



## Rare Disease Day Inspires, Offers Resources and Hope

BY DANA TALESNIK

People with rare diseases and their loved ones, clinicians, researchers and patient advocates were among the more than 2,000 participants in this year's Rare Disease Day at NIH on Feb. 28. The Natcher Conference Center was abuzz with hundreds of in-person attendees while many more tuned in via videocast for the hybrid event, co-sponsored by the National Center for Advancing Translational Sciences (NCATS) and the Clinical Center.

The day's sessions expounded on rare disease clinical research and resources as well as ways to improve the patient experience. Several panels focused on an

often-underserved population in the rare disease world—adolescents and young adults.



NCATS Director Dr. Joni Rutter (r) poses with Grace Moon of Mitre, a non-profit that supports NIH and other federal health projects.

PHOTO: NCATS

During welcoming remarks, Clinical Center CEO Dr. James Gilman said, "This is a day when the head acquires new information and knowledge. It's also a day when the heart renews hope and finds sources of new inspiration."

A rare disease is one that affects fewer than 200,000 people in the U.S. There are

more than 10,000 different rare diseases affecting more than 30 million people nationwide and more than 350 million people worldwide. Most rare diseases have few if any effective treatments.

Critical to finding new therapies is securing science funding, an effort driven by a cadre of bipartisan policymakers and legislators who support the research. Several members of the Rare Disease Congressional Caucus delivered virtual remarks, voicing their support for continued research to find new therapies for rare ailments.

"This day is about pausing to remember loved ones we've lost," said NCATS Director Dr. Joni Rutter, "to celebrate the big and small wins, to support those who work tirelessly in caring for people with rare diseases, to honor all people with a rare disease and recognize that struggle, and to hear about new ideas and projects that spell hope."

SEE RARE DISEASE, PAGE 6



NIH hosts White House Covid Response Team. See story, p. 3.

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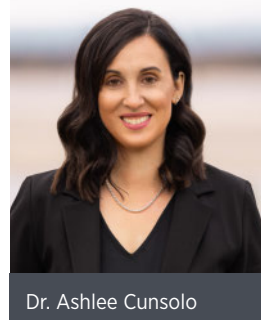
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## ECOLOGICAL GRIEF, ANXIETY Cunsolo Discusses Mental Health Impact of Climate Change

BY AMBER SNYDER

Climate change has many far-reaching effects on physical health—such as cardiovascular and respiratory disease, and other injuries/illnesses caused by extreme weather events—but how does it affect mental health?

Dr. Ashlee Cunsolo studies ecological grief and anxiety globally, and has spent 15 years working with Inuit communities in Canada on this topic. She spoke recently with National Institute of Mental Health Director



Dr. Ashlee Cunsolo

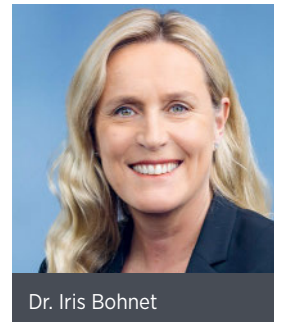
## AWARENESS IS NOT ENOUGH Bohnet Discusses Gender Bias in the Workplace

BY ERIC BOCK

Raising awareness is only the first step to addressing gender bias in the workplace, said Dr. Iris Bohnet during a recent Women's Scientists Advisors 30th Anniversary Seminar Series lecture.

"We have to apply real treatment to get real impacts," said Bohnet, Albert Pratt professor of business and government and co-director of the Women and Public Policy Program at Harvard University's Kennedy School of Government.

Implicit or unconscious bias shapes how



Dr. Iris Bohnet

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### Celebrate DNA Day on Apr. 25

In recognition of National DNA Day, the National Human Genome Research Institute (NHGRI) will host a symposium to commemorate two special milestones: the 20th anniversary of the Human Genome Project's completion and the 70th anniversary of the discovery of the DNA double helix.

The symposium will begin on Tuesday, Apr. 25 at 10 a.m. ET in Lipsett Amphitheater. Attendees can also register to join virtually at <https://bit.ly/3nD7duw>.

Symposium attendees will explore the evolution and future of genomics research, learn about the greater impacts of genomics on society and discover the wide array of careers in genetics and genomics—from scientists to social media specialists.

Former NIH Director Dr. Francis Collins will deliver the 2023 Louise M. Slaughter National DNA Day Lecture. The annual lecture honors the life and legacy of the late New York congresswoman, who was a strong advocate for genomics research. She was also responsible for passing the 2003 resolution in the U.S. House of Representatives that created National DNA Day. NHGRI established the Slaughter lecture in 2018.

National DNA Day commemorates successful completion of the Human Genome Project in 2003 and the discovery of DNA's double helix in 1953. For more information on National DNA Day, visit <https://www.genome.gov/dna-day>.

### NIMHD To Host 5K Walk/Run/Roll

Apr. 18

The National Institute on Minority Health and Health Disparities will host the "Minority Health 5K Walk/Run/Roll" on NIH's Bethesda campus at 11:30 a.m. ET on Tuesday, Apr. 18, in observance of National Minority Health Month.

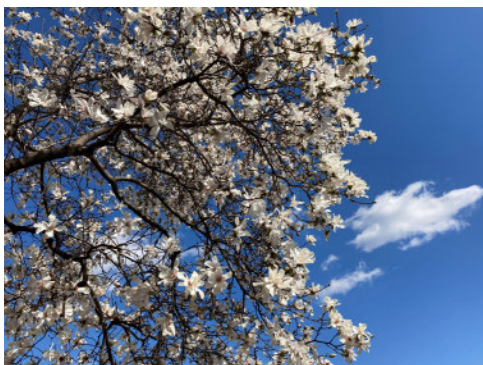
The course will begin (rain or shine) on the lawn in front of Bldg. 1. Participants will exit campus, travel the perimeter and return through security. To register, visit <https://forms.office.com/g/UCMbY6yRt4>.

To sponsor a water station and show your institute/center/office spirit, contact Shelly Pollard at Shelly.Pollard@nih.gov.



At the 2019 Minority Health 5K Walk/Run/Roll, then-U.S. Surgeon General Jerome Adams takes a selfie with crowd.

PHOTO: CHIA-CHI CHARLIE CHANG



**Signs of spring.** Above at left, a blooming tree. At right, yellow flowers in Seneca Creek State Park, Gaithersburg, Md. PHOTOS: DEBORAH HENKEN/NICHD Below, closer to home, fowl warning signs outside the Clinical Center also usher in the new season. PHOTOS: ERIN BUTLER/OD OCPL



The 2023 theme for the month is "Better Health Through Better Understanding" and focuses on how meeting cultural and linguistic needs can improve health outcomes.

Individuals who need sign language interpreters and/or reasonable accommodation to participate should contact Edgar Dews at [edgar.dews@nih.gov](mailto:edgar.dews@nih.gov) or (301) 594-8424. Requests should be made at least five days in advance of the event.

### All of Us Celebrates Fifth Anniversary

When the All of Us Research Program launched on May 6, 2018, it did so with the singular goal of building and nurturing relationships with potential participants, community partners, researchers and advocates from around the country. People from all backgrounds joined six live-streamed community events, including on the grounds of Ford Field in Detroit, the pews of Abyssinian Baptist Church in Harlem and the stalls of a town-wide farmer's market in Pasco, Wash., to learn about the promise of precision medicine.

The program was born from an aspirational idea: to realize a future of prevention, care and treatment that is tailored to the individual.

The goals of this precision

health initiative may have seemed ambitious in 2018 but are now within reach as the program marches closer to enrolling 1 million people who reflect the diversity of the United States, including 80% from populations historically underrepresented in medical research—a feat few research efforts could attempt.

These participants are fueling discovery led by more than 3,700 active registered researchers conducting more than 4,300 studies.

While these are impressive accomplishments for a research program of this size and scope, there is still so much more we can achieve—but it can only be done with the help of as many participants as possible.

Join the All of Us team on NIH's Bethesda campus May 8-12, when the program's Journey bus (shown below) will be on site. There, you can enroll in the program, answer health surveys, have biosamples (blood and urine) taken, or just learn more about All of Us. Mark your calendar and stay tuned for more information. You can even take the first few steps to enroll by visiting <https://allofus.nih.gov/atNIH>.





Dr. Lawrence Tabak (l) and other NIH leaders meet with Dr. Ashish Jha (r), White House Covid-19 response coordinator, and members of his team on Mar. 23. At center, the group sees a room on the CC special clinical studies unit. At right, Jha gets a demo of cell preparation by Alexandra Woo, NIH postbaccalaureate fellow.



Participating in the visit are (from l) Dr. Hugh Auchincloss, Dr. Karin Bok, Dr. Theodore Pierson, Jha, Dr. Richard Koup, Tabak, Dr. Tara Schwetz and Dr. Carl Dieffenbach.

PHOTOS: CHIA-CHI CHARLIE CHANG

Then, Dr. Cliff Lane, NIAID deputy director for clinical research, led them for a look inside the special clinical studies unit. NIAID Senior Investigator Dr. Richard Davey, medical director of the unit, talked about its unique features and the range of disorders and conditions treated there.

Later, NIAMS Deputy Scientific Director Dr. Mariana Kaplan described her laboratory research on Covid-19 in systemic autoimmune diseases. In her National Institute of Arthritis and Musculoskeletal and Skin Diseases lab, Dr. William Ambler, NIH clinical fellow, and Alexandra Woo, NIH postbaccalaureate fellow, demonstrated a cell preparation.

At a final briefing in the Medical Board Room, Dr. Gary Gibbons, director, National Heart, Lung and Blood Institute (NHLBI); Dr. Walter Koroshetz, director, National Institute of Neurological Disorders and Stroke; and Dr. Amy Patterson, NHLBI deputy director for clinical research and

strategic initiatives, offered updates on the RECOVER initiative.

NIH's Office of Communications and Public Liaison managed the visit. **R**



ON THE COVER: 3D rendering of yeast cells. Newly discovered gene helps some yeast endure toxins and can help scientists understand toxin resistance.

IMAGE: ERNESTO DEL AGUILA III/NHGRI

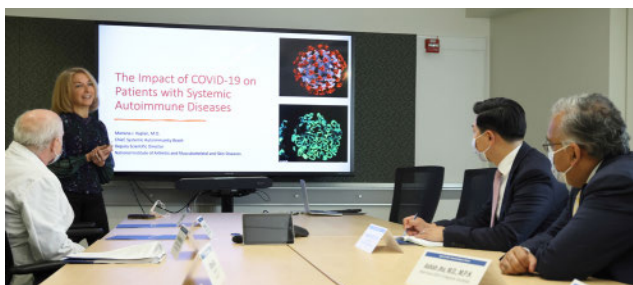
## Jha, White House Covid Response Team Visit

White House Covid-19 Response Coordinator Dr. Ashish Jha and several members of his team visited NIH on Mar. 23 for meetings and tours focused on NIH's current Covid activities.

The visit began at the Vaccine Research Center, where he was briefed by Dr. Hugh Auchincloss, acting director, National Institute of Allergy and Infectious Diseases (NIAID) and Dr. Richard Koup, VRC acting director. Auchincloss and Koup provided a VRC overview and discussed the facility's role in developing Covid-19 vaccines and other vaccines.

In addition, Dr. Bruce Tromberg, director of the National Institute of Biomedical Imaging and Bioengineering, gave an update on the Rapid Acceleration of Diagnostics (RADx) initiative.

Jha's team also toured the Clinical Research Center. The group was met in the CRC atrium by Clinical Center CEO Dr. James Gilman, who gave an overview of the building's layout.



Dr. Mariana Kaplan describes her laboratory research.



In a final briefing in the Medical Board Room, Jha (r) discusses the latest on the RECOVER initiative.

## The NIH Record

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## Eco-Grief

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Dr. Joshua Gordon as part of her lecture, “Ecological Grief and Anxiety: A Healthy Response to Climate Change.” The talk was part of the NIMH Director’s Innovation Speaker Series.

Cunsolo has been researching ecological grief and anxiety since 2008. She and a team of researchers were invited by the Inuit community of Rigolet on the North Coast of Labrador, Canada, to study the broader impacts of climate change on health, but Cunsolo pivoted to mental health research after asking community members a crucial question: How do you feel about the changes happening to your environment?

Labrador is one of the fastest-warming places on the planet and is the fastest-warming place in Canada. The climate there is warming almost four times faster than the global average and temperatures have already surpassed the 1.5°C average temperature increase that the rest of the world identifies as a limit under which to stay.

The Inuit “have been on the frontlines of climate change for decades,” Cunsolo said. That involves changes in sea ice, which is a major component of life for them. Inuit are “the People of the Sea Ice” and rely on months-long sea ice cover in winter for

things like hunting and traveling. But rapid warming is disrupting this system; sea ice extent in Labrador

has already declined by almost 75 percent. The so-called “highway of ice” used to last in previous times from approximately October to May, but now ice does not form until sometimes well into January and begins to melt in April.

One of the residents Cunsolo interviewed

★ ★ ★

*“People feel homesick for a home environment that’s no longer there.”*

-DR. ASHLEE CUNSULO

★ ★ ★

posed a profound, existential question in a documentary film, *Attutauniujuk Nunami/ Lament for the Land*: “If there’s no more sea ice, how can we be People of the Sea Ice?”

Depending on climate projections, Labrador is poised for an additional potential 6 to 11°C of warming, and the mood among Rigolet and Nunatsiavut inhabitants is full of concern and anxiety.

Inuit culture and well-being are deeply connected to the land. Having the ability to go out on the land—and particularly the ice—is “as much a part of our life as breathing,” one person told Cunsolo.

The “shoulder seasons” (when ice is melting/forming) inhibits people’s ability to get out on the land—and the length of the shoulder seasons are increasing as the climate warms. Such periods used to last only a couple weeks, but are now stretching six weeks or even longer, Cunsolo said.

Shoulder seasons are an “empty time” for people because they can’t enjoy the

outdoor territory. As a result, mental health professionals worry that residents may turn to harmful behavior in order to cope.

Australian researcher and philosopher Glenn Albrecht coined a term that has resonated with many people who are distressed by the changing climate: “solastalgia,” meaning “homesickness while you’re still at home.”

Cunsolo said she hears this sentiment from a lot of people in the region, particularly Elders.

“They haven’t moved but everything around has changed so much that it’s disorienting,” she explained. “People feel homesick for a home environment that’s no longer there.”

Many people around the world are also feeling grief and anxiety related to climate change. Researchers are still learning how to address this kind of slow and cumulative grief, Cunsolo said.

There are often mental health resources available for those experiencing natural disasters like wildfires and floods, but how do you learn to grieve “beyond the human” and what sort of resources are available to support those struggling with and experiencing ecological grief?

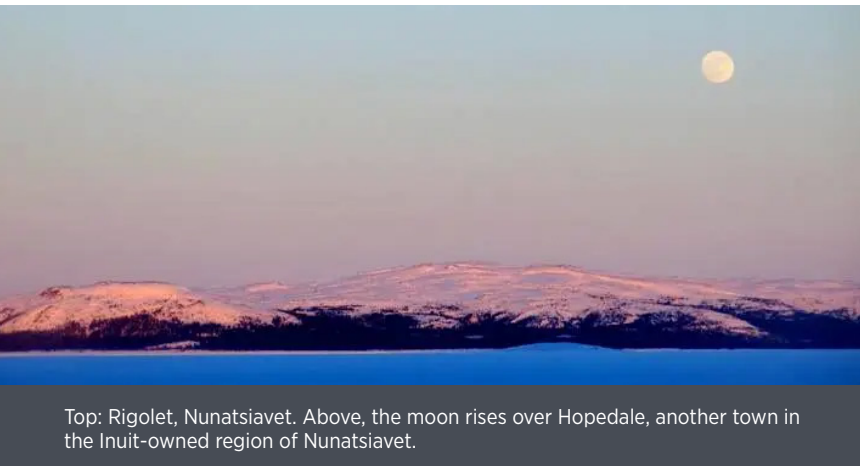
Cunsolo collaborated recently with Australian researcher Dr. Neville Ellis, who is studying emotional responses to extreme drought; they co-authored a paper on ecological grief. Study participants in both countries had amazingly similar responses to their respective ecological changes.

“You could take conversations and interviews with people from either place and replace sea ices with drought or vice versa, and you couldn’t tell [which group] the quotes were coming from,” Cunsolo said.

Ecological grief and anxiety can affect people from all walks of life, but one group that is drawing researchers’ focus around the world is young people.

A 2021 study of 10,000 people ages 16-25 from multiple countries surveyed participants about their feelings regarding climate change, as well as their thoughts on their government’s response to climate change.

About 84 percent of participants said they were moderately worried or higher, and many also reported a high proportion of negative thoughts, such as “people have failed the planet”—an opinion shared by 83



Top: Rigolet, Nunatsiavet. Above, the moon rises over Hopedale, another town in the Inuit-owned region of Nunatsiavet.

percent of participants. Mental health professionals are observing increasing numbers of young people who want to talk about their climate grief and anxiety.

So, how can people learn to cope as the climate crisis continues to make itself known?

Researchers are still working to understand the phenomenon's full effects on mental health, Cunsolo said. But, she also emphasized that feelings of ecological grief and anxiety are rational, reasonable responses to the climate crisis, and "embracing sorrow" is an important part of coping with the pain of climate change.

What do people need moving forward to learn to cope with this mental and emotional burden safely? According to Cunsolo, requirements include "accessible and safe spaces to explore these difficult emotional reactions, the political will to ensure that important strategies and supports are funded, and the research required to strengthen and support approaches of healing and resilience."

To view a recording of the lecture, visit <https://bit.ly/3McbYFB>. 

## VOLUNTEERS

### Insulin Study Recruits Volunteers

Did you know insulin is the primary hormone-regulating sugar in the blood? Think about enrolling as a healthy volunteer at NIDDK. Compensation will be provided. Contact the Clinical Center Office of Patient Recruitment at (866) 444-2214 (TTY users dial 711) or [ccopr@nih.gov](mailto:ccopr@nih.gov). Refer to study #76-DK-0006. Online: <https://bit.ly/3CUVs7T76DK0006>.

### Thyroid Study Needs Participants

Did you know your thyroid gland produces hormones that regulate body temperature, heart rate and metabolism? Physicians want to explore a better method to detect thyroid tumors by using a compound called 68Gallium-DOTATATE. This compound may bind to a tumor and make it visible during a PET/CT scan. The information will help guide future research and treatment. Contact the Clinical Center Office of Patient Recruitment at (866) 444-2214 (TTY users dial 711) or [ccopr@nih.gov](mailto:ccopr@nih.gov). Refer to study #000079-DK. Online: <https://bit.ly/3DQ6ADt000079DK>.

## NIMHD Seminar Looks at Past for Future of Health Disparities

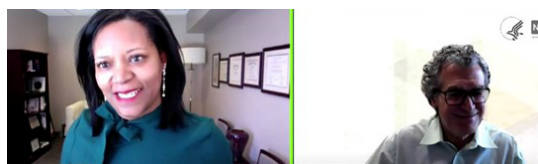
BY BETHANY HOFFMAN, SHELLY POLLARD, GINA ROUSSOS

The historical role of science and scientists in creating and perpetuating racism and racial health inequities was the topic of a recent NIMHD seminar that featured Dr. Consuelo Wilkins, senior vice president and senior associate dean for health equity and inclusive excellence at Vanderbilt University. "Although race is a sociopolitical construct, scientific researchers are still treating it as a biological justification for health inequities," she said in opening remarks.

Wilkins went on to discuss the continuing pervasiveness of racial and ethnic health disparities and the social and structural determinants that underlie them.

"In the last 20 years, disparities have narrowed very little," she said, noting how essential it is for researchers not to be "color blind" and to acknowledge race when studying health data. It is important because race does influence health outcomes, not necessarily by biology or genetics, but by differences in life experiences and conditions due to systemic racism.

Wilkins outlined how health leaders can address racial inequities, including reversing unjust practices and policies, making decisions based on diverse perspectives and connecting with frontline health care staff.



NIMHD Director Dr. Eliseo Pérez-Stable introduces Wilkins.



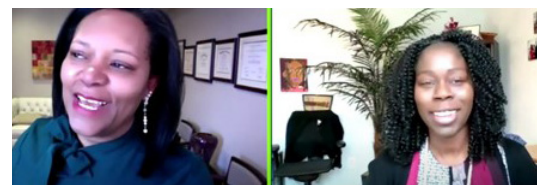
Wilkins suggested that early-career scientists not yet enmeshed in old practices are poised to create positive disruption in health disparities research by questioning the status quo.

For example, young scientists were instrumental in the call to remove race as a factor when calculating estimated glomerular filtration rate (eGFR) to diagnose stages of

kidney disease, a condition disproportionately affecting people in racial and ethnic minority populations such as African Americans.

Using race, a social construct, as a proxy for genetics, a biological factor, to calculate eGFR does not take into account the genetic diversity within racial and ethnic minority populations or the fact that many individuals identify as multiracial or multiethnic.

The scientific community can also take steps to promote racial equity in research, including improving transparency in clinical research enrollment and addressing exclusionary research practices; making investigators accountable for meeting scientifically valid diversity standards in their test groups, which should reflect the populations most affected by the disease being studied; investing in sustained, reciprocal relationships with communities that have been marginalized; and developing evidence-based guidance to inform inclusive research participation.



NIMHD Deputy Director Dr. Monica Webb Hooper (r) leads a Q&A session with Wilkins.

Wilkins emphasized the importance of community engagement and incorporating the perspectives of the population being studied. "Researchers usually set out presuming they have to educate the community they are studying, when it is actually the researchers themselves that may lack a meaningful understanding of what makes the community tick," she said.

Wilkins also discussed the importance of systems-level change in creating and integrating inclusive policies and initiatives to achieve health equity. She described how most health disparities researchers are trained to work at the individual level, studying individual health behaviors and responses to interventions. But the future of health disparities research requires investigators to explore upstream factors. She called on researchers to collaborate across fields and scientific departments to examine the higher-level social, cultural and environmental factors that disparately influence the health of racial and ethnic minority populations.

The lecture was NIMHD's Black History Month seminar.



Dr. Consuelo Wilkins

## Rare Disease

CONTINUED FROM PAGE 1

### Treatments on the Horizon

Most rare diseases emerge from a single gene variant, “so gene therapy is potentially a powerful approach for treating a wide range of rare diseases, including those viewed as too rare for commercial development,” said Dr. Lawrence Tabak, performing the duties of NIH director.

He outlined several initiatives—the Accelerating Medicines Partnership-Bespoke Gene Therapy Consortium, the Somatic Cell Genome Editing program and the Platform Vector Gene Therapy project—all underway at NIH to help speed development of new gene therapies. NCATS plays a leadership role in each of these initiatives.

Rutter noted that NCATS is developing platform-based technologies to enable more treatments to reach patients faster. And more help is on the way.

The Food and Drug Administration (FDA) is gearing up for a pilot program to authorize therapeutics that show promise. “This idea will allow faster clinical development with more real-time FDA-sponsored communications, which can ultimately move products as fast as possible through the regulatory pipeline,” said Rutter.

### Improving the AYA Experience

Several panels were devoted to the challenges and needs of adolescents and young adults (AYAs)—those 15-39 years old—living with rare diseases. One panel focused on the needs of AYAs with cancer.

“This age group had not seen improvement in survival of older and younger cancer patients in almost 30 years. Luckily, that is changing,” said Hilary Gan, director of hospital programs and services at Teen Cancer America, who spoke virtually from Los Angeles.

But difficulties remain and likely resonate among AYAs with any rare disease. AYAs don’t fit into the current health system, which is set up for children and adults. AYAs fall somewhere in between.

“They need age-specific, appropriate, targeted support even after they leave the hospital to address their unique challenges,” said Alison Silberman, CEO of Stupid Cancer, a national non-profit that offers resources



Aisha Dubose Campbell (l), director of resident services and family programming at the Children’s Inn, describes resources for young patients who stay at the inn, many of whom are adolescents and young adults. At right, event participants gather to chat near the All of Us Research Program table.

and community-building programs.

For AYAs who have gone through treatment, long-term side effects affect their physical, emotional and social well-being, noted Gan. AYAs need financial counseling and assistance; educational, occupational and psychosocial support; fertility counseling; and more targeted clinical research.

For adolescents with a rare disease, instead of thinking about fun teen stuff—friends, romance, graduation—they’re thinking about treatment and survival. They’re also thinking about mortality.

It’s one of the hardest conversations but AYA cancer patients should discuss their end-of-life preferences and wishes with family members and health care providers. Often, though, AYAs and their guardians don’t know how or when to broach the topic.

“We know adolescents have preferences for what they want and don’t want,” said Dr. Lori Wiener, director of the Psychosocial Support and Research Program in NCI’s Pediatric Oncology Branch.

After conducting a series of focus groups and a study that included participants at NCI and Georgetown, a resource was created for AYAs to facilitate communication about preferences for how they would like to be supported and comforted if they become seriously or terminally ill. That guide, *Voicing My CHOICES*, also covers issues of identity, spirituality, autonomy and commemoration. It was published in multiple

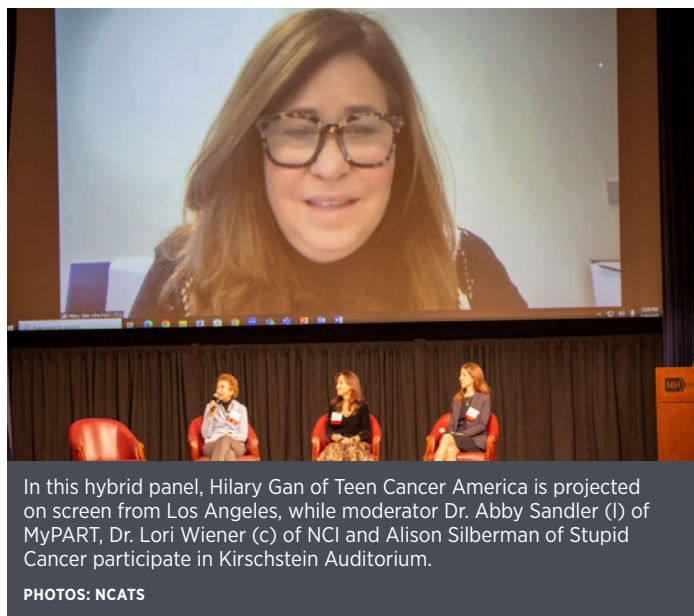
languages and has been requested in 42 countries.

“Perhaps the most important finding from a follow-up study at seven U.S. cancer centers was that anxiety around end-of-life planning decreased significantly immediately after [AYAs and their families] reviewed the document and a month later,” Wiener said. Follow-up has confirmed AYAs want to discuss end-of-life preferences and reap benefit from those conversations.

### Transitioning to Adult Care

Many rare diseases start in childhood and require specialty care throughout the person’s life. Two speakers shared their thoughts from a provider and patient perspective.

There are few adult-focused centers for rare diseases, particularly those that start in childhood, said Dr. Cary Harding, a pediatric specialist at a hospital in Oregon.



In this hybrid panel, Hilary Gan of Teen Cancer America is projected on screen from Los Angeles, while moderator Dr. Abby Sandler (l) of MyPART, Dr. Lori Wiener (c) of NCI and Alison Silberman of Stupid Cancer participate in Kirschstein Auditorium.

PHOTOS: NCATS

“I as a pediatrician, even though I’m trained as a geneticist and that handles the lifespan, don’t understand a lot of adult-onset disease,” he said. And since his main connections are in pediatrics, he struggles to find adult-specific resources for pediatric patients when they become adults.

Brittany Holmes is a nurse practitioner in a free-standing pediatric hospital—Boston Children’s—that does not have adult providers. Even if she had access to other local hospitals, that hospital might not be equipped with a rare disease specialist able to care for these patients.

Helping AYAs transition is a twisty journey of tracking down referrals and support services with little guidance in



Wiener (l) of NCI’s Pediatric Oncology Branch participates in a Q&A. At right, Brittany Holmes shares her perspective as both a rare disease patient and nurse practitioner, as Dr. Cary Harding looks on.

what often becomes fragmented care at multiple centers, frustrating patient and provider alike.

Holmes is both a provider and a rare disease patient. She can see both sides.

“I remember being a little afraid [of going to an adult-focused center],” she

shared. “I was treated at the same place since I was born. I trusted the health care team I had; starting over sounded scary.”

A collaborative approach is needed, Holmes said, to improve AYA protocols and guidelines. Expand options and resources, and continue research to broaden treatment choices, she urged.

As the Clinical Center continues to improve resources and emergency response capabilities for patients of all ages, Gilman acknowledged the many players in the rare

disease landscape—scientists and clinicians; patients and their families; advocacy organizations and policymakers.

“Rare Disease Day is unique in that it brings all the components of this ecosystem into the same orbit,” he said. “It’s a reminder that none of us are in this work alone.”

‘NOT SMOOTH SAILING’

Patient with Rare Disease Navigates Becoming the Captain

BY DANA TALESNIK

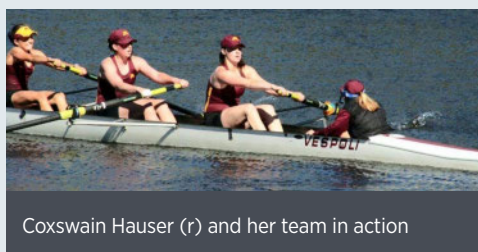
“My first memory of rare disease is one filled with fear and uncertainty,” said Abbey Hauser, a young adult rare disease patient and advocate, who shared her story during NIH’s Rare Disease Day.



When Hauser was six years old, she dislocated her knee while playing with toys in bed. Her family was outside having a yard sale; at first, nobody heard her screams.

“Unfortunately, it would not be the last time I’d feel alone or experience fear or uncertainty [which I would for many years] in relation to my medical care and diagnosis of Classical Ehlers-Danlos Syndrome,” she said.

Hauser grew up in Minnesota—the land of 10,000 lakes—where she was often around and on the water. Despite her EDS—a group of hereditary connective tissue disorders—she was a coxswain



Coxswain Hauser (r) and her team in action

on the University of Minnesota rowing team. Hauser’s love of the water steered her toward the nautical theme for her lecture that day.

On her rare disease journey, she said, “I quickly learned from a young age that I was not the captain of this ship.”

For young patients with rare disease, the transition from pediatric to adult care can be turbulent. Hauser likened rare disease to a boat. Everyone involved in the patient’s care has different roles to keep the boat afloat and functioning. The kid owns the boat but is not in charge, she noted. At age 18, the patient becomes the captain, regardless of whether they want or feel ready for that role.

“My rare disease boat was inefficiently run most of my childhood,” Hauser said. There was no one specialist coordinating her care and the chain of command was confusing. “I stayed afloat but it was not smooth sailing.”

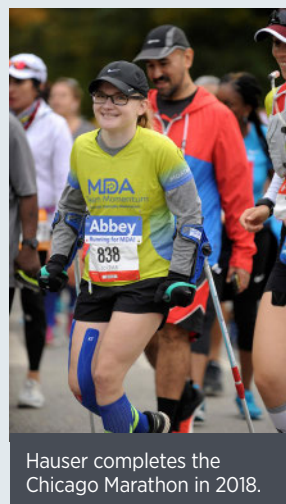
The week after Hauser turned 18, she had surgery on the knee that started her rare disease journey.

For all appointments leading up to the operation, she was a minor. Suddenly, she was the captain, signing documents and making decisions for herself in the adult wing of the hospital.

“I became the captain officially at one of the most vulnerable times we can have as a human—right before surgery,” she said.

During recovery, Hauser wanted to become a better captain. She used her new legal powers to request all of her childhood medical records from the various institutions involved in her care.

She had seen many specialists over the years. “My transition had consisted of waiting until I had symptoms that became intolerable enough to seek out care,” she said. She would then randomly pick a specialist from a web search and start the process all over again, repeatedly explaining her condition, sometimes receiving blank stares when calling it by name.



Hauser completes the Chicago Marathon in 2018.

Hauser offered several suggestions to help spare other young people from such a rocky transition. “We need coordinated clinics helping pediatric patients find adult care or support,” she said. From the time of diagnosis, educate patients early and often on the condition. And, she advised, parents should follow the child’s lead during the transition.

“We can help the kids of the future become strong leaders in their care.”

## Bohnet

CONTINUED FROM PAGE 1

people interpret the world, she continued. While these biases can help speed along decision-making during a busy day, they contribute to race, gender and socioeconomic inequality.

Bohnet noted many real-world examples of how gender biases create an uneven playing field. Recently, her colleagues at the University of Pennsylvania surveyed recruiters who attended a STEM job fair on campus about what they look for in candidates. The recruiters reported grade point average (GPA), internship experience and diversity were the most important qualities. Her colleagues then matched recruiters with candidates based on their preferences.

When it came time to hire, the employers did not hire students based on their own self-reported criteria. Researchers found that White male candidates with 3.75 GPAs were treated the same as women and minority candidates with 4.0 GPAs. Male applicants also got more credit for internships.

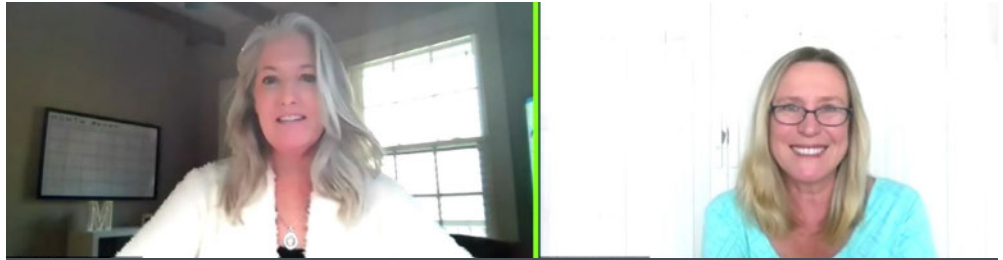
“That’s how these unconscious biases turn into real actions,” Bohnet noted.

Introducing hiring screens is one way to reduce gender bias in hiring. For example, the Boston Symphony Orchestra began “blind auditions,” where musicians perform behind a curtain. This mechanism increased the percentage of women who are part of the orchestra. Other organizations, such as the United Kingdom’s government, blind themselves to the names and addresses of job candidates.

In 2018, the Nobel Foundation asked Bohnet to review its nomination process. The foundation was aware that more than 90% of Nobel laureates were men from the Western hemisphere. The organization wanted to increase the number of women laureates. At the time, prize committees asked qualified individuals to nominate only one expert.

“I suggested to the foundation to explicitly invite people like me to nominate more than one person,” she said. “Our research strongly suggests that if we make decisions in bundles or batches, both accuracy and diversity increase.”

This approach ensures that decision-makers give more thought to who, for example,



NCI’s Dr. Mary Kearney (l), who chairs the NIH Women Scientists Advisors (WSA), and Bohnet participate in a question and answer session. Formed in 1993, WSA raises awareness of issues facing women scientists and works to improve women’s representation in the NIH faculty at all levels.

gets nominated for an award or considered for a position. It also expands the pool of candidates who get judged in the first place.

There is enough evidence to inform how to reduce bias in the hiring process, she said. Research suggests the best predictor of future performance is neither interview nor resume, but rather previous work sample tests that mirror the position’s responsibilities.

She warned about groupthink during panel interviews. Members of the panel will not come up with independent assessments. One person’s opinion can influence other panelists. Instead, Bohnet recommended one-on-one interviews.

“We have more gender diversity at the entry level, but much less at the top,” she said.

Bias in the performance review process influences who gets promotions, training opportunities and salary increases. Many organizations require employees to rate their own performance. Bohnet’s research has found that women, on average, give themselves lower performance reviews than men. In particular, women of color gave themselves the lowest self-evaluations. These ratings reduced women’s performance scores even though managers reduced female employees’ self-assessments less than those of their male counterparts, on average.

Bohnet has also discovered that women and minorities are given less support to begin with. This is called performance-support bias. These employees are often passed up for promotion because they have “thin files.”

Bohnet worked with a law firm that had this problem. She noticed that firm partners pursued first-year associates who were the same gender and race. Over time, the favored associates were included in important deals and received more feedback. The firm has

since changed how it allocates work to first-year associates.


Many women do “office housework”—the non-promotable tasks that keep an organization running. An example is reserving conference room space. While these important chores can be completed quickly, “the problem is that these little tasks accumulate,” Bohnet said. “When time comes up for promotion, we don’t acknowledge that people have supported our institution this way.” At the Kennedy School, tasks are explicitly taken into account in faculty’s workloads.

In addition to being aware of—and having the tools to address—bias, organizations must also be attuned to motivation. Deep exposure to people who are different from ourselves can motivate us to care about others. For example, men who mentor women better understand the issues women face and begin to consider them.

She urged others to become a “norm entrepreneur,” meaning someone who is interested in shaping the norms an organization holds. People want to be a part of the group.

Twenty years ago, the Kennedy School’s walls featured 50 portraits of leaders—all men. The school unintentionally signaled to its female population “they were not made to be leaders.” That has since changed. Now, there are many portraits of women from countries all over the world.

Finally, Bohnet noted that “too often we focus on the absence of women or people of color.” While it is true there aren’t enough women in positions of leadership, it’s important to communicate that “the train has left the station,” she concluded. “Gender diversity is happening. If you are not part of that club, you are on the outside.”

The full lecture can be viewed at <https://bit.ly/40TPjSq>. 



## NIH Researchers Discover New Autoinflammatory Disease

Scientists have identified an autoinflammatory disease caused by mutations in the *LYN* gene, an important regulator of immune responses in health and disease. Named Lyn kinase-associated vasculopathy and liver fibrosis (LAVLI), the identification sheds light on how genes linked to certain illnesses can potentially be targets for treatment by repurposing existing drugs.

The research, published in *Nature Communications*, was led by Dr. Adriana de Jesus and Dr. Raphaela Goldbach-Mansky of the translational autoinflammatory diseases section in NIAID's Laboratory of Clinical Immunology and Microbiology.

LAVLI was first discovered in a pediatric patient through genetic testing, which detected a mutation in *LYN*, the gene that encodes the Lyn kinase protein. Two additional, unrelated pediatric patients were later discovered to have two more mutations in the same gene. All three patients developed diseases linked to the *LYN* genetic mutation shortly after birth.

Two patients developed liver fibrosis—excessive amounts of scar tissue caused by inflammation and repeated liver damage—in the first year of life. All three patients had perinatal onset of neutrophilic cutaneous small vessel vasculitis, which is an immune disorder characterized by inflammation from high numbers of neutrophils—white blood cells of the immune system—that can damage small blood vessels.

The study revealed Lyn kinase was always active and unable to shut down in the three patients with the *LYN* mutation, which increased neutrophil migration, altered inflammatory signals and activated scar and fibrosis-inducing liver cells.

Results of this study suggest that Lyn kinase may be a potential therapeutic target for drugs that treat forms of non-syndromic small vessel vasculitis and other types of inflammation-induced liver fibrosis.

## Researchers Study Enhanced Model of Down Syndrome

Results of a new study, published in *Biological Psychiatry*, may help researchers develop more precise treatments to improve learning and memory in people with Down syndrome.

Researchers compared a new genetic animal model of Down syndrome to the standard model and found the updated version to be more similar to the changes seen in humans. Scientists often use different strains of mice as animal models to study human diseases because most genes in humans have similar counterparts in mice.

NIH researchers found the new mouse model, known as Ts66Yah, had memory difficulties and

behavior traits, but the symptoms were not as severe as seen with the previous mouse model.

“A mouse model that more precisely captures the genetics of Down syndrome has important implications for human clinical trials that aim to improve cognition,” said NICHD Director Dr. Diana Bianchi, who is also a senior investigator in NHGRI's Center for Precision Health Research and senior author of the study.



New study results may help researchers develop more precise treatments to improve learning and memory in people with Down syndrome.

PHOTO: DENIS KUYAEV/SHUTTERSTOCK

About 6,000 newborns are diagnosed with Down syndrome each year in the U.S. In most cases, these babies have a third copy of chromosome 21. An additional chromosome 21 adds an extra copy of more than 200 protein-coding genes to that person's genome, which causes difficulties with learning, speech and motor skills.

A previous mouse model has been considered the standard for Down syndrome research, used in preclinical studies for nearly 30 years. Along with some successful cognitive treatments, such as a recent hormone-based cognitive therapy, other treatments found effective in the mouse model were not as effective in humans.

Importantly, the previous mouse model's genome contains 45 extra genes that are irrelevant to human Down syndrome, a byproduct of how the model was developed. Humans and mice have very similar genomes, but the chromosomes that make up those genomes do not precisely align across those two species.

To create an enhanced mouse model of Down syndrome, researchers at the University of Strasbourg, France, removed these extra 45 genes using CRISPR gene-editing technology. Bianchi's group then compared the two mouse models and found that the extra 45 genes in the previous mouse model were affecting brain development and contributed to more severe difficulties with motor skills, communication and memory.

With this new and improved mouse model, Bianchi's group hopes to develop more precise treatments for improving cognition with the goal of independent living skills in people with Down syndrome.

## New Non-Invasive Imaging Tool Maps Labor Contractions

NIH-funded researchers have developed a new imaging tool, called electromyometrial imaging (EMMI), to create real-time, three-dimensional images and maps of uterine contractions during labor. The non-invasive imaging technique generates new types of images and metrics that can help quantify contraction patterns, providing foundational knowledge to improve labor management, particularly for preterm birth.

The tool lays the foundation toward potentially predicting who is at risk to deliver prematurely or whose labor pattern might result in the need for a cesarean section delivery.

The study team initially developed EMMI using a sheep model. In the new study, the team tailored EMMI for human clinical use and tested it among a group of 10 women with healthy pregnancies. Current clinical methods to measure contractions (i.e., tocodynamometry and an intra-uterine pressure catheter) can provide only limited details, such as contraction duration and intensity, while also being invasive.

EMMI integrated two types of non-invasive scans—a fast anatomical MRI to obtain an image of the uterus and a multi-channel surface scanning electromyogram that uses sensors placed along the belly to measure contractions during labor.

These data are then combined and processed into three-dimensional uterine maps, with warm colors denoting areas of the uterus that are activated earlier in a contraction, cool colors indicating areas that are activated later and gray areas showing inactive regions. A sequence of maps is generated over time, creating a visual time lapse that shows where contractions start, how they spread and/or synchronize, and potential patterns that are associated with a typical pregnancy versus one with complications.

Results from the pilot study also bring clarity to a longstanding question on how contractions begin—EMMI data suggest there is no fixed, pacemaker-like region in the uterus that initiates labor.

EMMI offers new possibilities for better understanding human labor and facilitating development of optimized, patient-specific interventions. The authors note that an EMMI contraction atlas generated from healthy pregnancies can serve as a resource to understand and diagnose preterm labor and possibly identify patients who would benefit from an induction versus those who may need a cesarean section.

The small study is supported in part by NICHD through its Human Placenta Project and other programs. Findings are published in *Nature Communications*.



Vickie Southers (l) with NIGMS EO Sally Lee

## NIGMS's Southers Retires

NIGMS Deputy Executive Officer Vickie Southers retired this past December after 22 years of federal service. Southers began her federal career at the U.S. General Accounting Office and then the National Institute of Standards and Technology. She joined NIH's Office of Human Resources in 2008, directing diverse programs to help NIH attract, engage and retain highly qualified employees.

In 2012, Southers joined NIGMS, serving as the principal advisor to Executive Officer Sally Lee. Southers shared in administrative management and provided leadership in strategic change, organizational design and effectiveness, and business process reengineering.

"Vickie will be deeply missed at NIGMS and NIH," said Lee. "She was a trusted and resourceful colleague and wonderful friend to many. She provided thoughtful mentorship and advice and was always willing to help others. She exemplified graciousness and teamwork."

Southers was committed to workforce and succession planning and the training and career development of employees. She provided expertise to NIGMS and NIH on important human resource issues, including helping to develop and implement a plan to identify areas of workforce underrepresentation, as well as potential barriers and retention issues.

Southers reinforced a culture of continuous improvement, cost effectiveness and enhanced business and management practices to meet the changing needs of NIGMS. Most recently, Southers led the effort to consolidate the institute's administrative functions into a single, centralized hub.

"NIGMS benefited greatly from Vickie's years of dedicated service," said Dr. Jon Lorsch, NIGMS director. "Her insights, guidance and good humor positively impacted many aspects of the institute. We wish her the best in her retirement."

## FAREWELL TO A FACE OF THE CC CC's Alexander Retires After 43 Years

After almost 43 years of dedicated service with NIH, Michael Alexander retired from his post as hospitality service coordinator for the Clinical Center's Office of Hospitality and Volunteer Services.

Alexander was a mainstay at the hospital's main lobby hospitality desk, greeting patients and their families. He is known for having an encyclopedic knowledge of CC locations, departments and resources.

In 2021, Dr. Francis Collins presented Alexander with an NIH Director's Award. He was recognized for his teamwork, professionalism and pleasant demeanor, which created a "welcoming atmosphere for patients and staff throughout the hospital."

"Michael was and always will be a part of the NIH family. He inspired people and brought positivity to patients every day," said Vivian Blair, chief of the Office of Hospitality and Volunteer Services. "We miss him already!"—**Donovan Kuehn**

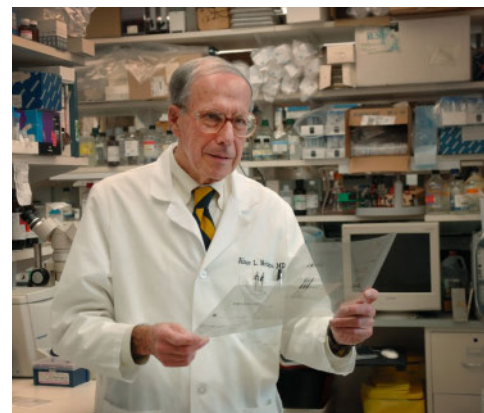


Michael Alexander



Alexander in the spot he occupied for 43 years

PHOTO: MARIA MASLENNIKOV



Dr. Abner Notkins

PHOTO: MICHAEL SPENCER

## SPENT SIX DECADES AT NIH Notkins, Architect of NIH IRP, Remembered

BY CHRISTOPHER WANJEK

Dr. Abner Notkins, a renowned expert on viral immunology and one of the architects of the modern NIH Intramural Research Program, died of complications from Covid-19 on Mar. 10 at age 90.

Notkins began his career at NIH in 1960, first as a research associate at the National Cancer Institute before moving to what was then called the National Institute of Dental Research (NIDR, now the National Institute of Dental and Craniofacial Research). He became a leader in understanding autoimmune diseases, particularly type 1 diabetes, which he revealed to be an autoimmune reaction associated with a viral infection, a fundamental discovery.

As chief of the NIDR experimental medicine section, Notkins continued to conduct novel research related to diabetes, insulin and other diseases associated with autoimmune antibodies throughout his 60-year NIH career. One standout achievement was his lab's isolation of a gene that encoded the protein islet antigen-2 (IA-2), now widely used to predict which children are at high risk for developing type 1 diabetes.

Other studies in his lab led to the discovery of immune interferon in the circulation of patients with diseases such as lupus, rheumatoid arthritis and scleroderma. He co-edited five books, held three patents and published nearly 500 peer-reviewed papers that employed molecular biology, genetics and immunology.

Notkins also was an extraordinary NIH citizen. He served as NIDR scientific director from 1985 to 1992. Upon his suggestion and subsequent organization, NIH held its first Research Day in 1986, an annual tradition that lives on today as the NIH Research Festival. He also has provided sage advice to numerous NIH leaders.

“Abner was truly a wonderfully wise and generous man,” said former NIH Director Dr. Francis Collins.

Notkins was born in New Haven, Conn., in 1932. His father was a physician, and his mother was one of the first female real estate brokers in the region. He graduated from Yale University and received his medical degree from New York University, before accepting a residency at Johns Hopkins University.

His many career awards include the David Rumbough Science Award from the Juvenile Diabetes Foundation and the Paul Ehrlich and Ludwig Darmstaedter Prize.

In a 2019 article from *Johns Hopkins Medicine* titled “Beyond the Dome: Abner Notkins,” he described his long career at NIH and his body of work as “more like a hobby in which I’m totally immersed.”

Notkins said, “NIH allowed me to explore my own ideas, which were mostly curiosity-driven. It also allowed me to spend most of my time in the lab, interacting with fellows and colleagues who were interesting and intellectually provocative. NIH was not always Camelot, but I can’t imagine a better place to do research. I have loved it.”

“Abner loved to chat about his latest research findings and then to question what you were focusing on in anticipation of identifying common ground for forward-thinking collaborative activities,” said Dr. Sharon Wahl, a former NIDCR branch chief and Notkin’s friend and collaborator. “He always had his eye on the goal of translational and ultimately, medically relevant insights. His six decades at NIDCR have certainly had a lasting impact, including the training of myriad fellows as part of his legacy, and his passing will leave a huge void.”

Notkins is survived by his wife, Susan.

Details about his life and career are captured in his 2017 NIH oral history, posted at <https://bit.ly/3GcOPAB>.

## NIAID’s Tramont Is Mourned

Dr. Edmund Tramont, associate director for special projects in NIAID’s Division of Clinical Research, died Mar. 5, after a brief illness. He was 83 years old.

Born and reared in Wallingford, Conn., Tramont developed a keen interest in microbiology as a student at Rutgers University. He graduated from Boston University School of Medicine in 1966. Tramont was drafted into the army in 1968, just two years after completing his medical degree, and began a residency at Walter Reed Army Medical Center. One of his first patients was Gen. Dwight D. Eisenhower.

Emerging as a leader in military infectious diseases, he established research programs at both Walter Reed Army Medical Center and Walter Reed Army Institute of Research. Through these organizations, he helped develop vaccines for myriad diseases that impact not just the armed forces, but also society in general. He also authored the Army’s first policies on managing HIV and AIDS, protecting both patients and force readiness.

After 23 years of service, Tramont retired from the Army in 1991 and was awarded the Army Distinguished Service Medal. He was recruited to join the staff at the University of Maryland, Baltimore, where he restructured the Medical Biotechnology Center and helped establish the Institute of Human Virology.

In 2001 Tramont became director of NIAID’s Division of AIDS (DAIDS). He helped lay the groundwork for the international focus of DAIDS clinical research activities, which are still flourishing today. He said the division should foster research in the populations that can most benefit. This approach bolstered the overall government approach to treatment and prevention of HIV through the PEPFAR program.

“I met Ed immediately after I arrived at NIAID in 2006,” said acting NIAID Director Dr. Hugh Auchincloss, in an email to staff. “He spent many hours teaching me the intricacies of conducting HIV research on a global scale. More recently...Ed has



Dr. Edmund Tramont

called several times, generously offering to provide advice and support at any time. I will personally miss his presence at NIAID.”


Dr. Richard Koup, acting director of the Vaccine Research Center (VRC), noted,

“When the VRC was in its early days, Dr. Tramont’s counsel helped to establish our early HIV vaccine development strategies... Throughout his career, he was a friend, advisor and colleague to many of us. Dr. Tramont’s passion, dedication, ingenuity and honesty made him an invaluable contributor to the research community, and he will be greatly missed.”

In more recent years, as an associate director for special projects, Tramont advised researchers throughout the world who were focusing on emerging diseases including SARS and Covid-19.

“Ed belongs in a hallowed category of dedicated souls who believed in doing good and striving always to heal the sick and prevent illness in the healthy,” said Abe Mittelman, senior advisor to the VRC director who served as the VRC’s first associate director for management and operations. “He was always sharply focused on achieving positive public health goals and on improving outcomes for mothers and babies exposed to HIV (especially in Africa), even when it wasn’t always at the top of other people’s priority lists. His work in the Department of Defense established baseline standards of care and, when he came to NIH, he worked to save lives at home and abroad...”

“I formed a trusting relationship with Ed,” Mittelman continued, “and he was always highly supportive of the need to ensure good management controls and good oversight of operational matters at the VRC. If, as I strongly believe, the VRC (embodied by its staff) has always had an uncommon zeal to stay focused and passionate about its work, Ed helped early on to inject his enthusiasm into that VRC ‘DNA’ and did his part to help keep it that way. The VRC has evolved much since the early days, but that zeal has never waned, and Ed’s character reflects that.”

Tramont’s survivors include a son, Dr. John Tramont, two daughters, Karen Altobello and Dr. Margaret Mulford, and seven grandchildren. 

## #PuppyCam Posts

“Happiness is a warm puppy,” Charles Schulz, creator of the well-known comic strip “Peanuts,” famously said. HHS leadership took the sentiment into account when they organized #PuppyCam, a livestream event intended to highlight various mental health initiatives across the department. The stars of the event came courtesy of Hero Dogs, Inc., a non-profit organization that provides trained service canines for U.S. military veterans and first responders with disabilities. The seven yellow labrador puppies, wearing miniature red service vests, romped around their enclosure and played with each of the speakers.

NIH was well-represented at the event: NCCIH’s Dr. Wendy Weber and Dr. Erin Quinlan discussed mindfulness, yoga, stress and pain management. Chaplain Michael Zoosman of the Clinical Center’s spiritual care department led a meditation exercise.

PHOTOS COURTESY TWITTER



At left, HHS Assistant Secretary for Health Admiral Rachel Levine (l), HHS Secretary Xavier Becerra and Assistant Secretary for Mental Health and Substance Abuse Dr. Miriam Delphin-Rittmon (r) pose with puppies. At right, NCCIH’s Dr. Wendy Weber (l) and Dr. Erin Quinlan cradle sleepy pooches. Below, the two NCCIH representatives share information about mindfulness, yoga, stress and pain management.



Pups take a “paws.” The seven woofers—eight weeks old at the event—are being raised to become service animals by Hero Dogs, a non-profit organization that serves military veterans and first responders with disabilities.



Above, Becerra and a companion extol the virtues of having a trusted friend (two-legged or four-legged) to help you through a mental health challenge. At right, CC Chaplain Michael Zoosman leads a calming meditation.