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FOR IMMEDIATE RELEASE

Dr. Steven Baker Awarded \$20,000 Young Investigator Draft Research Grant to Research Cerebral Creatine Deficiency Syndromes

Dr. Steven Baker is among 10 rare disease researchers to be awarded a grant at Uplifting Athletes' Sixth Annual Young Investigator Draft presented by CSL Behring on Feb. 4 at Lincoln Financial Field

Carlsbad, CA (January 13, 2023) – [Uplifting Athletes](#), a nonprofit organization dedicated to serving the rare disease community, has announced that [University of Utah's](#) Dr. Steven Baker will receive a \$20,000 research grant as a member of Uplifting Athletes' 2023 Young Investigator Draft Class.

Dr. Baker is among a group of 10 researchers who will each receive an unrestricted grant for medical research at Uplifting Athletes' sixth annual [Young Investigator Draft](#) presented by [CSL Behring](#) on Feb. 4, 2023, at Lincoln Financial Field in Philadelphia.

Uplifting Athletes is a nonprofit organization that connects student-athletes across the country as well as professional athlete ambassadors with their local rare disease community to raise awareness and funds, and bring inspiration and hope to those impacted by rare diseases, which is approximately one in 10 Americans.

The annual event, inspired by the NFL draft, invests in research which could lead to potential rare disease treatments and cures. Just as the NFL draft rallies around the hope and possibilities that the next generation of game-changing talent will bring, the Young Investigator Draft celebrates the early-career research investigators who are working toward a brighter future for those affected by rare diseases.

“The members of our 2023 Young Investigator Draft Class represent a group of early-career, diverse researchers who are tremendous assets to the rare disease community,” said Rob Long, executive director of Uplifting Athletes. “These individuals stand to make a profound impact on the 30 million Americans impacted by a rare disease. We are thrilled to celebrate these scientists with the same spirit and fanfare that elite athletes receive and the college and pro athletes who join us every year in this effort love sharing the stage with these deserving researchers who represent so much promise for the future,” Long added.

Including the grants to be presented at the 2023 Young Investigator Draft, Uplifting Athletes will have awarded more than \$820,000 in grant funding to 44 rare disease researchers across North America since 2018. Grant submissions for the Young Investigator Draft are evaluated by an expert panel of scientific advisors prior to the selection of each year’s Draft Class. Every researcher is nominated by a patient advocacy organization (PAO) recognized by Uplifting Athletes as a priority partner, with grants equally co-funded by Uplifting Athletes and the nominating PAO. The PAO that nominated and will co-fund Dr. Baker is the [Association for Creatine Deficiencies](#).

Dr. Baker is an assistant professor in the department of pathology at the University of Utah. After completing a bachelor of science in biological sciences at Cornell University, he joined the MD/PhD training program at Baylor College of Medicine. He performed his graduate studies in the laboratory of Dr. Huda Zoghbi studying the molecular basis of Rett syndrome. He continued his medical training as a clinical pathology resident and then transfusion medicine fellow at Stanford University. While at Stanford he carried out research on Cerebral Creatine Deficiency Syndrome (CCDS) as a postdoctoral fellow in the laboratory of Dr. Thomas Montine. His work on CCDS focused on utilizing single cell sequencing resources and immunofluorescence to characterize the creatine synthetic pathway in mammals. Following the completion of his medical training, Dr. Baker joined the faculty of the University of Utah as an associate medical director of transfusion medicine and medical director of the immunohematology reference laboratory at ARUP.

Creatine Transporter Deficiency (CTD) is caused by mutations in the SLC6A8 gene . While patients with CTD have the necessary AGAT and GAMT enzymes to form creatine, the creatine transporter does not function properly. This results in creatine in the bloodstream, but not in the brain and muscles. Individuals affected by Creatine Transporter Deficiency struggle with



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symptoms including intellectual disability, speech delay, seizures, behavioral challenges and low muscle tone.

“Currently there are no effective therapies for children affected by Creatine Transporter Deficiency,” said Dr. Baker. “Funding from the Uplifting Athletes Award will help us test a novel approach for gene therapy to one day help these patients. I couldn’t be more excited to have the support of this amazing organization.”

CSL Behring is a vital partner to Uplifting Athletes, serving as the title sponsor for the Young Investigator Draft since its inception in 2018.

In addition to honoring the new Draft Class, the 2023 Young Investigator Draft will provide a forum to recognize the [newest members of Uplifting Athletes’ Rare Disease Champion Team](#)—an exemplary group of leaders in college athletics who have made a positive and lasting impact on people affected by rare diseases.

For more information or to purchase tickets for the Uplifting Athletes 2023 Young Investigator Draft presented by CSL Behring, please visit <https://charity.pledgeit.org/YID2023>.

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About Uplifting Athletes: Uplifting Athletes is a nonprofit organization founded in 2007 that harnesses the power of sport to build a community that invests in the lives of people impacted by rare diseases. Since its inception, Uplifting Athletes has raised more than \$7 million by engaging athletes in order to positively impact the rare disease community through driving action and funding research. To learn more about Uplifting Athletes visit upliftingathletes.org.

About ACD: The Association for Creatine Deficiencies’ mission is to eliminate the challenges of CCDS. ACD is committed to providing patient, family, and public education to advocate for early intervention through newborn screening, and to promote and fund medical research for treatments and cures for Cerebral Creatine Deficiency Syndromes. Because CCDS mimic symptoms of other medical conditions, patients are often first diagnosed with autism, cerebral palsy, epilepsy, and other disorders. Proper diagnosis and early intervention are critical to establishing interventions needed to improve life quality and longevity for the CCDS patient. For more information regarding ACD, please visit creatineinfo.org.



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About University of Utah Health: [University of Utah Health](#) provides leading-edge and compassionate care for a referral area that encompasses Idaho, Wyoming, Montana, and much of Nevada. A hub for health sciences research and education in the region, U of U Health has a \$458 million research enterprise and trains the majority of Utah's physicians and health care providers at its Colleges of Health, Nursing, and Pharmacy and Schools of Dentistry and Medicine. With more than 20,000 employees, the system includes 12 community clinics and five hospitals. U of U Health is recognized nationally as a transformative health care system and provider of world-class care.