

## ‘PATIENTS ARE THE KEY’

### Families, Researchers Share Insights at Rare Disease Day

BY DANA TALESNIK AND AMBER SNYDER

For the 30 million Americans who suffer from a rare disease, finding answers is personal and urgent. Many people with rare diseases first endure a long and complicated journey to get an accurate diagnosis. If and when they receive one, that complex journey isn’t over. For most rare diseases, few if any treatments exist.

“We at NIH believe that caring for people with rare diseases is at the heart of everything we do, because NIH can be a focal point for this incredibly diverse and challenging community,” said NIH Director

Dr. Monica Bertagnolli during Rare Disease Day at NIH recently.

More than 2,200 people registered from around the world, many of them



The zebra is the official mascot for rare disease patients. Shown with NCATS Director Dr. Joni Rutter, this striped ambassador works for Global Genes, a rare disease advocacy and education organization.

PHOTO: NCATS

watching the day’s presenters via videocast. The annual event, held in the Natcher Conference Center, is sponsored by the National Center for Advancing Translational Sciences (NCATS) and the Clinical Center.

This year’s presentations highlighted new models for developing treatments; a new FDA-approved drug for Duchenne muscular dystrophy; and the promise of data science and personalized strategies to accelerate the arrival of new therapies.

An often-repeated theme is the integral role of people with the disorders. As NCATS Director Dr. Joni Rutter said in opening remarks, “Stories from patients and families are the thing that guide our scientific journey and spur the meaningful change that we need to see.”

### Patient and Caregiver Odysseys

Ashley Eakin’s journey has taught her the importance of visibility. She held off

SEE RARE DISEASE, PAGE 6

## ‘MILLIONS LIKE ME’

### Consultant Advocates for Better Accessibility

BY DANA TALESNIK

More than 60 million American adults live with a disability. Many struggle with inadequate accessibility tools and accommodations. Some challenges are common, others less obvious.



Dr. Matthew Weed

Dr. Matthew Weed counsels companies and agencies on ways to improve physical and virtual accessibility for people living with mobility, cognitive, vision, hearing and other impairments. Weed speaks from personal experience: he is totally blind and has type-1 brittle diabetes.

SEE WEED, PAGE 4

### Display in CC Commemorates Anniversary of Chinese Exclusion Act’s Repeal

BY ERIC BOCK

A new display at the NIH Library commemorates the 80th anniversary of the repeal of the Chinese Exclusion Act, which banned Chinese laborers from entering the United States.

“The repeal of the Chinese Exclusion Act was a really important event for science in the United States,” said Dr. Michael

SEE DISPLAY, PAGE 8



Dr. Nina Schor (c), flanked by Dr. Michael Gottesman (l) and Susan Wong, cuts the ribbon for the display at the NIH Library.



President Biden’s Executive Order boosts women’s health research. See story, p. 3.

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Former HHS Secretary Dr. Louis Sullivan, at a History of Medicine event in 2016

PHOTO: LISA HELFERT

### NIMHD To Hold Fireside Chat with Former HHS Secretary Sullivan

April is National Minority Health Month (NMHM), a time to raise awareness about the importance of improving the health of racial and ethnic minority communities and reducing health disparities. NMHM builds awareness about the disproportionate burden of premature death and illness in people from racial and ethnic minority groups and encourages action through health education, early detection and control of disease complications.

In observance of NMHM, the National Institute on Minority Health and Health Disparities (NIMHD) will host several events, including a Fireside Chat with Dr. Louis W. Sullivan, president emeritus of Morehouse School of Medicine and former secretary of the Department of Health and Human Services.

The virtual event is scheduled for Monday, Apr. 22 from 1:30 to 2:30 p.m. ET. Watch via NIH videocast.

For questions about NMHM events, email [Seppideh.Sami@nih.gov](mailto:Seppideh.Sami@nih.gov).

### Construction Continues on the CC's Newest Addition

Construction for the Clinical Center's surgery, radiology and laboratory medicine (SRLM) wing continues. In the photo at right, taken on Mar. 19, workers can be seen in the project's deep excavation pit. Multilevel Parking (MLP)-14 is shown in the back left corner.

Scheduled to open in 2029, the 547,290-square-foot addition will house three departments—perioperative medicine, radiology and imaging sciences, and laboratory medicine—and National Cancer Institute and National Heart, Lung and Blood Institute laboratories and patient care areas.

During construction over the next four or five years, Center Drive from the gate at Old Georgetown Road to the front of the Northwest Child Care Center, and Convent Drive north of the MLP-9 garage entrance will be closed.

For more information about parking and traffic changes associated with the SRLM and other construction on the main campus, visit <https://traffic.nih.gov/Pages/default.aspx>.

### NIMH Hosts Event for Autism Acceptance Month, Apr. 16

The National Institute of Mental Health and the Office of National Autism Coordination will host the 11th annual Autism Acceptance Month event, "Sound Tracks: An Artistic Journey to Belonging" on Tuesday, Apr. 16 from 2 to 4 p.m.

Autism spectrum disorder (ASD) is a neurological and developmental disorder that affects how people interact with others, communicate, learn and behave. People with ASD often have differences in social behavior and communication, intense interests and repetitive behaviors.

The hybrid event will feature autistic photographer Blair Bunting, autistic violinist Laura Nadine and neurodivergent filmmaker John Schaffer. The event will include a showing of the film *Sound Tracks*, directed by Schaffer and featuring Bunting and Nadine, as well as Q&A with the artists and remarks from Denise Resnik of First Place AZ, the nonprofit organization that sponsored the film.

Attend in person in Lipsett Amphitheater, Bldg. 10, or virtually via videocast at <https://videocast.nih.gov/watch=54247>. Pre-registration is recommended. Register online at <https://bit.ly/3PGodLh>.

Autism spectrum disorder can usually be reliably diagnosed by the age of two. If you notice signs, seek an assessment to get the earliest diagnosis, interventions and services possible.



The autism acceptance event features (clockwise, from top l) Blair Bunting, Laura Nadine, John Schaffer and Denise Resnik.

It's recommended that all children be screened for developmental delays beginning at their 9-month well-child visit and specifically for autism at their 18- and 24-month visits.

Learn more at <https://go.nih.gov/nLv8oCK>.



PHOTO: ERIC BOCK

## NIH Leaders Attend Signing of Executive Order on Women’s Health

NIH leaders attended the historic signing of an Executive Order (EO) to advance women’s health research and innovation. The directive, signed by President Joe Biden on Mar.

18, integrates and prioritizes women’s health across the federal research portfolio.

“For far too long, scientific and biomedical research excluded women and undervalued the study of women’s health,” said Biden at the White House signing. “The resulting research gaps mean that we know far too little about women’s health across women’s lifespans, and those gaps are even more prominent for women of color, older women and women with disabilities...It is time to pioneer the next generation of discoveries in women’s health.”

The EO’s directives will enable research that closes gaps to improve the health of all women. This guidance will bolster NIH-wide efforts to prevent, diagnose and treat health conditions unique to women, paving the way for better, more personalized care.

“I was honored to be a part of this

historic event,” said Dr. Kimryn Rathmell, director of the National Cancer Institute, in a social media post. “Supporting research on women’s health isn’t just about equality, it’s about understanding unique health challenges, advancing treatments and empowering women to live healthier, fuller lives.”

Rathmell was one of several NIH institute directors and leaders in the Office of the



Attending the historic signing at the White House are (from l) NICHD Director Dr. Diana Bianchi; NIAID Director Dr. Jeanne Marrazzo; NIAMS Director Dr. Lindsey Criswell; NCI Director Dr. Kimryn Rathmell; NIH Director Dr. Monica Bertagnoli; NIH Deputy Director for Program Coordination, Planning and Strategic Initiatives Dr. Tara Schwetz; ORWH Director Dr. Janine Clayton; and NHLBI Director Dr. Gary Gibbons.

Director to join NIH Director Dr. Monica Bertagnoli at the signing that was also attended by Vice President Kamala Harris, First Lady Dr. Jill Biden and Secretary of the Department of Health and Human Services Xavier Becerra.

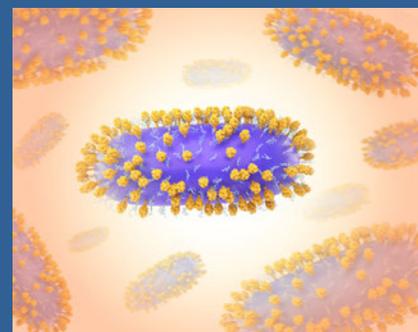
The full text of the EO can be found at <https://bit.ly/3xttFLh>.



President Joe Biden signs an Executive Order to advance women’s health research and innovation, as supporters of the directive applaud.



Vice President Kamala Harris (l) smiles with Bianchi (c) and Rathmell.



ON THE COVER: Creative artwork featuring 3D renderings of respiratory syncytial virus (RSV)—a common contagious virus that infects the human respiratory tract. The viral envelope is purple, G-glycoproteins are light blue, and F-glycoproteins are orange. F-glycoproteins allow the virus to fuse with and infect human cells.

IMAGE: NIAID

### The NIH Record

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## Weed

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“This talk is about the tens of millions of Americans with chronic health challenges, all of whom can benefit from greater understanding on the part of health professionals and scientists of the challenges we face in managing our health, whether in the clinic, the research laboratory or at home and work,” said Weed. He spoke at the recent inaugural talk in the NIH Diversity, Equity, Inclusion and Accessibility (DEIA) Lecture Series, sponsored by the Clinical Center, National Eye Institute (NEI) and the Office of Intramural Research (OIR).

Weed, who holds a doctorate in genetics from Yale and master’s degrees from Harvard and Princeton, began losing sight as a baby and became totally blind at age 8. In childhood, he suffered seizures and multiple comas caused by out-of-control blood sugar from diabetes.

“In the early 1970s, we had no at-home blood glucose monitoring and didn’t [yet] have recombinant insulin,” he recounted. Doctors told his family he wouldn’t survive to adulthood. Today, he’s still going strong at 53.

Weed began advising NIH in 1994 on improving access to facilities and databases. More recently, he has worked with institutes and centers across the agency to bolster website accessibility.

Greater accessibility helps all people with disabilities participate more fully in society. But disparities in the community, in particular, need greater attention. Women, people who are socio-economically disadvantaged and certain ethnic groups are more likely to report disability.

“These folks are the ones least likely to appear in their appropriate numbers in clinical trials,” said Weed, “so we may be mispending a lot of money on trials because we are not matching them to people who need them the most.”

There are other blatant gaps in research. Scientists with a long-term disability

earn far less than the median scientist, noted Weed. NIH data from 2022 also showed principal investigators with a self-reported disability on their application had a decreased grant success rate.

“Yet these are the people doing research on the issues we have to deal with and [that] NIH is chartered to reduce the risk and impact of going forward,” Weed said.

### Doctors, Take Note

“Millions like me have complex medical routines that health professionals and scientists must be aware of in prescribing medications in clinic or for use at home,” Weed said.

For example, many devices are not accessible. Some require tiny movements, such as

getting a small drop of blood onto a test strip.

“There’s no way for me to do that,”

Weed pointed out. And most glucose monitors don’t speak, so even if he somehow managed to get the droplet onto the strip, he’d struggle to get a reading.

“[Doctors] and patients need to work together to find creative solutions to these problems—solutions that can literally be the difference between life and death or at least [alleviate] great suffering for your patients and trial subjects,” he said.

Another challenge is differentiating among identically shaped medicine bottles. Blind patients need Braille

labels, which many pharmacies aren’t equipped to produce. Weed uses tape on preloaded insulin pens, but the caps can inadvertently get switched, resulting in medication or dosing errors.

“Any story I tell is likely repeated many times in many different ways across our nation,” he said.

Also keep in mind, he said, “The choices health professionals make and attitudes they carry can have huge impacts on patients’ outcomes and compliance.”

Be respectful, he pleaded. Don’t yell or patronize. Data may point to a better protocol but it might not be practical for that patient. He urges doctors to reduce barriers by seeing patients as people and keeping an open mind.

### Tech Challenges

It’s tough for a blind person to learn how to use a computer in the first place. Weed recounted a friend using cardboard cut-outs to explain the concepts of Windows and icons.

Navigating online presents its own challenges. Many websites and apps are not accessible for screen readers. And even screen readers are daunting. Weed played a clip of a common reader spitting out words so fast, the audio sounded garbled.

Almost all of the top one million high-trafficked web pages had at least one accessibility hurdle, noted Weed. And, he

said, nearly half of federal government websites failed the accessibility test on at least one of their three most popular pages.

He also shared examples of pitfalls at NIH. In one institute’s strategic plan, for instance, line numbers made the draft more digestible for the sighted but impeded screen readers from skimming it. There also wasn’t an accessible tool for Weed to insert his feedback.

Another NIH institute uploaded a report that was a “non-navigable block PDF.” The institute initially responded to

Weed’s concerns by citing budget and staffing constraints.

“If you create a major report and are having trouble financing full navigability,” Weed said, “please seek help from your agency in financing to get it to the point where it’s usable.”

Another tip: In out-of-office



A typical blood glucose monitor is tough to use for a blind person with diabetes

PHOTO: MIKHAILOV-STUDIO/SHUTTERSTOCK



Weed highlighted the challenge for blind patients, like himself, to differentiate among identically shaped medicine bottles and pens.



Weed delivers virtual lecture from his home.

autoresponder messages, include the email or phone number, not just the name, of your backup person. “A lack of access to information for someone who can’t see is also a lack of access, in this case, for any constituent of federal resources.”

### Training, Reality Differences

Weed argued there is a disconnect between the realities many patients face and the training health professionals and scientists receive to alleviate them.

“This is a cultural problem tied to training,” Weed said. He encouraged NIH and other federal agencies to become more engaged in how scientists and health professionals are trained so tools like screen readers and other assistive technologies are made known and available to those who need them. Such a change, he said, could greatly improve outcomes for millions of people.

### Getting Around

People with disabilities need physical spaces to be accessible both indoors and outside so they can get to work, access care and travel everywhere else they need to go. Weed’s talk specified compliance measurements for ramps and landings, parking lots, doors and auto-door openers, hallways, elevators and exam rooms.

He concludes, “The more we can find ways to reduce risk, grow our knowledge, improve accessibility of both [information] and places and improve empathy, the less people who already face barriers will have to climb health care and regular-life mountains while working to recover, maintain and hopefully improve their health and wellbeing.”

Learn more about Weed: <https://www.drmatthewweed.com/>. 

## Nobel Laureate Doudna To Speak, Apr. 15

Dr. Jennifer Doudna, 2020 co-recipient of the Nobel Prize in Chemistry, will deliver an NIH Director’s Lecture on Monday, Apr. 15 at 1 p.m. as part of the Wednesday Afternoon Lecture Series (WALS).

Titled the “Future of CRISPR: What’s Ahead for Genome Editing,” the lecture will be held in Masur Auditorium, Bldg. 10, and viewable online at <https://videocast.nih.gov/watch=54303>.

Doudna is a Howard Hughes Medical Institute investigator as well as the Li Ka Shing chancellor’s chair in biomedical and health sciences and a professor of molecular therapeutics at the University of California, Berkeley.

In 2012, she and her colleague Dr. Emmanuelle Charpentier, then at Umeå University in Sweden, proposed that CRISPR-Cas9, a set of enzymes that bacteria use to regulate immunity, could be employed for high-precision genome editing in higher-order species. The finding set off what has been called the CRISPR revolution and earned the two researchers the Nobel Prize and numerous other accolades.

Doudna has since become the face of genome editing. Indeed, “DNA” is in her name. Current research in her lab focuses on discovering and determining the mechanisms of novel CRISPR-Cas and associated proteins; developing genome-editing tools for use in vitro, in plants and in mammals; and advancing anti-CRISPR agents.

For her WALS Director’s talk, Doudna will discuss FDA approval of the first CRISPR therapy and ongoing research aimed at expanding access and reducing costs of CRISPR medicines. She will explain how CRISPR genome editing works and how CRISPR is used to treat patients with sickle cell disease. Then she will discuss ongoing research into the accuracy and delivery of CRISPR therapies that will make them more widely available in the future.

Learn more about this lecture on the WALS website at <https://oir.nih.gov/wals>.—**Diana Gomez**



Dr. Jennifer Doudna

PHOTO: CHRISTOPHER MICHEL

## Take Your Child to Work Day Is Apr. 25

NIH’s 30th Take Your Child to Work Day will take place on Thursday, Apr. 25 from 9 a.m. to 4 p.m. ET. Parents and their children can have in-person, hands-on experiences at this year’s event,



TYCTWD 2017

PHOTO: CHIA-CHI CHARLIE CHANG

which also will include Earth Day activities. Virtual and prerecorded activities will once again be offered for remote workers and employees at other NIH locations.

Volunteer opportunities will also be available for students in grades 9-12 to help with virtual activities.

For details, visit

<https://takeyourchildtowork.nih.gov/Pages/default.aspx>. Email questions to [Take-Your-Child-To-Work@nih.gov](mailto:Take-Your-Child-To-Work@nih.gov).

## Postbac Poster Day 2024 Set

NIH Postbaccalaureate Poster Day is a two-day event that will take place May 1-2 in Natcher Conference Center (Bldg. 45) on the main campus. Intramural postbacs are provided an opportunity to share with each other and the broader NIH community the research they have been conducting. At the same time, they develop scientific communication and networking skills.

Posters will be reviewed and judged by teams composed of graduate students, postdocs and staff scientists/clinicians. Authors of the top 20 percent will receive a letter acknowledging their accomplishment.

Investigators, staff scientists and scientific administrators can make a particularly important contribution by visiting posters and engaging authors in discussion.

For more information, visit <https://www.training.nih.gov/me/ppd/>.

## Rare Disease

CONTINUED FROM PAGE 1

pursuing her dreams of being a filmmaker for years because she thought the career was out of reach for a person like her.

Eakin has a rare condition called Maffucci syndrome, which causes genetic mutations that affect the growth of long bones and may also cause tumors. She has had more than 28 surgeries over her lifetime and has beaten ovarian cancer twice.

“I was ashamed of being different for most of my life,” she said. A speaker with a visible disability in a film business class in college changed her mindset.

Another lesson in inclusivity came later, when she worked as a director’s assistant for Jonathan Chu on the film *Crazy Rich Asians*. Eakin saw what the representation meant to the Asian community and realized that her shame about her own disability could stem from a lack of representation. She struck out on her own as a director in 2017 and has focused on projects that feature authentic characters who have disabilities.

“Go toward fear and face it,” she said. “It could change your life.”



Dr. Dominique Pichard of NCATS shares her family’s story. Above, a family photo with daughter Catalina, who has Rett syndrome

## Clinical Trials Crucial

Dr. Dominique Pichard has devoted her career to rare disease research and advocacy, inspired by personal experience. Her daughter Catalina lost verbal language as a baby. Pichard suspected Rett syndrome but was immediately dismissed by the pediatrician.

“I was not seen; I was not heard. But in my gut, I knew I was right,” said Pichard, director, Division of Rare Diseases Research Innovation (DRDRI) at NCATS.

As a clinician, she knew her way around the medical system. She got referrals and met with specialists. Yet it still took a year to confirm that diagnosis. “[DRDRI] works really hard so that, one day, families don’t have to go through this.”

Catalina is now a teenager. For most of her life, no treatments existed for Rett syndrome—until now. Last year, the FDA approved a treatment, the culmination of more than a decade of clinical research.

Pichard reiterated her gratitude to clinical trial participants. “Patients are the key to finding treatments for rare diseases.”

## Understanding, Empathizing Important

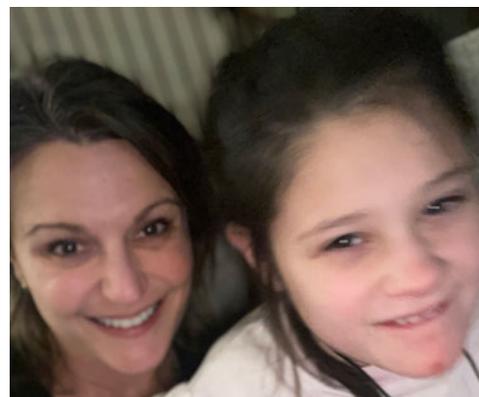
Jessica Swanson shared her family’s diagnostic journey, which took more than nine years. She knew of their family’s genetic predisposition to spinocerebellar ataxia, but tests revealed that wasn’t the cause of her daughter Isla’s spasms and relentless seizures.

Isla began displaying unusual movements at seven months old. When she was three, a medication tested during a clinical trial stopped her seizures, but the uncertainty continued without a diagnosis.

“The challenge of conveying years of medical history in mere minutes to new doctors, the weight of the four-inch medical binder, and the trust placed in new hands never got easier,” recounted Swanson.

In 2021, the family connected with NIH’s Undiagnosed Diseases Network (UDN). A year later, a diagnosis arrived: CDKL5 deficiency disorder.

The diagnosis “provided a label, a community and hope for the future,” Swanson said. “With exciting medical



Jessica Swanson describes her decade-long journey to find a diagnosis for daughter, Isla. She found her answer through NIH’s Undiagnosed Diseases Network.

treatments in the immediate pipeline, we would have missed the opportunity to improve Isla’s life because we didn’t have that diagnosis until the UDN.”

Swanson urged researchers, clinicians and advocates to take Isla’s story as a call to action.

“The need for a unified medical system and more collaborative approaches to patient care is evident,” she said. “Our experience underscores the importance of not just looking, but seeing; not just testing, but understanding; and not just treating, but empathizing.”

## Finding New Treatments

Of the more than 7,000 known rare diseases, only a few hundred have active research programs, leaving a significant gap in available treatments. Investigators like Dr. James Hickman want to change that by developing new platforms for rare diseases.

The co-founder and chief scientist for Hesperos, Inc., Hickman spoke about his company’s work creating custom, human tissue chip models to reproduce and develop treatments for a wide range of rare and ultra-rare diseases. Using cells derived from patients, he and his team create interconnected, multi-organ models that accurately reproduce specific disease states and a patient’s response to treatment. These models can predict patient outcomes more accurately than traditional animal models and are actively accelerating rare disease treatments today.

Hickman emphasized that these tissue chips “aren’t just laboratory things you might have in 10 years.” The various disease

models he showed in his presentation are all commercially available.

Hesperos was the first company to enable authorization of a clinical trial using only data from a tissue chip model (NCT04658472).

The drug was repurposed from another

indication to treat chronic inflammatory demyelinating polyneuropathy/CIDP, a rare autoimmune disorder.

Hickman also empathizes with rare disease patients on a personal level; he has struggled with an undiagnosed peripheral neuropathy for 15 years.

“These [tissue chip] systems create hope for all patients battling a rare disease,” he said. “As the technology continues to gain adoption, we expect therapeutic development to continue accelerating for this underserved population.”

Promising research continues through the NIH-funded Rare Disease Clinical Research Network, which connects scientists to rare disease organizations and advocacy groups, and through the public-private Accelerating Medicines Partnership Bespoke Gene Therapy Consortium. Developments are on the horizon.

### Expanding Access to Care

In a disadvantaged border town in Texas,

a collaborative project gives hope to children with rare diseases and serves as a model that could be replicated and expanded.

Dr. Seema Lalani, a professor at Baylor College of Medicine in Houston, discussed the NCATS-funded Project GIVE (Genetic Inclusion by Virtual Evaluation), which enrolled about 100 families in the Rio Grande Valley (RGV). The medically underserved population there is largely Hispanic, living below the poverty line, with limited access to specialized care. Houston, where Lalani works, is five hours away.

“Reducing health disparities and improving access to care in RGV is a priority for us because these are children who have endured years of health decline with no prospects of preventing late morbidities and chronic diseases,” Lalani said.

Launched in 2022, Project GIVE used [consultagene.org](https://consultagene.org), a virtual platform designed by Baylor, to enable doctors to refer local Latinx children and provide families a

tool for genetic evaluations.

Assisted by a bilingual social worker, the children gave biosamples at the study site, a clinic at the University of Texas-RGV. The clinic had a kiosk where families could subsequently go online for consultation and follow-ups with Baylor researchers.

So far, 61 families received genome sequencing results and 17 received diagnoses. Surveys revealed the families understood the genetic results and believed doctors would use those results to improve their child’s health.

For millions of people, the journey continues. Earlier, Swanson said, “The rare disease community taught us rare does not equate to alone; there is strength in shared experiences.”

“We are not done,” Bertagnolli pledged. “We have so much more to do. And we will continue to look for ways to work with everyone it takes as research partners to make life better.” **R**



At Rare Disease Day 2024 (from l) Ashley Eakin, NIH Director Dr. Monica Bertagnolli and Dr. Seema Lalani all give impassioned talks during the day-long event.



At left, event attendees—decked out in zebra stripes—attend an art exhibit as part of the event. At right, Dr. James Hickman **PHOTOS: NCATS**



Wong (l), chief scientific resources liaison with the Office of Research Services, came up with the idea for the display commemorating repeal of the Chinese Exclusion Act. FAN chair Dr. Jake Liang (c) says, “If it weren’t for the repeal, I most likely wouldn’t be here.” At right, Schor, NIH deputy director for intramural research, lauds the display’s statement.

PHOTOS: CHIA-CHI CHARLIE CHANG

## Display

CONTINUED FROM PAGE 1

Gottesman, senior investigator at the National Cancer Institute (NCI) and former NIH deputy director for intramural research, during the display’s opening ceremony. “Our scientific workforce depends on the incredible contributions of tens of thousands of Chinese scientists.”

Chinese workers first began to immigrate during the California Gold Rush, said Devon Valera, acting senior curator in the Office of NIH History and Stetten Museum (ONHM),



The display consists of two glass-enclosed panels located on either side of the NIH Library’s entrance.

who conducted research for the display. Anti-Chinese sentiment rose among other workers as the number of Chinese laborers increased.

In 1882, Congress passed the Chinese Exclusion Act—the first federal law that denied entry of a specific ethnic group. The legislation placed new requirements on people of Chinese descent already living here, ended the possibility of citizenship and made it difficult for them to return if they ever left. The act set a precedent for exclusionary immigration policies that targeted other Asian countries.

In 1943, Rep. Warren G. Magnuson—the legislator the Clinical Center is named after—proposed the Chinese Exclusion Repeal Act. It was signed into law by President Franklin D. Roosevelt.

Even though the act was repealed, Chinese immigration was still limited because of an ethnic quota system that favored European immigrants, said Valera. The system was in place until the Immigration Act of 1965 that removed race and ancestry as a basis for resettling here.

“The Chinese Exclusion Act is part of my family’s history,” said Susan Wong, chief scientific resources liaison with the Office of Research Services. “It resulted in the separation of my family in the early part of last century. The repeal allowed some of my family to reunite.”

Dr. Jake Liang was another beneficiary of the repeal of quota-based immigration laws. Soon after the policy reversals, his parents started talking about immigrating to the U.S. for a better life.

“My family ended up here in the mid-1970s,” said Liang, a distinguished investigator at the National Institute of Diabetes and Digestive and Kidney

Diseases and chair of NIH’s Federation of Asian American, Native Hawaiian and Pacific Islander Network (FAN). “If it weren’t for the repeal, I most likely wouldn’t be here.”

After the act’s repeal, several fellows and other research trainees joined NIH components, including NCI and what’s now the National Institute of Arthritis and Musculoskeletal and Skin Diseases, said Michele Lyons, retired ONHM senior curator, who was instrumental in gathering objects and information for the display.

One of those scientists, Dr. Jacqueline Whang-Peng, became world renowned for

her research on cytogenetics, or the study of chromosome inheritance. She was the first person to discover and confirm that cancer is related to chromosome variation. A display featuring Whang-Peng’s contributions to science can be found in the alcove near the pediatric clinic in Bldg. 10.

“I feel very proud to be part of a country that is not afraid to admit it made a colossal mistake,” said Dr. Nina Schor, NIH deputy director for intramural research. “There’s always someone, such as Congressman Magnuson, who has the courage to stand up and say, ‘we must reverse this. It’s not the right thing to do.’”

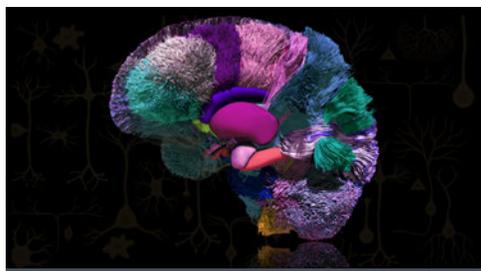
The display commemorating the anniversary of the repeal is located near the NIH Library’s entrance in Bldg. 10.



Candace Townsend (l), deputy director of the NIH Library, thanks NIH’s Federation of Asian American, Native Hawaiian and Pacific Islander Network and ONHM for curating and installing the display. Dr. Carl Hashimoto, director of faculty development in the Office of Intramural Research, offers remarks.

ONHM researched, wrote, designed and installed the display in-house, based on an idea by Wong. Contact ONHM at [history@nih.gov](mailto:history@nih.gov) for more information about the display, which is part of a rotating project that will be dismantled and replaced likely in late May, following AANHPI Heritage Month.

An agreement between ONHM and the NIH Library brings new stories and perspectives to the space about every six months. **B**



This image won second place in the photo category of the BRAIN image contest in 2022.

PHOTO: SAHAR AHMAD, YE WU, PEW-THIAN YAP

## BRAIN Initiative Photo & Video Contest Open Until Apr. 15

The Brain Research Through Advancing Innovative Neurotechnologies® (The BRAIN Initiative®) Photo and Video Contest is back.

All researchers, regardless of career stage, are invited to submit stunning, colorful and inspiring images captured by today's advances in neurotechnology by Monday, Apr. 15.

A committee will review anonymized submissions to narrow down the field to the top six photo and top six video entries they feel capture the creative spirit of the U.S. BRAIN Initiative.

Following the review, photo and video finalists will be displayed and open for public voting at <https://braininitiative.nih.gov/>. Winners will be announced at the 10th annual BRAIN Initiative Conference on June 16.

For details about eligibility and rules, visit <https://braininitiative.nih.gov/news-events/brain-initiative-photo-and-video-contest>.

## Stanford's Awaad To Give Spirituality & Health SIG Talk

NIH's Religion, Spirituality and Health Scientific Interest Group (RSH-SIG) will host a virtual presentation on Tuesday, Apr. 16 from 2 to 3 p.m. ET, featuring Dr. Rania Awaad of Stanford University. Her topic is "Enhancing Competencies in Providing Whole-Person Care for Muslim Individuals." The event is free, open to the public and will be recorded. Registration is not necessary. More information on Awaad and her work may be found at [https://profiles.stanford.edu/Rania\\_Awaad](https://profiles.stanford.edu/Rania_Awaad). For details about RSH-SIG and to get the Zoom link, visit <https://oir.nih.gov/sigs/religion-spirituality-health-scientific-interest-group>.

## Spring Vendor Expo Scheduled, Apr. 17-18

The NIH Recreation and Welfare Association and the Technical Vendor Association will hold a Spring Vendor Expo on Apr. 17-18 from 10 a.m. to 3 p.m. on the FAES Terrace in Bldg. 10. Visit about 40 different scientific vendors each day and learn about advances in technology that will help develop breakthroughs in medical research. Food trucks will also be in Lot 10H selling lunches both days. Visit <https://technicalvendor.com/2024-exhibitors/>, to see the list of participating vendors.

## 2024 AWARDEES TO PRESENT

### ODP Selects Three Speakers for Early-Stage Investigator Lectures

NIH's Office of Disease Prevention (ODP) announces its week of Early-Stage Investigator Lectures (ESIL) featuring the 2024 awardees—Drs. Andrea Schneider, Jessica Perkins and Benjamin Xu. Their lectures will take place via Zoom at 11 a.m. ET on May 6, 7 and 8, respectively.

ODP's ESIL recognizes early-career prevention scientists who are poised to become future leaders in prevention research.

For more information about this annual award, visit [prevention.nih.gov/ESIL](https://prevention.nih.gov/ESIL).



Dr. Andrea Schneider

The week will begin with a lecture by Schneider on Monday, May 6. Her presentation, "Powered by Numbers: Leveraging Epidemiology to Foster Prevention of Traumatic Brain Injury-Related Sequelae," will discuss the current epidemiology of traumatic brain injury (TBI) and efforts focused on improving the characterization of lifetime history of TBI in ongoing prospective cohort studies in diverse populations.

Read more lecture details at <https://go.nih.gov/DqHVHlf>.

Schneider is an assistant professor of neurology in the Division of Neurocritical Care at the University of Pennsylvania Perelman School of Medicine, with a secondary appointment in the department of biostatistics, epidemiology and informatics. Her work has been funded by the National Institute of Neurological Disorders and Stroke.

Perkins will give her lecture, "Misperceived Social Norms as Drivers of HIV Prevention: Exploring Opportunities for Individual and Social Change," on Tuesday, May 7. She will describe misperceived HIV-related norms and why these misperceptions matter for multiple levels of HIV prevention, as well as how to enhance the uptake of HIV-related health behavior interventions.

Find more information about the talk at <https://go.nih.gov/wPHlwpG>.

Perkins is an assistant professor at Vanderbilt University's Peabody College of Education and Human Development and serves as core faculty at the Vanderbilt Institute for Global Health. Her work has been funded by an NIH Career Development Award from the National Institute of Mental Health.



Dr. Jessica Perkins

On Wednesday, May 8, Xu will give the final ESIL, covering recent advances in early detection and prevention of primary angle closure glaucoma using optical coherence tomography (OCT) and artificial intelligence (AI). His presentation, "Early Detection and Prevention of Angle Closure Glaucoma Using OCT and Artificial Intelligence," will highlight how combining OCT imaging and AI can help overcome existing clinical limitations.

For details about his lecture, visit <https://go.nih.gov/4BQ4uOw>.

Xu is the chief of the Glaucoma Service at the University of Southern California Roski Eye Institute. His work has been funded by the National Eye Institute.

To register for each lecture, visit <https://go.nih.gov/3ISjcg6>. The lectures will be recorded and available on ODP's website approximately four weeks after each session.



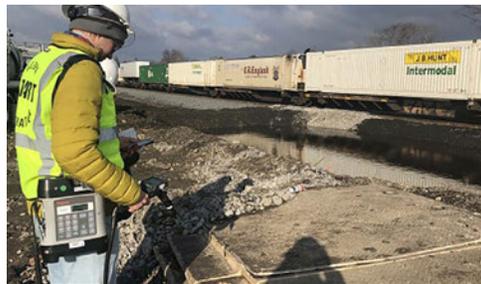
Dr. Benjamin Xu

# Six NIEHS Grants Announced to Study Health Effects of Ohio Train Derailment

BY ROBIN MACKAR

It was the evening of Feb. 3, 2023, when a Norfolk Southern Railway Company train carrying hazardous materials derailed in the village of East Palestine, Ohio. Some of the train cars caught fire and some spilled their loads onto the ground. These substances traveled into local waterways and flowed miles downstream.

For the past year, many local, state and federal agencies, including the National Institute of Environmental Health Sciences (NIEHS), have been part of a coordinated response to support the communities affected by the derailment in East Palestine and surrounding areas of Ohio and Pennsylvania.



Environmental Protection Agency contractors conduct air monitoring during rail excavation.

PHOTO: ENVIRONMENTAL PROTECTION AGENCY

From the early days of the disaster, NIEHS has been actively engaged to understand the community concerns about potential health impacts, and to identify opportunities for needed research. Much of this was done through NIH's Disaster Research Response (DR2) Program.

During a visit to East Palestine on Feb. 16, 2024, President Joe Biden acknowledged the "herculean efforts" made by the community and agencies since the tragedy. He announced the award of six NIH "grants to some of America's best research institutions to study the short- and long-term impacts of what happened here."

NIEHS made the awards through its Time-Sensitive Research Grants Program to conduct research and community engagement activities in East Palestine. Many of the grantees have already been working in the area and have established good working relationships with the community. The six recipients are:

- **Case Western Reserve University | Healthy Futures Research Study: Linking Somatic Mutation Rate with Baseline Exposure**  
Principal Investigator (PI) Dr. Fredrick Ray Schumacher and



Aerial view of derailment in the village of East Palestine, Ohio

PHOTO: NATIONAL TRANSPORTATION SAFETY BOARD

team will engage community partners and talk with East Palestine residents to better understand their experiences and concerns during and after the disaster. The team will also collect, and store for future research, blood and saliva samples and analyze them to understand how the mixtures of chemicals impact health both short- and long-term.

- **Texas A&M University | Responding to Air Pollution in Disasters Air Sampling and Symptom Monitoring**

PI Dr. Natalie Johnson and team will apply a mobile air-sampling strategy to rapidly characterize potential health risks from hazardous volatile organic compound exposure in the aftermath and recovery phases of the East Palestine environmental disaster.

- **University of California, San Diego | Uncovering the Short-Term Public Health Impact of Toxin Release: Outcomes and Effect Modifiers**

PI Dr. Beatrice Golomb and colleagues already have a community partnership in East Palestine and have conducted interviews with affected residents. The researchers will begin recruiting a cohort of residents to assess the short-term (approximately two to three years) health impact of being exposed to a mixture of toxins.

- **University of Kentucky | East Palestine Train Derailment Health Tracking Study**

PI Dr. Erin Haynes leads a research project that will include a health tracking study (via an online

survey) to collect longitudinal measures of health symptoms, stress and well-being of East Palestine residents. A researcher network will also be established to help report back and disseminate research findings successfully to the community.

- **University of Pittsburgh | Profiling the Post-Accident Exposome**

PI Dr. Peng Gao leads a research project that will collect soil, water and sediment samples to capture the extent of the contamination and to document the ongoing impact on the local environment of this region and its highly interconnected waterways.

- **University of Pittsburgh | East Palestine Community-Engaged Environmental Exposure, Health Data and Biospecimen Bank**

PI Dr. Juliane Beier will lead a research project that will use citizen science strategies to engage community members actively in collecting environmental samples, biospecimens and health outcome data to better understand the interaction of psychosocial stress and vinyl chloride exposure on their health and to find ways to reduce the immediate and long-term impacts of these exposures.

To learn more about time-sensitive research, visit <https://www.niehs.nih.gov/research/supported/timesensitive>.

To read details of the East Palestine grants, go to <https://go.nih.gov/LJa7SUw>.



Lead scientists on the NIEHS grants are (from l) Dr. Juliane Beier, Dr. Peng Gao, Dr. Beatrice Golomb, Dr. Erin Haynes, Dr. Natalie Johnson and Dr. Fredrick Ray Schumacher.

## Repurposed Drug Shows Promise Against Endometriosis-Related Pain



PHOTO: VORONAMAN/SHUTTERSTOCK

Fenopropfen, a nonsteroidal anti-inflammatory drug (NSAID), successfully alleviated pain and inflammation in a rodent model of endometriosis. In an NICHD-funded study, researchers chose the drug after using a computer algorithm to evaluate nearly 1,300 existing compounds for their ability to reverse gene expression related to endometriosis disease.

Analysis by researchers at the University of California, San Francisco, using publicly available data from people with endome-

triosis, returned 299 candidate compounds with seven considered top candidates. These drugs included commonly used treatments for the disease, such as aspirin, as well as those not yet studied for this purpose. Researchers chose fenopropfen for further evaluation because it returned the highest gene expression reversal score and belongs to a drug class—NSAIDs—that is one of the first-line treatments for endometriosis.

Fenopropfen is a prescription drug approved for the relief of mild to moderate pain and is often prescribed for arthritis. The researchers analyzed electronic medical records from five University of California health care institutions and found the drug had been prescribed for less than 1% of patients with endometriosis or related conditions. They then tested fenopropfen in a rodent model of the disease, observing that it successfully alleviated vaginal hyperalgesia, a surrogate marker for endometriosis-related pain.

If future studies in people confirm these findings, researchers suggest fenopropfen could be prescribed more frequently to treat endometriosis pain. The work also supports continued use of their computer-based approach to repurpose other existing drugs as potential therapeutic candidates for endometriosis.

Endometriosis occurs when tissue similar to the uterine lining grows outside of the uterus, often causing severe pain and infertility. The disease affects an estimated 10% of U.S. women, yet diagnosis is often delayed. Many existing treatments have challenging side effects, do not treat the source of the disease and leave a chance for recurrence.

## Severe Lung Infection from Covid-19 Can Damage the Heart

SARS-CoV-2, the virus that causes Covid-19, can damage the heart even without directly infecting the heart tissue. This NIH-funded study, published in the journal *Circulation*, specifically looked at damage to the hearts of people with SARS-CoV-2-associated acute respiratory distress syndrome (ARDS), a serious lung condition that can be fatal. But researchers said the findings could have relevance to organs beyond the heart and also to viruses other than SARS-CoV-2.

Prior imaging has shown that more than 50% of people who get Covid-19 experience some inflammation or damage to the heart. Scientists did not know, however, whether the damage occurs because the virus infects the heart tissue itself, or due to systemic inflammation triggered by the body's well-known immune response to the virus.

Researchers focused on immune cells known as cardiac macrophages, which normally perform a critical role in keeping the tissue healthy but can

turn inflammatory in response to injury such as heart attack or heart failure. Researchers analyzed heart tissue specimens from 21 patients who died from SARS-CoV-2-associated ARDS and compared them with specimens from 33 patients who died from non-Covid-19 causes. They also infected mice with SARS-CoV-2 to follow what happened to the macrophages after infection.

In both humans and mice, scientists found the SARS-CoV-2 infection increased the total number of cardiac macrophages and also caused them to shift from their normal routine and become inflammatory.

"This study shows that after a Covid infection, the immune system can inflict remote damage on other organs by triggering serious inflammation throughout the body—and this is in addition to damage the virus itself has directly inflicted on the lung tissue," said senior study author Dr. Matthias Nahrendorf of Harvard Medical School. "These findings can also be applied more generally, as our results suggest that any severe infection can send shockwaves through the whole body."



IMAGE: KATERYNA KON/SHUTTERSTOCK

## Antibiotic Shows No Benefit for Dry AMD

The drug minocycline, an antibiotic that also decreases inflammation, failed to slow vision loss or expansion of geographic atrophy in people with dry age-related macular degeneration (AMD), according to a phase II clinical study at NEI. The drug has shown beneficial effects for diabetic retinopathy, but had not previously been tested for dry AMD.



Retina, showing a large region of geographic atrophy

PHOTO: NEI

Dry AMD affects the macula, the part of the eye's retina that allows for clear central vision. In people with dry AMD, patches of light-sensing photoreceptors and their nearby support cells begin to die off, leaving regions known as geographic atrophy. Over time, these regions expand, causing people to increasingly lose their central vision.

Microglia, immune cells that help maintain tissue and clear up debris, are present at higher levels around damaged retinal regions in people with dry AMD. Scientists have suggested that inflammation—and particularly microglia—may be driving the expansion of geographic atrophy regions.

Led by Dr. Tiarnan Keenan, a Stadtman tenure-track investigator at NEI, the study tested whether inhibiting microglia with minocycline might help slow geographic atrophy expansion and its corresponding vision loss. The trial enrolled 37 participants at the Clinical Center and at Bristol Eye Hospital, United Kingdom.

After a nine-month period where the researchers tracked each participant's rate of geographic atrophy expansion, the participants took twice-daily doses of minocycline for two years. The researchers found no improvement in those who took minocycline.



## Music in the Atrium Features Hampered Dulcimer

PHOTOS: JANICE DURAN

The Dulcetones, an intergenerational musical quintet formed in 2022 in Arlington, Va., recently gave a lunchtime concert in the Clinical Center (CC). The group's sound includes the distinctive strains of a hammered dulcimer, a percussion-stringed instrument consisting of strings stretched over a trapezoidal resonant sound board.

Dulcetones performers include Paul Saunders on hammered dulcimer, David Roth on guitar, Phyllis Shea on violin and vocals, Mary Hynes on bass and Tim Felker on vocals. The group enjoys

creating fresh and unique takes on pop, rock, jazz, classical and folk classics, highlighting both their varied backgrounds and a shared love of musical collaboration.

Featuring more than a dozen songs, the Mar. 12 playlist at the CC ranged from Jim Croce's folk/rock classic *Time in a Bottle* and the bossa nova/jazz hit *Girl from Ipanema* to Irish folk ballads such as *Fields of Athenry* and *Danny Boy* and blues standard *The Thrill is Gone*.

The concert was part of the CC's Music in the Atrium program. To see the schedule of performances, visit <https://clinicalcenter.nih.gov/ocmr/music.html>.



The Dulcetones feature (from l) Dave Roth, guitar; Tim Felker, vocals; Phyllis Shea, violin/vocals; Mary Hynes, bass; and Paul Saunders, hammered dulcimer.

