pittsburgh, PA. GeneDx staff and genetic counselors will participate at the conference as individual speakers, poster presenters and exhibitors. GeneDx is also sponsoring a networking reception with partners and colleagues. Details are below:

GeneDx BOOTH: #115

[RSVP here](#) for the Annual GeneDx Happy Hour

When: Thursday, October 22, at 8:00pm

Where: Il Tetto Rooftop Bar at Sienna Mercato

GeneDx Lunch Session and Presentation

Diagnostic Testing, Evolving Phenotypes, and Impact on Patient Care: A GeneDx Update on XomeDxXpress and Inherited Cancer Testing

When: Friday, October 23 at 11:45 pm

Where: Allegheny Grand Ballroom, Westin Hotel

Who: Sara Knapke, MS, CGC; Audra Bettinelli, MS, CGC; Stephanie DeWard, MS, CGC

GeneDx Individual Speaker Presentations

Pre-Conference Symposium - *Diagnostic Exome Sequencing as the Standard of Care*

When: Wednesday, October 21 at 9:00 am

Where: Room 315/316

Who: Jane Juusola, PhD, FACMG

Concurrent Paper Session (Clinical Care: Pediatrics & Adult Track) - *The Clinical Utility of a Multi-Gene Panel for Neuromuscular Disorders (#1311)*

When: Saturday, October 24 at 3:15 pm

Where: Room 315/316

Who: Meg Bradbury, MS, CGC, MSHS

GeneDx Poster Presentations

NOTE: Odd Numbered Posters: Thursday, October 22, 2015 2:00 pm – 3:00 pm

Even Numbered Posters: Friday, October 23, 2015 1:15 pm – 2:15 pm

Poster# 64

Comparing Yields and Referral Criteria for the Lynch/Colorectal High Risk Panel and the Colorectal Cancer Panel

Anna McGill, MS, LCGC

Poster# 78

Yield of Genetic Testing for Hereditary Cancer Among Male Patients

Kristin Theobald, MS, LCGC

Poster# 153

Genetic Testing Strategies for Patients With Epilepsy and Neurodevelopmental Disorders

Amy Decker, MS, CGC

Poster# 154

Mutations in SPATA5 Are Associated With a Novel Autosomal Recessive Disorder of Microcephaly, Intellectual Disability, Seizures and Hearing Loss

Stephanie DeWard, MS, CGC

Poster# 169

Whole Exome Sequencing Identifies the First PANX1 Germline Mutation in an Individual with Intellectual Disability, Hearing Loss, Endocrine Dysfunction and Skeletal Abnormalities

Kara Levine, MS, CGC

Poster# 192

De Novo Pathogenic Variants in DDX3X Are a Novel Cause of Intellectual Disability in Females

Leah Williams, MS, CGC

Poster# 194

Whole Exome Sequencing Identifies POGZ Mutations as a Cause of Neurodevelopmental Disorders and Microcephaly

Nora Alexander, MS, CGC

Poster# 218

Mutations in ARID2 Are Associated with Syndromic Intellectual Disabilities

Rebecca Willaert, MS, CGC

Poster# 268

Compound Heterozygosity of Two MECP2 Deletions with Paternal Inheritance of a Late-Truncating Mutation in a Female With Atypical Rett Syndrome

Dana Stolar, MS, CGC

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