

PCD NEWSLETTER

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DIRECTOR'S CORNER

Dear members of the Division of Precision and Computational Diagnostics (PCD),



Welcome to the thirteenth issue of our Divisional Newsletter. We are excited to welcome three new members to our division. Heather Mitchell, BS, joined the CPD team as a technologist, Chris Herzog, PhD, joined the CPD team as a Variant Analyst, and Tom Dibello, BA as a new technologist in the Molecular Pathology Lab. We are excited to have them join the PCD and look forward to working with them.

In this month's letter I wanted to take the time to recognize our various staff members that have shown great passion in and outside of the work place. In particular, Amanda Rosen and Chris Leid. Amanda brings together art and science in this year's exhibit at the Perlman Center for Advanced Medicine. This exhibit is dedicated to Bernett L. Johnson Jr., a scientist that exemplified science is an art form in its own right. Chris strives to complete one of the items on his bucket list by traveling to Africa this month. His goal is to visit all seven continents within the next seven years. So far he has made it to five out of the seven continents. We acknowledge his great progress and applaud his tenacity to accomplish his goals. These achievements are inspiring and instructive for living out goals and passions.

As a division we should always recognize professional growth as well as personal growth. With that said we are looking forward to the future and the accomplishments we can all achieve together.

Thank you for being excellent at what you do and your continued contributions to the service of our patients. Best wishes for a productive remainder of the year.

Kojo Elenitoba-Johnson, MD

PUBLICATIONS

Reese P, Van Deerlin VM, Gentile C, Smith J, et al. **Twelve-Month Outcomes After Transplant of Hepatitis C-Infected Kidneys Into Uninfected Recipients: A Single-Group Trial.** *Ann Intern Med* 2018 Sep 4;169(5):273-281.

This manuscript summarizes Penn's experience with the first 20 HCV+ kidney transplants in the THINKER study. The conclusion is that all 20 HCV-negative recipients of HCV-infected kidneys experienced HCV cure, good quality of life, and excellent renal function. Kidneys from HCV-infected donors may be a valuable transplant resource.

Suh E, Grando K, Van Deerlin VM. **Validation of a Long-Read PCR Assay for Sensitive Detection and Sizing of C9orf72 Hexanucleotide Repeat Expansions.** *J Mol Diagn.* 2018 Nov;20(6):871-882.

Described here is work in Vivianna Van Deerlin's research lab on validating a commercial assay for C9orf72.

Goldman JS, Van Deerlin VM. **Alzheimer's Disease and Frontotemporal Dementia: The Current State of Genetics and Genetic Testing Since the Advent of Next-Generation Sequencing.** *Mol Diagn Ther.* 2018 Oct;22(5):505-513.

Co-written by Vivianna Van Deerlin and a Genetic Counselor at Columbia University, this manuscript elucidates the current state of genetics and genetic testing with next generation sequencing for Alzheimer disease and frontotemporal dementia.

WELCOME

**Heather Mitchell,
BA, MB (ASCP) CM**



Heather is a Clinical Genomics Lab Technician at the Center for Personalized Diagnostics.

Heather received her Bachelor of Arts degree in Biology from Rutgers University. She comes to us with 15+ years experience in clinical CLIA, CAP, and NYSDOH accredited clinical molecular laboratories either diagnosing infectious diseases or cancers of unknown primary. She obtained the American Society for Clinical Pathology (ASCP) Molecular Biology Technologist (MB) certification in 2015.

Heather is a certified pistol and rifle instructor as well as a Range Safety Officer. She utilizes her certifications as a local chapter leader and instructor for a national organization of women shooters. She enjoys going to sporting events like the Flyers and Phillies and participating in the sports of snowboarding and CrossFit.

Chris Herzog, PhD

Chris is a Variant Analyst at the Center for Personalized Diagnostics.

Chris is taking on the role as our newest Variant Analyst. Chris received his PhD in Molecular Cancer Research. With that said he has several years of experience in the oncology field, as well as variant review based assessments.

Tom Dibello, BA

Tom is a medical technologist in Molecular Pathology.



Tom graduated from Rutgers University in Camden, NJ, with a BA. Previously, he worked for the National Molecular Lab of the American Red Cross as a technologist III. Tom mostly, spends time with his fiancé and two boys. He has a six year old son and a 1 month old son. He loves sports, especially soccer. Also, he is a huge Eagles fan. Tom enjoys the outdoors and camping. He was born and lived in Frankford for 25 years. Now he resides in Gibbsboro, New Jersey. Tom is a technologist at HUP in the molecular pathology lab and is looking forward to starting a career at Penn.

FANTASY FOOTBALL UPDATE

PCD Battle Royale is Underway!

This is the first year of the PCD fantasy football league and it is shaping up to be neck and neck right up until the playoffs! The league, run by commissioner Ashkan Bigdeli, includes ten teams from various members of the PCD, split into two conferences. See the standings and other league highlights below and to the right!

Other members of the division already mentioned wanting to join next year - we welcome everyone to play, no prior knowledge about football required!

STANDINGS IN THE EAST DIVISION

1. Akshay Chitturi
2. Jackie Roth
3. Ashkan Bigdeli
4. Amanda Rosen
5. David Lieberman
6. Robyn Sussman

STANDINGS IN THE WEST DIVISION

1. Chris Orr
2. Joe Milano
3. Sal Priore
4. Jennifer Morrissette



Highest weekly score:
David Lieberman
(170 points!)

Most points scored overall:
Akshay Chitturi (902 points!)

First player drafted:
Todd Gurley II (Team Chitturi)

Creative team names include:
*Team Huge Deal (David)
*Perfect McChampionFace (Ashkan)
*The Underdog (Amanda)
*JuJu Wanna Build A Snowman (Jackie)

TEAM LOGOS



SEMINARS

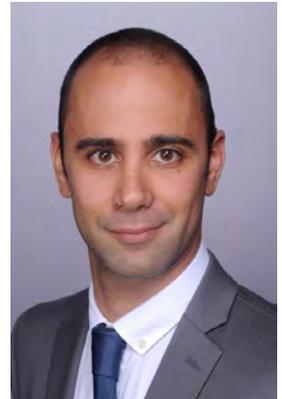


October 4th: Robyn Sussman, PhD delivered an AMP webinar regarding "Mutational Signatures in Cytogenetics Risk Groups of Newly Diagnosed Acute Myeloid Leukemia (AML) and Myelodysplastic Syndrome (MDS)."

October 5th: Robyn Sussman, PhD presented "Kicking the tires before driving off to clinical validation: using reference standards to define performance characteristics of new NGS assays" at the World Clinical Biomarkers and Companion Diagnostics meeting in Boston.

October 19th: Robyn Sussman, PhD presented "Mutational Signatures in Cytogenetic Risk Groups of De Novo AML and MDS" at the American Society for Human Genetics annual meeting in San Diego.

November 8th: Ashkan Bigdeli, MS and Robyn Sussman, PhD presented in a Genomeweb webinar "Defining the Performance Characteristics of New NGS Assays with Reference Standards."





THE CLOUDS OF UHURU
Uhuru Peak is the highest point in Africa and the summit point of Mount Kilimanjaro, the tallest free standing mountain on Earth. The trek to the 19,341' high summit took Chris five days. He has aspirations to visit all seven continents within the next seven years. From the ebbing snow cap of Mount Kilimanjaro to the disappearing Great Barrier Reef in Australia, Chris is on a race with time as the landscape of the Earth changes dramatically. His visit to Tanzania, Africa marks his 5th continent, with the glaciers of Antarctica and cities of Europe remaining.

**CHRIS LEID
TRAVELS
AROUND
THE WORLD**



Congrats!

**JESSICA AND
CARMELLA**

WE WISH YOU THE BEST



ART & LIFE

An exhibit in
memorium

Amanda Rosen presents
her work, "Outer Limits."

This year's 'Celebration of Art and Life,' which opened this September, featured the artistic talents of Penn Medicine patients, faculty, staff, and CPD's very own Amanda Rosen. The juried annual art exhibition is dedicated to the memory of Bernett L. Johnson, Jr., M.D., former Chief Medical Officer of HUP, who was also an accomplished artist.

Amanda's painting is an acrylic and mix media piece. She first painted the canvas black, then created a top layer consisting of a mixture of colors. She rotated the canvas to move the paint and colors to fill any negative space. During this process, natural lines and curves formed, to which she added beads while the paint was still wet. This emphasized the the lines and curves and gave the paintings more movement and depth. Most of her paintings are influenced by modern art/artists, specifically with regard to their use of bright and bold color combinations. With this style of painting, she can never predict the final outcome, which is part of the fun. Even when she uses the same initial colors, the paintings can diverge significantly.



JASON ROSENBAUM, MD

CONVERSATIONS

IN THE CENTER FOR PERSONALIZED DIAGNOSTICS

Jason is an Associate Professor in the Division of Precision and Computational Diagnostics at the University of Pennsylvania. He grew up in the San Francisco Bay Area in the halcyon days before "tech bro" entered the lexicon and forest fires became ubiquitous, and was an undergraduate at the University of California Berkeley. He studied medicine at Northwestern University before pursuing AP/CP training at University of Wisconsin and molecular genetic pathology training at Washington University. He is known for his ability to generate esoteric knowledge on demand and with sardonic irreverence. Over Samarian Sunsets, we discussed his career path and the future of the field. And, dont worry, I made sure to deploy Ferengi Rules of Acquisition #33. -Priya Velu

What inspired you to choose molecular genetic pathology over a career in comedy?

I thought the entertainment industry relied too much on good luck and knowing the right people, so I wanted the security of a career in science. As it turned out, the joke ended up being on me.

Technologies are always rapidly changing in molecular genetics. How do you see the next five to ten years evolving for clinical laboratories and medical applications, especially with regards to new advances in sequencing (fourth-generation) and resolution of sequencing (single-molecule)?

30 years ago, radiologists probably imagined a future in which everyone would routinely get full-body MRIs. Now, the imagined future is one in which everyone routinely has their genome sequenced. The fact that something is technologically possible does not necessarily mean it is clinically useful or scalable. The more data each advance brings, the more challenging the interpretation becomes, and the more risk there is for overinterpretation and unnecessary intervention. There will always be a place for new technologies that advance patient care, but the key is that they have to demonstrate clinical utility. I think both fourth generation sequencing and single molecule sequencing may find clinical uses, but I have yet to see a clear "killer app" for either technology that offers a clear, cost-effective, scalable clinical advantage over existing techniques.

What are your opinions on direct-to-consumer testing (DCT)? Though fraught with liabilities and issues in interpretation, do you think there is any value in democratization of sequencing?

I think there is value in increasing public awareness of genetics and genomics, and certainly in reduced costs to consumers. I have deep concerns about "democratization" of interpretation; in my experience even trained professionals with experience in molecular biology may not have a good depth of understanding of the pitfalls associated with laboratory testing, such as relative risk vs. overall risk, pre-test probability, analytical vs. clinical sensitivity, etc. No matter how much reading a consumer is required to do, and no matter how many waivers and attestations of understanding they sign, I am not convinced that these tests should be offered outside of the context of a trained medical professional.

The trajectory of telecommunications use in third-world countries had a "leapfrog" phenomenon, where many inhabitants completely skipped traditional land-lines and jumped to cell phone usage. Do you see something similar potentially happening in medicine with DCT and computational imaging technologies?

Possibly, but I'm not sure that's a good thing, due in part to the difficulty in rendering an appropriate interpretation. Another example of a "leapfrog" phenomenon comes from Europe, where there has been a long-standing resistance to American-style drip-brewed coffee, until the advent of coffee pods. I'm told the pods are becoming ubiquitous. But pods offer inferior coffee, generally speaking, and have negative environmental consequences. "Leapfrogging" is not always desirable – sometimes a technology is better envisioned and implemented as one option in a spectrum of possibilities than as an endpoint.

If you were tasked to design a tricorder, what functionalities would you include?

- 1) Comprehensive genomic profiling
- 2) Automatic question answering
- 3) Philately (stamp-collecting, says my tricorder)

You often have laugh inducing one-liners. Do you think of them on the spot or have them ready and waiting in the wings? In general, what inspires your humor?

I use my tricorder.

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