



Children'sSM
Healthcare of Atlanta



Unprecedented Discoveries, Global Impact

Children's Healthcare of Atlanta
and Emory University School of
Medicine Department of Pediatrics
2021 Research Report



EMORY
UNIVERSITY

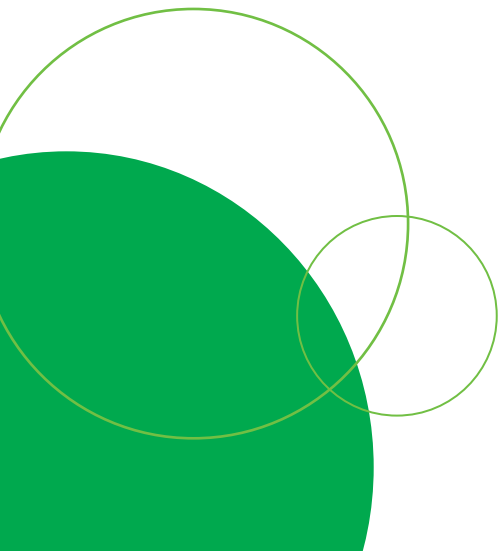




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A Message from Dr. Lucky Jain



Dear Friends,

4 For the past two years, our research and clinical teams at Children's Healthcare of Atlanta and the Emory University School of Medicine Department of Pediatrics have shown what it takes to combat a global pandemic.

In Atlanta and across the world, we pivoted from our day-to-day research to collaborate on multidisciplinary projects, working together to pursue innovative and nimble solutions to the evolving challenges presented by COVID-19. Indeed, our team's tireless efforts and personal sacrifices that began in 2020 have produced incredible advances and unforgettable impact in 2021.

In the past year, we participated in efforts that have led to: the development of serological tests and assays of neutralizing antibodies; authorization of pediatric vaccines with 250 children enrolled and 150 committed to begin the Pfizer/BioNTech and Moderna pediatric studies; the availability of at-home tests (after evaluating more than 70 different technologies in the National Institutes of Health (NIH) Rapid Acceleration of Diagnostics (RADx) program); authorization of new drugs for treatment such as the baricitinib-remdesivir combination; and identification of potential biomarkers. Overall, our researchers and physicians have published 145 COVID-19 studies since the pandemic began, with 29% appearing in high impact journals. Of these studies, 17 focused on MIS-C and two identified potential biomarkers of COVID-19.

We helped change the course of the virus and touched the lives of many in need. On top of it all, our regular research program continued to achieve significant milestones.

Representative of our many cutting-edge breakthroughs, the 2021-2022 U.S. News & World Report rankings are another testament to our success. Children's ranked among the nation's top pediatric hospitals with four specialties ranking in the top 10 and nine specialties ranking in the top 20. Our clinicians are empowered with innovative treatments discovered through basic, translational and clinical research.

While deeply involved in today's most vexing challenges, we always have an eye to the future. As part of our research expansion plan, it's important to note the unprecedented support we received this past year. A \$31 million gift from the Marcus Foundation is helping to establish a Center for Pediatric Cellular Therapies; a five-year public and private initiative from the Georgia Research Alliance will advance exploration into sickle cell disease; and a previous \$90 million transformational gift from the Robert W. Woodruff Foundation continues to facilitate 30 new positions for research-focused faculty members.

These donations are in addition to \$101.4 million in total extramural funding, including \$72.5 million in NIH funding awarded to our researchers in the last year.

This growth plan also enabled the design and construction of the new Health Science Research Building II (HSRB-II), expected to open in 2022. A significant portion of the building is dedicated to pediatric research. As such, HSRB-II will ensure we have the translational research environment required to enable research breakthroughs that will reduce the burden of disease for kids and their families. This is in addition to new clinical research space in the Children's Healthcare of Atlanta Arthur M. Blank Hospital scheduled to open in 2025.

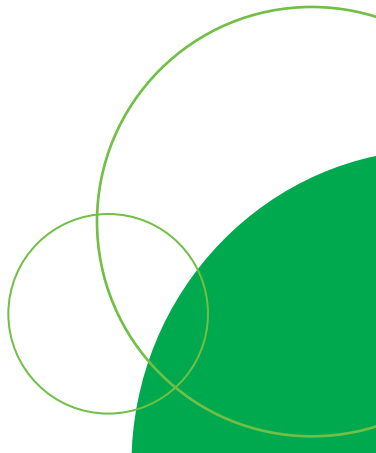
We have accomplished so much, and I'm honored to share with you this 2021 Research Report highlighting some of our incredible feats. I look forward to the innovation and life-changing work to come in 2022 and beyond.

All the best,



Lucky Jain, MD, MBA

Chief Academic Officer,
Children's Healthcare of Atlanta
George W. Brumley Jr. Professor and Chair,
Emory University School of Medicine Department of Pediatrics





Research Growth, Brick by Brick

Arthur M. Blank Hospital is Built for Breakthroughs

As Children's grows, so, too, does its research capabilities.

Arthur M. Blank Hospital, now under construction on Children's 78-acre North Druid Hills campus, is the largest healthcare project ever in the state of Georgia. Designated to be the only dedicated Level 1 pediatric trauma center in Georgia, the hospital is on schedule to open in 2025. In addition to overall patient care, the 1.5-million-square-foot facility will help accelerate research by creating a dedicated space for breakthroughs, including clinical research and trials, that will help transform the future of pediatric healthcare for generations to come.

To that end, the hospital will house a technologically advanced good manufacturing practice (GMP) facility to produce cell therapies for The Marcus Center for Pediatric Cellular Therapy which will be used in clinical trials for many pediatric diseases, including asthma, cancer, osteogenesis imperfecta, and others, with hopes of finding cures and treatments.

"Arthur M. Blank Hospital will provide the best care to the kids who need us most for many generations to come," says Donna Hyland, Chief Executive Officer of Children's. "We are proud of the pediatric future we are building for our city, state and country."



Arthur M. Blank and Donna Hyland

Emory University Builds Innovative Biomedical Research Facility

The new Health Sciences Research Building II (HSRB-II) at Emory University will be an eight-story facility with research space for imaging sciences, biomedical engineering, cardiovascular medicine, cancer, inflammation, immunity and immunotherapeutics, emerging infections and other innovative interdisciplinary research programs. The new facility is funded in part by the Robert W. Woodruff Foundation and Children's with approximately two floors of laboratory space reserved for pediatric research. HSRB-II is scheduled to open in 2022.

"This expansion of laboratory space will allow us to align our research across specific disease groups to optimize collaboration between investigators," says Doug Graham, MD, PhD, Chief of Aflac Cancer and Blood Disorders Center of Children's. "We will also be able to recruit top national researchers into this state-of-the-art environment, which will help us in our goal to be one of the premier research centers in the country. The research performed in this space will directly advance the care we can provide to children."





In 1956, the board of trustees for both Emory University and the former Henrietta Egleston Hospital for Children signed an agreement to make Egleston a first-rate teaching hospital.

Partners United

Children's and Emory University work together to facilitate leading-edge pediatric research, training and innovation. This long-standing partnership between one of the country's largest freestanding pediatric healthcare systems and a leading research university is mutually beneficial. Emory University's expertise and oversight are invaluable during clinical trials, and Children's provides a training site for the Emory University Pediatric Residency and Fellowship Program. Discoveries in Emory University's research laboratories are deployed in real lifesaving situations at Children's. Together, our partnership advances research and facilitates evidence-based quality care for children. **Watch the video at choa.org/researchreport.**

"We're innovating diagnostics and treatment of childhood and adolescent disease," says Wilbur Lam, MD, PhD, Pediatric Hematologist/Oncologist at Aflac Cancer and Blood Disorders Center of Children's and Principal Investigator for the National Institutes of Health (NIH) Rapid Acceleration of Diagnostics (RADx) program in Atlanta. "The culture here that is so cooperative and so willing to help each other is unlike any I have seen in the rest of the country."

2021 Research by the Numbers

Ranked
No. 4
nationally in NIH funding for
pediatric departments*


**\$72.5
million**
in funding
from NIH

**\$101.4
million**
in total extramural
funding



3,211
Children's patients
enrolled in clinical
research



3,715
visit hours
in the
Children's
Pediatric
Research
Unit



723
residents and fellows
in training



1,892
publications
in **885** journals



391
active clinical
trials



1,617
hours of CME
credits through
261
educational
activities

"What is so unique about Children's is that we have the clinical volumes to drive clinical trials," says James Fortenberry, MD, Chief Medical Officer of Children's. "As discoveries are made, we can quickly make them available to children."

*NIH funding period from
Oct. 1, 2020, through Sept. 30, 2021

Visit choa.org/researchreport to learn more.

Changing the Course of a Pandemic

From the moment COVID-19 turned the world upside down, Children’s researchers joined the fight to meet it head on.

Sixty principal investigators from Children’s and Emory have led projects to study SARS-CoV-2 with 145 publications. Their work in 2020 and 2021 helped change the course of the pandemic and allow for the gradual return to everyday life, by leading to authorization of pediatric vaccines, availability of at-home tests, authorization of new drugs for treatment, and identification of potential biomarkers. These discoveries have helped. Kids are back in the classroom and participating in social activities in person once again—and it was all made possible, in part, by discoveries made in our labs and clinics.

Watch COVID-19 research videos at choa.org/researchreport.



Making Pediatric COVID-19 Vaccines Available

Children’s and Emory participated in trials that have led to the authorization of COVID-19 vaccinations for more children.

Evan Anderson, MD, Pediatric Infectious Disease Physician at Children’s and Professor with the Emory Department of Pediatrics, has advocated for COVID-19 vaccine trials in children from the

beginning. He is serving as Principal Investigator of the Moderna and Pfizer-BioNTech trials for children at the Emory-Children’s Center (ECC), which has enrolled more than 250 children. Data from these studies has contributed to the FDA’s decision to grant emergency use authorization for vaccinations in children as young as age 5 (Pfizer-BioNTech) and young adults as young as age 18 (Moderna). Already, more than 2.84 million children have been vaccinated in the U.S.

According to Dr. Anderson, the work of his team has helped “make it possible for children to receive the same type of immune protection now provided to adults.” The Pfizer and Moderna trials continue to produce critical data, and Dr. Anderson and his team have also participated in Johnson & Johnson and Novavax studies.



Closer to a COVID-19 Treatment

Vaccines are proving to be effective at preventing COVID-19. But research at Children's and Emory is also making an impact on the lives of those who contract the virus.

Drug discovery expert and Professor of Pediatrics at Emory, Raymond F. Schinazi, PhD, DSc, has served as lead virologist for a major COVID-19 treatment discovery. Baricitinib—an anti-inflammatory drug and JAK inhibitor originally developed for treating rheumatoid arthritis—has now been tested in several clinical trials in adults with severe COVID-19. The results have been encouraging, as patients who received baricitinib were 46% less likely to die from the virus, and the drug has been authorized for solo use for hospitalized patients needing supplemental oxygen, ventilation or life support.

Clinical trials for its application in Atlanta were led by Vincent Marconi, MD, Professor of Medicine and Global Health at Emory. He and Dr. Schinazi have previous experience studying a similar drug for HIV patients, working with Christina Gavegnano, PhD, Assistant Professor in the Department of Pathology and Laboratory Medicine at Emory.

Assessing Antibodies and Variants

As new COVID-19 variants emerge, researchers at Children's and Emory's are hard at work determining if—and how—they will respond to the vaccines already developed.

A report published in the *New England Journal of Medicine* by Mehul Suthar, PhD, Assistant Professor of Pediatrics at Emory, indicates antibodies induced by mRNA vaccines are less effective in neutralizing the Delta and Kappa variants of COVID-19. Using lab assays with live SARS-CoV-2 virus, Dr. Suthar and his team found that vaccine-induced antibodies are 2.9 times less able to neutralize Delta and 6.8 times less able to neutralize Kappa. However, they reported the antibodies will be strong enough in many people and protective immunity is “most likely retained” against Delta and Kappa.

For the study, the research team obtained blood samples from 24 people who had recovered from COVID-19 in 2020, 15 people who had received the mRNA-1273 Moderna vaccine and from 10 people who had received the Pfizer–BioNTech vaccine.

Serendipitous COVID-19 and MIS-C Biomarker Discovery

One wouldn't think that research into children with traumatic brain injury (TBI) would be useful in studying COVID-19. But an unexpected discovery was made in the Children's and Emory pediatric neurotrauma lab, led by Andrew Reisner, MD, Medical Director of Neurotrauma and Pediatric Neurosurgeon for Children's.

The team discovered a molecular indicator, or biomarker, for COVID-19 and MIS-C severity while studying the same marker in children with TBI. The lab noticed similarities between the inflammatory responses of TBI and the virus and analyzed whether the plasma osteopontin biomarker could also identify those children with COVID-19. And, indeed, it could as levels of the biomarker were correlated with clinical data and were significantly elevated in children with moderate or severe COVID-19 and MIS-C compared to mild or asymptomatic children.

Findings were published in the peer-reviewed journal, *Experimental Biology and Medicine*, with collaboration from Children's Neuropsychologist Laura Blackwell, PhD, Stacy Heilman, PhD, Iqbal Sayeed with the Pediatric Neurotrauma Lab, and Infectious Disease Physicians Evan Anderson, MD, Andi Shane, MD, and Christina Rostad, MD. While larger follow-up trials are needed to determine the specificity and predictability of this marker for widespread clinical use, it is stable, easily accessible through a blood test, and can be quickly and affordably measured at the point of care. Plasma osteopontin therefore has the potential to scale up and inform the prognosis of anyone with COVID-19.



Andrew Reisner, MD and Laura Blackwell, PhD



Evaluating COVID-19 Diagnostics

Participating in the NIH's RADx program is one of the most significant COVID-19 contributions undertaken by our investigators to date.

The research was initially funded by a \$31 million grant, the largest singular NIH award in the history of research at Children's or Emory, and additional NIH grants, which catapulted the entire research program to No. 1 in NIH funding for pediatric departments in 2020.*

Led by Wilbur Lam, MD, PhD, the RADx program in Atlanta has contributed to the FDA, NIH and White House recommending and fast-tracking COVID-19 diagnostic tests, making them readily available in stores nationwide since soon after the pandemic began. As one of only five RADx sites in the nation and the only pediatric site, Atlanta is now also evaluating new variants of the virus, developing a pediatric self-administering protocol, and continuing to assess tests with a new focus on scaling at-home diagnostics to market. Dr. Lam and his RADx team encapsulate the unique opportunity available in Atlanta with a laboratory at Georgia Tech, tenured professorship at Emory and patients at Children's.



Wilbur Lam, MD, PhD

"The synergy between the three institutions allows me and my team to do what we do," says Dr. Lam, who sees patients at Children's while serving as a professor at Emory University and the Georgia Institute of Technology, where his lab is located.

2021 RADx Atlanta highlights:

- **January:** Children's appointment-only RADx collection site reaches 10,000 specimens.
- **February:** The first at-home, non-prescription, over-the-counter, self-administered COVID-19 diagnostic evaluated by RADx Atlanta is made available to the public nine months after the study launched.
- **May:** Dr. Lam is selected as the 2021 Health Care Heroes Award recipient in the Health Care Innovator/Researcher category from the *Atlanta Business Chronicle*. He commented: "We answered the challenge for the country, and today you can go to retail outlets and buy tests."
- **August:** At-home tests with a 90% accuracy rate for positive cases recommended to the FDA by RADx Atlanta are in high demand as the Delta variant spreads.
- **October:** The RADx Atlanta site is chosen to continue participating in diagnostic evaluations. The NIH invests \$70 million to boost the supply of at-home tests by accelerating test developers' progress through the regulatory authorization process.
- **November:** The RADx Atlanta team reaches 5,000 children enrolled into the research protocol, making it one of our largest pediatric clinical studies to date.

*According to rankings from the Blue Ridge Institute for Medical Research published Feb. 11, 2021.



Autism Research Exemplifies Pandemic-Friendly Study Method

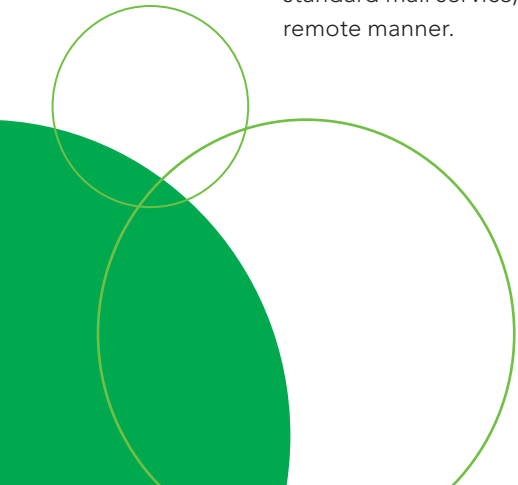
The earlier autism can be detected in children, the sooner they can begin treatment.

And autism researcher Gordon Ramsay, PhD, Director of the Spoken Communication Laboratory at Marcus Autism Center at Children's, is exploring novel ways to make early detection happen through the use of wearable sensors on babies.

Voice recorders track the sounds that babies make in their first three years of life to help his team determine if certain vocalizations may be an early indicator of autism. The recorder is light and portable, weighing only two ounces and tucking into the baby's clothing. Parents turn on the recorder when the baby wakes up in the morning, put it inside the pocket of the onesie, and leave it running all day.

To date, the team has tracked vocalizations from 450 babies, capturing 6,000 days and more than 60,000 hours of their early speech. Using this method, the team discovered boys produce significantly more protophones, the precursors to speech, including vowel-like sounds, squeals and growls, than girls, and this difference was even more pronounced in infants at high risk for autism. Thus, their findings may lead to a vocal biomarker for autism in the early stages of development, hopefully encouraging parents to seek treatment sooner.

Also, the study is an exemplary model of how to acquire data from a distance, which enabled the research to proceed during the pandemic. Dr. Ramsay and his team send out and receive the recorders using standard mail service, and all data was collected in a pandemic-friendly, remote manner.



Achieving Pediatric Research Milestones

Even with the demands presented by COVID-19, our researchers continued to reach new heights in their regular disciplines.

Significant milestones were achieved in cell therapy, HIV, cancer and blood disorders, neonatology, genetic research and more—much of it driven by NIH funding. Displaying diligence and expertise, our pediatric research program is one of the best in the country.



Edwin Horwitz, MD, PhD with students

New Center for Pediatric Cellular Therapy is the Way of the Future

While drugs have been the primary method of treatment for most diseases for many years, cell therapy may be the way of the future.

So says Edwin M. Horwitz, MD, PhD, Pediatric Hematologist/Oncologist at the Aflac Cancer and Blood Disorders Center

of Children's and cell therapy expert for 25 years. To help him on that journey of discovery, he has received a \$31 million grant from the Marcus Foundation for a new Center for Pediatric Cellular Therapy, which will provide the infrastructure needed for clinical trials to make this a reality.

Cell therapy, in contrast to drugs, works by using natural or modified cells to treat disease. Methods include regenerative medicine; rebuilding diseased or damaged tissues with cells; and cellular immunotherapy, which modulates the immune system by boosting, calming or redirecting it, such as CAR-T therapy for cancer.

As Co-Director of the Center for Pediatric Cellular Therapy, Dr. Horwitz will promote the use of cells in all disciplines throughout pediatrics at Children's and Emory. Already, seven clinical trials are in progress at the Center, studying such diseases as graft versus host disease (GVHD), asthma, neuroblastoma, dilated cardiomyopathy and osteogenesis imperfecta. The Center will include a good manufacturing practice (GMP) lab, known as the Marcus Cellular Production Facility, to be

housed at Arthur M. Blank Hospital for generating cells to treat patients; a Cell Therapy Trials Office for managing clinical trials; and a Cell Therapies Correlative Studies Laboratory to analyze patient samples.

“We are in the midst of an evolutionary change in the way we take care of patients,” says Dr. Horwitz. “Drugs were the mainstay of therapy for many years, but more and more, cells will be used to do things that drugs cannot do.”

Beating Heart Cells Return to Earth

An out-of-this-world experiment may lead to new ways to treat heart disease in children and adults.

Upon safely returning to earth and the Emory-Children’s Center, researchers determined that cardiac stem cells grew into numerous, mature, beating heart cells while aboard NASA’s SpaceX-20 mission in March 2020 after more than a year of analysis. Chunhui Xu, PhD, Associate Professor in the Department of Pediatrics at Emory, Kevin Maher, MD, Pediatric Cardiologist for Children’s, and their colleagues dived into the data from the space experiment, seeking to determine the effects of real zero gravity conditions on stem cells and to optimize the generation of clinically relevant cardiac muscle cells on earth.



Jessica Meir, PhD, NASA Astronaut,
Brody Parker and Kevin Maher, MD

Their research is critical in finding ways to apply cell therapy so that kids, such as Brody Parker, pictured above, will have enough robust heart cells for surgery, or children with damaged heart valves may one day receive a replacement valve grown from their own cells. Before having astronauts conduct the experiment on the International Space Station (ISS), the team used space-simulation machines to enhance the ability of pluripotent, or immature, stem cells to turn into cardiac muscle cells.

“These cells have the potential to treat heart disease in kids and adults, but repairing a damaged heart requires a large number of mature heart cells,” says Dr. Xu, Department of Pediatrics at Emory. “This space experiment may teach us to identify a more effective way to generate these cells.”



Ann Chahroudi, MD, PhD

Searching for a Pediatric HIV Cure

While the world is understandably focused on COVID-19, research continues on another deadly disease: human immunodeficiency virus (HIV).

Despite major advances in prevention and treatment, millions of children and adolescents continue to live with HIV each day. Ann Chahroudi, MD, PhD, Infectious Disease

Specialist at Children's and Associate Professor of Pediatrics at Emory, was awarded \$27.6 million from the NIH to accelerate the search for a cure for HIV in children and adolescents, as co-principal investigator with the Yerkes National Primate Research Center and Johns Hopkins University School of Medicine.

The grant is part of the Martin Delaney Collaboratories for HIV Cure Research program called the Pediatric Adolescent Virus Elimination (PAVE) Collaboratory, which uses a multidisciplinary, multicultural and iterative approach to study pediatric HIV.

"With this funding and a truly outstanding group of investigators and industry partners, we are thrilled to synergize the pediatric cure research efforts globally. The award will also allow us to apply state-of-the-art scientific tools to understand HIV persistence in children and adolescents with the ultimate goal of achieving HIV remission or eradication," says Dr. Chahroudi.

The PAVE Collaboratory aims to identify and harness the unique immunovirological features of HIV infection in children and adolescents. PAVE team members will conduct preclinical safety and effectiveness research studies of novel treatments, focusing on developing procedures, tools and techniques, including imaging, specifically for infants, children and adolescents.



Novel 'Atlanta Protocol' for Hemophilia A is Now Worldwide

Researchers at Aflac Cancer and Blood Disorders Center of Children's developed a new method for treating patients with severe hemophilia A in 2019. And that method—the "Atlanta Protocol"—is now being tested on a larger group of patients in an international, multisite, five-year trial.

Hemophilia A is an inherited bleeding disorder that causes abnormal blood clotting, resulting in more bleeding than normal due to low levels of factor VIII, a protein needed to form blood clots. About one-third of severe hemophilia A patients will develop even more bleeding due to inhibitors, antibodies generated by the patient's body against their treatment.

The Atlanta Protocol involved combining an old approach, immune tolerance induction (ITI), with the drug, emicizumab. The treatment was effective in four out of seven patients who were able to rid their bodies of the disease's stubborn antibody inhibitors. Given the successful results, Robert Sidonio Jr., MD, a Pediatric Hematologist/Oncologist at the Aflac Cancer and Blood Disorders Center of Children's and Associate Professor of Pediatrics at Emory, is further investigating the approach as co-principal investigator of the 120-patient international trial. The protocol was originally developed by Dr. Sidonio, and Pediatric Hematologists/Oncologists Glaivy Batsuli, MD, and Shannon Meeks, MD.



Robert Sidonio Jr., MD, Glaivy Batsuli, MD, and Shannon Meeks, MD

Aiming to Prevent Severe GVHD

The potentially life-saving results of bone and marrow transplant (BMT) procedures are sometimes compromised by a complication—one that the Aflac Cancer and Blood Disorders Center of Children’s is striving to overcome.

For hematologic cancer patients undergoing unrelated donor BMT, a condition known as acute graft-versus-host disease (GVHD) may occur after stem cell transplantation. This may result when the donated cells turn against the patient or host and begin attacking the patient’s organs. In the most severe forms, half of the transplant recipients die.

Providing new hope to patients, the drug abatacept was shown to reduce the risk of acute GVHD by 10 times among children and adults during a seven-year, multisite trial conducted by hematologists and oncologists at the Aflac Cancer and Blood Disorders Center. Benjamin K. Watkins, MD, and Muna Qayed, MD, led the trial at Children’s, published their findings in the *Journal of Clinical Oncology*, and in December 2021 received FDA approval for the therapy.

“The incidence of severe disease dropped to only 3% in our abatacept treated group, compared to 30% for the control group, so it’s really a striking response,” says Dr. Watkins, an Assistant Professor of Pediatrics at Emory.

“Up until now, there has not been a groundbreaking drug to change the risk of getting GVHD in the first place,” says Dr. Qayed, Director of the BMT Program, an Associate Professor of Pediatrics at Emory and co-first author with Dr. Watkins. “Abatacept has the potential to save many lives by preventing this devastating disease.”



Benjamin Watkins, MD



Muna Qayed, MD



Claudia Morris, MD

Making Gains in Pediatric Emergency Medicine Research

Emergency medicine research has seen tremendous gains since the emergency department at Egleston Hospital was selected a Pediatric Emergency Care Applied Research Network (PECARN) site in 2019.

Under the leadership of Children's Pediatric Emergency Medicine Physicians Claudia R. Morris, MD, Professor of Pediatrics at Emory, and Harold Simon, MD, MBA, Vice Chair of Pediatrics at Emory, the program now receives millions of dollars annually in federal grants and is a high-enrolling site for several NIH-funded multi-center studies—with one of the largest being an \$8.7 million NIH grant for the Sickle cell disease Treatment with ARginine Therapy (STARt) trial.

Several other studies made significant strides in 2021:

- Rob Grell, MD, Pediatric Emergency Medicine Fellow of Children's and Emory, received the Warshaw Fellow's Research Grant to study the use of a virtual reality device to detect concussion in children presenting to the emergency department with head injury. He is collaborating with Michelle C. LaPlaca, PhD, Professor at the Wallace H. Coulter Department of Biomedical Engineering at Georgia Tech and Emory.

- During the pandemic, the emergency department was an active contributor to enrollment for evaluating COVID-19 diagnostics for RADx Atlanta, as many children with COVID-19 and MIS-C presented there for acute care.
- A longitudinal surveillance study of SARS-CoV-2 antibodies in pediatric healthcare workers, led by Dr. Morris and Miriam Vos, MD, Pediatric Hepatologist with Children's and Professor of Pediatrics at Emory, identified novel mechanisms of disease with potential therapeutic opportunities. The study, funded in part by the Emory Woodruff Health Science Center Synergy Award, enrolled more than 640 participants, was published in many multidisciplinary publications, and will now look into SARS-CoV-2 antibodies in response to vaccination.



Miriam Vos, MD

- The work of Chris A. Rees, MD, MPH, Pediatric Emergency Medicine Physician at Children’s and Assistant Professor of Pediatrics and Emergency Medicine at Emory, examined race and ethnicity of children enrolled in clinical trials in the United States, and observed they were not representative of the nation as a whole. He presented these findings at the 2021 American Academy of Pediatrics National Conference & Exhibition. “Our study identifies key areas in which we, as a pediatric research community, can improve enrollment in clinical trials to be more equitable for all groups,” said Dr. Rees. “Results from clinical trials that lack American Indian, Alaska Native, Asian, and Native American-Pacific Islander participants may not be generalizable to all populations that may benefit from trial results.”

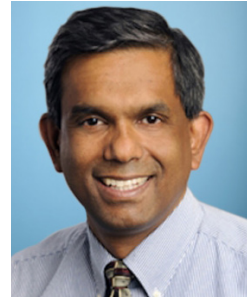


Chris Rees, MD, MPH

First Whole-Genome Study of IBD in African Americans

Race plays a role in genetic risk factors for inflammatory bowel disease (IBD), according to breakthrough research from Children’s and Emory.

Results of the first whole-genome study of IBD in African Americans show the genetic risk landscape is very different in African Americans than people with European ancestry. Lead author Subra Kugathasan, MD, Pediatric Gastroenterologist at Children’s, says future clinical research on IBD needs to take ancestry into account.



Subra Kugathasan, MD

Findings of the multi-center study, which analyzed the whole genomes of more than 1,700 affected individuals with Crohn’s disease and ulcerative colitis and more than 1,600 healthy controls, were published in the American Journal of Human Genetics. As part of the analysis, the researchers developed an algorithm that corrects for ancestry when calculating IBD polygenic risk scores. These scores are tools for calculating gene-based risk for a disease and are used for IBD as well as other complex conditions such as coronary artery disease. The study showed that a gene, PTGER4, that plays an important role in determining the risk for IBD in African Americans, plays a relatively minor role in European populations. In contrast, two gene loci that are important in Europeans—NOD2 and IL23R—play smaller roles in African Americans.

“Even though the disease destination looks the same, the populations look very different, in terms of what specific genes contribute to risk for IBD,” says Dr. Kugathasan, Marcus Professor of Pediatrics and Human Genetics at Emory. “It shows that you can’t develop a polygenic risk score based on one population and apply it to another.”

Medical Genetics Selected a NORD Rare Disease Center of Excellence

The National Organization for Rare Disorders (NORD) has named the Division of Medical Genetics a NORD Rare Disease Center of Excellence. A partnership of Children's and Emory, the newly designated center is one of 31 medical centers nationwide that are part of an innovative network aimed at expanding access, advancing care and enhancing research for rare disease patients in the U.S.

Children and adults living with rare diseases frequently face many challenges in finding a diagnosis and quality clinical care. The average rare disease diagnosis can take several years and require many tests and visits to specialists. There are over 7,000 rare diseases—most of them with a genetic origin—and 25-30 million Americans are estimated to be currently living with rare diseases.

Within the Division of Medical Genetics, researchers focus on a number of specialty areas, such as fragile X syndrome and lysosomal storage disorders. But clinicians have broad experience with many rare diseases. The center is also responsible for follow-up after newborn screening for metabolic diseases within the state of Georgia, and hosts a genetic counseling training program. In addition, genetics researchers counsel families who learn that a child has a previously unrecognized genetic disorder. Several clinical trials are testing the first therapies available for a specific condition, such as an ongoing study of the medication vosoritide, which enhances bone growth in children with achondroplasia; and another study of trofinetide for girls and adolescent females with Rett syndrome.

"We are delighted that NORD has recognized our team's commitment to patients and their families with rare disease," says

Michael J. Gambello, MD, PhD, Clinical Geneticist at Children's and Vice Chair of the Department of Human Genetics at Emory. "We have an

exceptionally dedicated group of geneticists, genetic counselors, metabolic dietitians and support staff."



Michael Gambello, MD





Looking to the Future

Our pediatric research enterprise has experienced astounding growth over the past decade, and we are excited to continue that trajectory into the future. We are proud of the fact that our research enterprise pivoted effectively to meet the challenge of COVID-19 and made a significant contribution to this global emergency. Moving forward, we continue to implement our bold strategic growth plan in research and education that began in 2020. Over a five-year period, Children's and Emory University will lead and accelerate research in a number of critical disciplines, recruit 30 new investigators and add 45 pediatric residents and subspecialty fellows. We will add 40,000 square feet of research space in 2022, and in 2025 we will move into the state-of-the-art Arthur M. Blank Hospital made up of 1.5 million square feet. This growth will give us the manpower and facilities needed to provide the quality care and better health outcomes that kids deserve.





As our research portfolio and extramural funding continue to grow, we are well positioned to enhance our national reputation as one of the leading pediatric research partnerships in the country, especially in these areas:

- **Pediatric technologies** will focus on microsystems-engineered point-of-care technologies, nanomedicine for pediatric cancers and technology-enabled tissue engineering, such as 3D printing and bioprinting, often in collaboration with engineers at the Georgia Institute of Technology.
- **Health disparities research** will include outcomes and population-based research, growing our Sickle Cell Disease Program, and behavioral and mental health initiatives.
- **Sickle cell disease research** will highlight stem cell biology, the basis for blood formation; cellular therapies, including stem cell transplant and gene-based therapy; and drug discovery and development, including small molecules and biologicals. We are recruiting “game changing” faculty to lead and support this effort.
- **Behavioral and mental health research** will enhance behavioral and mental healthcare, improve access to community resources, pioneer prevention through innovative programs, and build a strong foundation through leadership recruitment and partnerships.
- **Cell therapies research** will involve rapid translation of “homegrown” discoveries into early phase clinical trials in our newly established Marcus Center for Pediatric Cellular Therapy.
- **Clinical informatics research** will accelerate the development of evidence-based practices and their implementation into clinical care by partnering with frontline doctors and nurses, data analysts and electronic health record (EHR) developers.
- **Bioinformatics research** will nurture next-generation genome and large biological data-driven discoveries for better treatment of complex and heterogeneous diseases.

We are proud to share with you these examples of some of the exciting work that reflects our progress. As we look to the future, we will continue to leverage our nationally leading clinical volumes and the unique Atlanta partnership of Children’s, Emory University and Georgia Tech to improve the health of children in Georgia, across the nation and around the world.



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