- Molecular marvels
- A cure for Caroline
- Genetic counseling in the genomic era
- The skin microbiome
Four-year-old Caroline Fletcher snuggles with her mother, Kate. Jackson Laboratory Professor Robert Burgess and collaborators are working to develop a personalized gene therapy to treat Caroline’s rare neuromuscular disease.

Pictured is a Schlemm’s canal.

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These are qualities that define us — not just the scientists and staff of The Jackson Laboratory (JAX) but also our supporters. All of these qualities come together in this issue of The Search, especially in our feature story, “A cure for Caroline.” Finding a cure for a complex disease is often a bigger challenge than any one scientist, or any one institution, can solve. Thanks to a determined surgeon and JAX donor who sought out researchers working on the rare disease affecting his granddaughter, a partnership has been forged between JAX Professor Robert Burgess, Ph.D., and collaborators at other institutions. Together, they are making discoveries that offer new hope for patients like Caroline.

Ultimately, support for JAX is about impact: maximizing the potential of research that will transform medicine, and doing so with a sense of urgency, because real lives are depending on our discoveries. That requires collaboration and creativity. At JAX, we are determined to transform human health through genomic discovery and precision medicine — and to empower scientists and clinicians around the world in this quest. Assistant Professor Albert Cheng, Ph.D., profiled in this issue, embodies that commitment. Cheng’s research focuses on developing new, more precise and powerful technologies for gene editing: techniques that are already being used by JAX colleagues and that ultimately will give scientists around the world powerful, precision tools to develop models for research and to treat a wide range of diseases.

Innovative approaches like those Cheng is spearheading often face barriers, not least of which is funding. That’s why philanthropic support for promising new areas of research, pilot studies and inventive technologies is so vital. Whether your support is focused on a particular disease area or on scientific innovation more broadly, it makes an impact by fueling discovery, empowering scientific collaboration, and — ultimately and most important of all — transforming the way we treat patients.

That’s a vision that inspires us all.

Edison Liu, M.D.
President and CEO, The Jackson Laboratory

President’s message

The Search

President and CEO, The Jackson Laboratory

The Jackson Laboratory

President and Chief Executive Officer
Edison Liu, M.D.

Senior Director of Strategic Communications
Stephanie Wasco

Mission
We discover precise genomic solutions for disease and empower the global biomedical community in our shared quest to improve human health.

Locations
Bar Harbor, Maine
Farmington, Conn.
Sacramento, Calif.

Printed July 2016

DETERMINATION.
INNOVATION.
COLLABORATION.
INSPIRATION.

GROWING OUR WORLD-CLASS TEAM

CHA-LIN WEI, PH.D., APPOINTED JAX DIRECTOR OF GENOME TECHNOLOGIES

Cha-Lin Wei, Ph.D., an international leader in genomics and sequencing, has joined The Jackson Laboratory as director of genome technologies.

In her new role leading genome technologies across the Laboratory, Wei is based at The Jackson Laboratory for Genomic Medicine in Farmington, Conn., developing and providing genomics and sequencing services for JAX and external researchers.

Wei comes to JAX from the U.S. Department of Energy’s Joint Genome Institute in Walnut Creek, Calif., where she was head of production sequencing and led the genomic technology development effort. She also held the post of senior staff scientist in the biosciences division of Lawrence Berkeley National Laboratory.

S. CATHERINE (KATY) LONGLEY JOINS THE JACKSON LABORATORY AS CHIEF FINANCIAL OFFICER

Katy Longley of Brunswick, Maine, will join The Jackson Laboratory as vice president and chief financial officer.

Beginning her new role in August, Longley will provide strategic leadership, direction and management for all financial activities of The Jackson Laboratory, including leading the development of financial strategies, plans and budgets and managing a world-class financial services team.

For nearly 14 years, Longley has served as the senior financial officer responsible for the development to sterility. Accepting the Janeway Distinguished Chair position will enable Braun to expand his research program.

JAX RECEIVES $1.5M GIFT TO CREATE THE JANeway DISTINGUISHED CHAIR

The Jackson Laboratory has received a philanthropic gift of $1.5 million from Weslie Janeway, a longtime member of the institution’s Board of Trustees, to support the creation of the Janeway Distinguished Chair. Janeway’s gift ensures ongoing support for the research of the faculty member holding the chair.

Janeway’s past and current philanthropic support of the Laboratory includes the Pyewacket Scholars program, which provides funding for postdoctoral fellows working in the labs of early-career faculty members.

“With the establishment of an endowed chair,” says JAX President and CEO Edison Liu, M.D., “Weslie Janeway has underscored the critical role philanthropy plays in supporting our scientists at every stage of their careers: from creating opportunities for postdoctoral training, to providing funds for faculty who are just getting established as principal investigators, to supporting and recognizing the work of established and world-renowned researchers.”

Liu has appointed Professor Robert Braun, Ph.D., to the Janeway Distinguished Chair. Braun is one of the nation’s leading reproductive biologists, studying germine stem cells and how they develop into eggs and sperm. His lab explores how germine stem cells balance self-renewal with differentiation, and how imbalances between the two can lead to problems ranging from tumor development to sterility. Accepting the Janeway Distinguished Chair position will enable Braun to expand his research program.
Susan Mockus wants to overcome a language barrier. It’s not between English and Spanish — it’s the genetic vocabulary that oncologists and cancer researchers should share but too often don’t.

Mockus is The Jackson Laboratory’s manager of clinical genomics and curation. She and her team develop genomic tools to improve the accuracy of cancer diagnosis and treatment, and are also working to integrate more detailed genetic profiles of patients and their tumors into clinical trials of new chemotherapies.

Today many clinical trials of cancer treatments involve some patient screening to identify a few genetic mutations, but given the vast number of mutations and genes that could potentially be involved in a patient’s cancer, their oncologists aren’t getting the full picture. Moreover, there’s a terminology problem in classifying those mutations.

To solve this problem and ensure the cancer screening is as helpful to patients and oncologists as it should be, The Jackson Laboratory’s Cancer Treatment Profile next-generation sequencing service implements standardized terminology for mutations. This service holds promise not only to help oncologists find the best available treatments for their patients, but also to build a data resource on specific cancers that will guide precision cancer treatments in the future.

“If we are to truly enter the era of precision cancer care, we must gather as much information as possible about cancer mutations, identify them accurately and share what we learn with the world’s oncology community.”

Susan Mockus

Learn more about Susan Mockus at www.jax.org/spotlight.
...science fiction was a big thing on the big screen. The movies depicted things both large (“Jurassic Park”) and small (“Outbreak”) designed to thrill and scare audiences. Cheng’s response was somewhat unusual, however, and foreshadowed his future in research.

“In the movies I think that it’s very exciting to see people work together as a team to solve problems or create something beautiful,” he says, now an assistant professor at The Jackson Laboratory (JAX) in Connecticut. “If I were a science fiction character, I’d like to be Tony Stark — Iron Man — who doesn’t have a superpower but solves his problems through science and engineering.”

Cheng’s science and engineering marvels aren’t as showy as a flying metal suit, but they are remarkable nonetheless. He is a leader in the relatively new field of genome engineering and works to develop improved ways to edit genomes, manipulate function and activity within the genome, and engineer artificial proteins for binding DNA and RNA. In a nutshell, his work has the potential to make feasible entire areas of research that were previously impossible. The difficulties he faces are profound. Precisely and reliably tweaking something as immense, complicated and seemingly chaotic as a genome takes ingenuity and patience. But it’s this sort of challenge that appeals to him the most.
“What drives me now is figuring out how to make things work,” says Cheng. “If we spend a lot of time and effort to build something and it fails, I keep thinking about new ways we might find a solution. I get so wrapped up in it and excited about it sometimes that I have trouble getting to sleep.”

**Mentors’ legacy**

Encouraged by teachers from a young age to explore science and participate in projects and competitions, Cheng entered Hong Kong University of Science and Technology well prepared to jump right into bench research. And that’s what he was able to do.

“I would say I have been lucky to have very good science teachers all the way through my education,” says Cheng. “My secondary school STEM teachers were very inspiring, and they encouraged me to participate in science/engineering competitions. My university professors gave me the chance to just go into the lab and do research right away as a freshman, which definitely kept me interested in science. I’ve had that kind of encouragement and support along the way through getting my Ph.D. and then coming to JAX.”

Cheng moved from Hong Kong in 2007 to pursue his Ph.D. in computational and systems biology at the Massachusetts Institute of Technology (MIT) in the labs of Christopher Burge and Rudolf Jaenisch. Their influences are apparent in his current program. Burge studies gene expression and regulation, identifying genes in genomic sequences and investigating how messenger RNA is processed before being translated to a protein. Small changes in RNA processing can have profound effects on the proteins that are eventually produced. Jaenisch also studies gene regulation, but in different ways. His research has led to new understanding of stem cells, both embryonic and induced pluripotent stem cells (iPSCs), which can be created from mature cells. Jaenisch was an early innovator with the CRISPR genome editing technology as well, applying it in new ways to accelerate research in his lab.

**Opportunities knock**

As Cheng was finishing his doctorate, a friend of his in Jaenisch’s lab, Haoyi Wang, Ph.D., a postdoctoral associate, was recruited to join the JAX faculty in Bar Harbor. Cheng was already aware of JAX for mouse genetics and the Courses and Conferences program, and he was interested in continuing to work with Wang.

“We were like ‘How about we just work together at JAX’ so I joined Haoyi’s lab as a postdoc,” Cheng recalls. “After I came here, I was amazed by how strong the research program is and how much support JAX provides for researchers. We have centralized services with the latest equipment and technology, grant writing and application support, and administrative support. It’s very complete.”

Cheng was also one of the first to be named a JAX Scholar, a program established for exceptional postdoctoral associates. JAX Scholars are expected to work with greater independence than usual and to pursue independent funding under the mentorship of faculty. Once again, he embraced his opportunity and followed through with excellent work, becoming a productive and integral part of the Wang lab.

“Albert is one of the most inspiring people I have had the pleasure to meet and work with,” says Wang. “More importantly, he is such a smart, kind and fun person that we became really good friends through collaborating and just simply hanging out together. We want our friendship and scientific collaboration to be lifelong, and for our work to contribute to the improvement of human health.”

When JAX started building out its new facility in Connecticut for what is now The Jackson Laboratory for Genomic Medicine, Cheng was perfectly positioned for yet another leap forward.

“When JAX launched its genomic medicine campus, I was so excited,” he says. “I decided to apply to join the faculty there, and luckily I was accepted. I’m very happy to be starting my own research here as an independent principal investigator.”
The future

For Cheng, becoming independent didn’t mean becoming isolated, and he has continued to collaborate with Wang after setting up his laboratory at JAX. A project he set his sights on early was to take some of the key attributes of CRISPR/Cas — targeting very specific places in the genome and being able to bind to the DNA sequence there — and make the system more versatile. Initially, the only function was to cut the DNA at the targeted place and perhaps insert a different sequence (known as genome editing), but Cheng and Wang figured out how to make the CRISPR/Cas system perform many more tasks. And they made it so that different tasks can be carried out in the genome at the same time. The system, named Casilio, promises to be a very powerful tool for researchers and allow them to perform experiments that were previously impossible to do.

Cheng’s research and the tools he develops are highly technical and not specific to any particular disease, but he always keeps in mind the larger goals for them.

“I hope what we do will have an impact for patients, that it will improve human health,” he says. “One thing our tools can do is to help other scientists look at processes that are not possible with existing methods and allow them to take new approaches to help understand disease. We also hope we can further develop these tools to provide new therapeutic opportunities and improve diagnostic technologies.”

The quiet, painstaking work Cheng does is not the stuff of comic books. But, if one thinks about it for a minute, his impact on the human condition might be more profound than that of your average superhero.

“Developing the tools needed to understand — really understand — our own biology and how it works in health and disease provides a way to profoundly affect medicine and our ability to not only treat disease, but to avoid it whenever possible. And that’s just about the happiest ending possible.”

“Doing this work, step by step, sometimes you think it’s impossible to make it work,” says Cheng. “But at the end, when it actually does work, I think that’s the most rewarding part.”

CRISPR/Cas9 is a natural system that helps bacteria defend against viral attack. Adapting the system to use in the laboratory, researchers use the two components, a guide RNA and a nuclease (a protein that cuts DNA), to accurately target and edit specific DNA sequences. With CRISPR/Cas9, they can delete or correct mutations at the targeted site, like a simple word processor. It makes genome editing faster and more accurate than ever before, but the system is limited to one target sequence and one function at a time. Albert Cheng developed Casilio to expand the capabilities of CRISPR/Cas9.
For the last decade, Robert Burgess, Ph.D., a professor at The Jackson Laboratory (JAX), has studied Charcot-Marie-Tooth disease, a degenerative nerve disorder. He experiments with mice that model the disease’s genetic origins. Occasionally he will get an email or a call from someone who’s found his work online: Can he help a loved one who’s been diagnosed with the disease? “No,” Burgess typically replies with regret. “I can’t even help most of my mice,” he says.

Such was the case when Dr. Stephen Fletcher, a pediatric neurosurgeon from Houston, emailed him in the summer of 2013, asking if Burgess could help with his granddaughter, Caroline. As a toddler Caroline had been diagnosed with a form of Charcot-Marie-Tooth known as type 2D (CMT-2D), and her case was unusually severe.

Charcot-Marie-Tooth is a group of peripheral nerve disorders that cause muscle weakness and wasting in the feet, legs, hands and arms, and reduced sensation in the limbs. The disease is robbing Caroline of muscle strength, preventing her from standing or walking, restricting the use of her hands, and even making it difficult to breathe. “It was a compelling story,” Burgess recalls, “[I said] I’m really sorry to hear about this, but my lab works exclusively on mice, and I don’t have any clinical practice or facilities. We have the long-term goal of understanding this disease and developing a treatment, but that’s a long-term goal.”

Burgess suggested that Fletcher contact scientists at medical centers who might be involved in clinical trials of potential Charcot-Marie-Tooth therapies. He thought that would be the end of his involvement.

JAX Professor Robert Burgess and collaborators are developing personalized gene therapy for a Texas child suffering from a neuromuscular disease.

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A collaboration forms

A few months later, one of those clinical scientists, Scott Harper, Ph.D., a principal investigator in the Center for Gene Therapy at the Research Institute at Nationwide Children’s Hospital in Columbus, Ohio, received an email from Fletcher. As Burgess had done, he was ready to refer Fletcher to someone else.

“My initial impression was to simply inform him that I wasn’t working on CMT, was busy with several other projects, and didn’t think I would have time to start a new one from scratch,” says Harper, who researches muscular dystrophy. “However, I wasn’t familiar with the form of the disease his granddaughter had, CMT-2D, and I figured that before I responded to him, I should at least understand exactly what it was I was refusing to consider.”

Harper researched the disease online and came upon some intriguing scientific papers by Burgess describing CMT mouse models he had identified and characterized.

“After reading Rob’s work, I came to realize that many of the strategies we had been developing to treat dominant muscular dystrophy could be applied to CMT-2D,” Harper recalls. “He had the tools and expertise needed to make this project happen.”

Harper called Burgess, and the two quickly bonded. They discovered they were both from the Greater Tri-Cities region of Central Michigan and had studied at rival schools in the state, Harper at the University of Michigan as a graduate student and Burgess at Michigan State University as an undergraduate.

More importantly, their conversation left both men thinking they might be able to help Caroline after all. The pair realized that each scientist held half of a potential solution for her disease.

“I could have made the mouse, but I couldn’t have done anything with it,” Burgess says. “Scott could have developed a gene therapy vector, but would have never known if it worked in vivo [in a living organism]. Once Scott called me, it’s like, ‘This makes total sense. We should do this project.’”

And so they are. They have been working together for the last two years.

A third scientist Fletcher contacted, Anthony Antonellis, Ph.D., associate professor of Human Genetics and Neurology at the University of Michigan Medical School, is also collaborating with them. Antonellis has performed important cell-culture studies of Caroline’s disease and has worked with Burgess previously on mouse models.

Just as Fletcher’s determination to find hope for Caroline sparked this collaboration, his philanthropic support has fueled its growth. Fletcher’s gifts to support Burgess’ research (as well as the work of other scientists studying CMT) illustrate the power of philanthropy to accelerate scientific research and, in particular, to serve as a catalyst for innovative research projects that might otherwise not get off the ground.

A complex disease

Charcot-Marie-Tooth is named for the three physicians who first described it in 1886. There are no treatments to stop or slow its progression in the 2.8 million people who have the disease.

Even for the type 2D form of the disease, Caroline’s symptoms presented earlier and more severely than is typical.

“By the time she was 11 months I knew something was up,” says Fletcher. “I started noticing some weakness in muscle groups. We thought she was getting worse.

As time went on we noticed her hands weren’t working very well. I think she’s got progressive problems because she can’t gain weight due to atrophy of her muscles.”

More recently Caroline has had trouble breathing, requiring multiple stays in the hospital. “She lost a nerve that makes her diaphragm work,” Fletcher says. “That’s why she’s had so many pulmonary problems.”

Analysis of her family’s DNA revealed that Caroline alone carries a unique genetic mutation responsible for the disease. The mutation prevents her peripheral neurons — nerve cells connecting the brain and spinal cord to the rest of the body — from sending vital electrochemical signals to her muscles.

Caroline lacks part of a gene that instructs cells to make glycyl-tRNA synthetase, or GARS, an enzyme essential for protein production. A malformed GARS enzyme is believed to be toxic to axons, the long nerve cell fibers that transmit signals to motor and sensory cells in the limbs and other parts of the body.

“You can literally think of this as electricity,” Burgess says. “If you degrade the axon, your connection is gone. This is just like cutting the wire between the fuse box and the lamp.”

Caroline has one good copy of the GARS gene and one mutated copy.

“Unfortunately the mutation in the bad copy overrides the good gene,” Harper explains. “We think that reducing or eliminating the bad GARS gene will allow her good copy to function normally, and could offer a treatment for her disease.”

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Personalized gene therapy

In the coming months Burgess and Harper will attempt a gene-silencing strategy in mice that are being engineered to contain the exact genetic mutation underlying Caroline’s disease.

“This is the height of personalized medicine,” Harper says.

Burgess will inject the mice with Harper’s viral vectors, altered to deliver small, specific pieces of RNA, the molecule that translates DNA into instructions for making proteins. The vectors insert the microRNA into peripheral neurons and other cells, where it will match up with, and disable, faulty RNA.

The challenge with this “RNA interference” technique will be to silence the genetic instructions for making the faulty GARS enzyme without silencing the code for making normal GARS, because survival without GARS is unlikely, Burgess says.

If the mouse studies establish proof of principle that the technique works, the next step will be to determine safety through toxicity studies in mice and possibly larger animals, Harper says. Then investigators can apply to the Food and Drug Administration for a clinical treatment plan for Caroline, most likely to be carried out at Harper’s institution.

“The Center for Gene Therapy at Nationwide Children’s Hospital, led by Dr. Jerry Mendell, is arguably the world’s leader in translating gene therapy for neuromuscular disease,” Harper says.

Meanwhile, he and Burgess are proceeding cautiously.

“This is the first time that people have tried this,” Burgess says. “We’re doing our absolute best to do this right and get all the ducks in a row first.”

Harper says his own standard for proceeding to the clinic is, “Would I be comfortable delivering this therapy to my own children?”

Similar gene therapy approaches for other diseases are in clinical trials in the United States and Europe, and they should “help pave the way for translating the work Rob and I are doing,” Harper says.

“Afier decades of development, and after clearing some roadblocks, we are now finally starting to realize the great promise of gene therapy,” he says. “Importantly, we’ve learned a number of lessons from these early studies that are now finally reaching the clinic, and this gives me great confidence that we will be able to get there with our strategy as well, although I expect it will take time.”
Hope for Caroline

Caroline, now 4 years old, is undefeated by her illness and its daily limitations.

Though she can't walk, she zips around in a motorized wheelchair (“She calls it her magic chair,” Fletcher says.) She enjoys playing with her fraternal twin, Henry, who is unaffected by CMT, and other relatives and neighborhood friends.

“She doesn’t think she has a problem because she gets out and does stuff with them,” Fletcher says. “She can play. She’s very good and able to adapt to things. Nothing seems to impede her.”

As a grandfather, Fletcher is hopeful for a treatment, but as a physician, he knows how much time, money, work and technology are required to translate a theory into a therapy.

“Bench to bedside, that’s a difficult thing in medicine,” he says. “The clinical application of research is a totally different ballgame.”

He believes it may be too late to reverse the muscle loss Caroline has suffered but thinks gene therapy may be able to stop her disease from progressing.

“I’d like to see her live a long time,” he says. “She’s smart. She’ll have something to contribute.”

If relief for Caroline is possible, Fletcher believes the trio of scientists he has catalyzed on her behalf — Burgess, Harper and Antonellis — will be the ones to deliver it.

“I haven’t met them personally, but I love the hell out of them,” he says. “They are three researchers who work well together, but don’t have egos. That mouse model they’ve created, and what they’ve published in the last few years, I think they’re the go-to guys.”

Maximizing Impact, Funding Innovation

The Director’s Innovation Fund at JAX matched the philanthropic support from the Fletcher family to kick-start research into Caroline’s illness.

Tucker Taft and Phyllis Yale have been involved with The Jackson Laboratory for decades — but when asked what excites them most about JAX, they say it’s the future that inspires them.

Taft — a 1969 Summer Student Program alumnus — grew up hearing about JAX from family and neighbors with connections to the Laboratory, including his godfather Bob Little, son of JAX founders C.C. Little. “JAX is in his blood,” laughs Yale. And after 34 years of marriage, Yale is very much a part of the JAX family as well.

When the couple made a recent major gift commitment to JAX, they directed their support to the Director’s Innovation Fund (DIF). It’s a way to support the entirety of JAX’s mission while targeting work that can benefit the most from philanthropic investment.

“No one disease is most important to us,” explains Taft. The DIF was a natural fit with their interest in maximizing the impact of their gift by funding promising research at a stage when it’s so new that it would be difficult to obtain grant funding. The DIF, Taft observes, leverages the power of donor support and makes it possible for research with great potential to gain traction.

Yale concurs, adding that the escalating cost and accelerating pace of research make philanthropy more important than ever before in advancing JAX’s mission.

What draws them to JAX — and to the DIF in particular — is the multiplier effect of their support. “We look for institutions that have that [multiplier effect] and can also help the broader community.” Through the DIF, Taft and Yale are fueling scientific discovery both at JAX and, ultimately, on a global scale.

Taft sums up what makes JAX unique, noting that the scientists he’s talked to have come to JAX because of “the camaraderie, the resources, the people” — and a willingness to take scientific and creative risks. And thanks to supporters like Taft and Yale, that spirit of innovation has the fuel it needs to thrive.
The skill sets held by genetic counselors are a unique combination of intense science, critical thinking and empathic counseling.

– Joy Larsen Haidle, certified genetic counselor

Genetic Counseling in the GENOMIC ERA

The genomic era has given us an eye-popping 32,000 genetic tests for 5,800 medical conditions involving 3,900 genes, according to the Genetic Testing Registry, a database sponsored by the National Institutes of Health.

The number of tests has more than doubled in the last two years and continues to grow dramatically, driven by new technologies.

How then is a patient or even a physician supposed to understand this avalanche of new tests and make smart medical decisions about what the tests reveal?

Who to call

“Genetic counselors can be an ideal resource to help you navigate the evolving genetic testing landscape and protect your health,” says Joy Larsen Haidle, a certified genetic counselor at the Humphrey Cancer Center in Robbinsdale, Minn., and president of the National Society of Genetic Counselors (NSGC). “An informed consumer is also an empowered consumer.”

Counselors can be a critical part of a patient’s health-care team. They are medical professionals who hold specialized master’s degrees focused on genetics, health and related psycho-social issues. Their training is designed to guide patients through a potentially difficult time by discussing with them how their particular genetic makeup could impact them and their families, now and in the future.
Genetic counselors can offer two valuable things that many doctors can’t: deeper knowledge of genetics and genomics, and ample time to counsel patients. Most doctors receive little training in genetics or genomics in medical school, so they tend to welcome the expertise of genetic counselors.

Consultations typically last from 30 to 90 minutes, and follow-up sessions may be required.

Counseling sessions usually cost a few hundred dollars, while genetic tests can cost several hundred or a few thousand dollars, depending on the type.

Genetic counseling is widely, but not universally, covered by insurance, especially among patients who can document a high risk for a condition or disease. Many insurance companies consider genetic counseling and testing to be a cost-effective way to prevent and treat diseases.

Patients who get counseling and testing are protected from potential genetic discrimination by their employers or health insurance carriers by laws such as the Genetic Information Nondiscrimination Act of 2007, also known as GINA. (However, GINA does not apply to life, disability or long-term care insurance. Some states have enacted laws to fill these gaps.)

The Jackson Laboratory offers a clinical and continuing education program to help doctors and other health-care providers better integrate genomics-based medicine and genetic risk assessment into their practices. The program offers workshops and online courses for Continuing Medical Education credit. Learn more at www.jax.org/ccep.

What’s next

Genetic counselors keep pace with rapid technological change through continuing education, a requirement for maintaining their certification by the American Board of Genetic Counseling.

“I’ve seen so many changes,” says Schwartz, the UConn Health counselor who has practiced for 40 years. “Having the ability to counsel people based on specialized genetic test results has always been changing, and technology continues to emerge.”

Schwartz, an adviser to JAX’s CCEP, says incorporating genomics-based, precision medicine will be the profession’s next big challenge and opportunity.

“I think there’s tremendous promise,” she says. “We’re finding out things that are unique to an individual that helps doctors find better and more precise treatments. That’s an area that’s going to be expanding. We’re just now learning how to identify that uniqueness in individuals.”

Counselors will then collect the patient’s personal medical history, lifestyle habits and family medical history—still the “Holy Grail” of disease risk assessment after a half century.

“Family history is the core of precision medicine,” says Larsen Haidle. “It can be used to determine the chances of developing certain health problems, determine if surveillance should begin earlier than the general population or be done more frequently, and determine if genetic testing would be beneficial.”

Then follows a discussion of health risks that can be assessed; which genetic tests are most appropriate; which disease-prevention or management strategies might flow from the tests; and any emotional, financial and life-planning issues that might impact patients and their families.

“Genetic counselors are great partners in working with providers and patients to help select the best test and to discuss its benefits and limitations so the patient can make an informed decision,” says Kate Reed, a certified genetic counselor who directs JAX’s clinical and continuing education program CCEP for health-care providers. “This type of collaboration is necessary to ensure the right treatment to the right patient at the right time with everyone engaged in the decision making.”

The skill sets held by genetic counselors are a unique combination of intense science, critical thinking and empathic counseling,” says Larsen Haidle. “It is unique to understand complicated scientific concepts and be able to explain them in a meaningful and supportive way.”

Patients are often referred to genetic counselors after a physician assesses their family medical history or after a diagnostic test raises a red flag about a particular condition or disease. Patients can also engage counselors on their own. (The NSGC sponsors a free directory of counselors at www.findagenc counselor.com.)

What to expect

When you visit a genetic counselor, expect to be asked a lot of questions. Counselors are trained to follow the adage, “Seek first to understand, then to be understood.”

“One essential piece of the entire genetic counseling process is sitting down with the patients and discussing, ‘What brings you in today?’” says Robin Schwartz, a certified genetic counselor and assistant professor of genetics and genome sciences at UConn Health in Farmington, Conn.

“The Jackson Laboratory offers a clinical and continuing education program to help doctors and other health-care providers better integrate genomics-based medicine and genetic risk assessment into their practices. The program offers workshops and online courses for Continuing Medical Education credit. Learn more at www.jax.org/ccep.
The human body is covered in microbes of many varieties: bacteria, virus, fungi and Archaea. And if you had to pick a part of your skin most prone to environmental changes, you might pick the palms of your hands. Your hands are constantly touching new, microbe-covered surfaces — think doorknobs, handrails and food — and they are probably the part of your body you wash with soap most frequently. Eating with your hands and opening doors might introduce new members to your community, while washing your hands might decrease the number of microbes and open up space for new members to fill.

Yet, according to Assistant Professor Julia Oh, Ph.D., despite these near-constant assaults, the microbial community on your hands is remarkably stable. In a recent paper in Cell, describing work she did while at the National Institutes of Health, Oh and colleagues used metagenomic shotgun sequencing to monitor changes to the skin community at various body sites for a short (one to two months) and long time period (one to two years) on the same person. Even after two years, the community on an individual’s hands remained very stable. This suggests that environmental factors like constant touching and washing are relatively insignificant to the acquisition of new bacteria on the skin. Moreover, this suggests that our microbial partners are in it for the long haul — or at least for two years.

The skin community does change pretty remarkably during the transition from childhood to adulthood. The change is likely due to the increase in sebum (oil) on the skin during and after puberty. Oh and colleagues discovered this transition in prior research published in Genome Medicine in 2012. However, this new evidence implies that once the environment of the skin is stable, so too are the microbes that live on it.

As an interesting side point, other researchers have determined that the palm microbiomes of cohabitating couples are more similar to each other’s than to those of other healthy adults (eLife, 2013). Thus, the skin microbiome is not completely free of influence from the outside world, and the influence of cohabitation on the microbiome of mice is well documented. In the future, it would be interesting to monitor the pre- and post-effects of cohabitation with a limited life cycle — say, between college roommates — and whether the effect is long-lasting after the roommates have separated.
The Jackson Laboratory for Genomic Medicine and UConn Health co-hosted a day-long workshop for science journalists, who were able to tour both campuses and meet researchers. Here, JAX Professor, Florine Deschenes Roux Chair and Director of Genome Sciences Yijun Ruan, Ph.D., explains his work investigating the three-dimensional structure of the genome to journalists Cathy Shufro and Kelly Servick.

Photograph by Pete Morenus for UConn