An update from the Consortium of Eosinophilic Gastrointestinal Disease Researchers

Fall 2020

RDCRN Funds CEGIR for a Second Cycle

Consortium to continue research of EGIDs

The Consortium of Eosinophilic Gastrointestinal Disease Researchers (CEGIR) is a multicenter clinical research collaborative focused on EGIDs. CEGIR formed in 2014 as part of the Rare Diseases Clinical Research Network. Last year, the RDCRN awarded $7.57 million to CEGIR for a second 5-year cycle.

In its second cycle of funding, CEGIR will continue longitudinal assessment of OMEGA data. Investigators seek to answer questions such as "What happens to EGID patients over time?" and "What symptoms and genes are associated with EGIDs?" Eosinophilic gastroenteritis (EGE) was also added as a subset of EGID that CEGIR will focus on in Cycle 2, which will advance the understanding of this condition.

Investigators also aim to create a validated endoscopic scoring tool for eosinophilic gastritis (EG), as well as create diagnostic consensus criteria for EG.

The clinical trial that CEGIR will be conducting in this cycle will focus on assessing the utility of an anti IL-13 and IL-4 monoclonal antibody (dupiliumab) in patients with EG and EGE.

Two pilot studies will also be supported, one which looks at alpha-1-antitrypsin (a protein encoded into a specific gene) in EoE, and the other, co-funded with APFED, will examine gastric motility in patients with EG.

CEGIR's annual meeting of investigators will take place virtually on Nov. 17. The meeting will give the consortium participants an opportunity to discuss data trends and direction of new research.

Read more about how CEGIR is advancing the field through collaboration in this article published in *Gastroenterology*.

CEGIR is dedicated to improving the lives of individuals with eosinophilic gastrointestinal disorders through innovative research, clinical expertise and education via collaborations between scientists, health care providers, patients, and professional organizations.

The team has a multidisciplinary approach and integrates expertise in pediatric and adult clinical specialties, including gastroenterology, allergy, immunology and pathology.

Funded by the National Institutes of Health (NIH), and by its patient advocacy group partners APFED, CURED, and EFC, CEGIR is part of the Rare Diseases Clinical Research Network (RDCRN).

Learn more about CEGIR at rdcrn.org/cegir.

CEGIR Studies Currently Enrolling

7801: OMEGA Outcome Measures for Eosinophilic Gastrointestinal Diseases across Ages A Prospective, Multicenter Study to Compare and Validate Endoscopic, Histologic, Molecular, and Patient-Reported Outcomes in Pediatric and Adult Patients with Eosinophilic Esophagitis (EoE), Gastritis (EG), and Colitis (EC)

7808: Use of Unsedated Transnasal Esophagoscopy to Monitor Dietary Management of Eosinophilic Esophagitis in Children

CEGIR Welcomes New Trainee Scholars into Training Program

CEGIR supports investigators who are focusing their research efforts on eosinophil disease research. CEGIR trainees have access to in-person and web-based opportunities to help them be successful in EGID research. Here, we are pleased to introduce the new trainees who were selected to enter CEGIR's 2020 training program.

Paroma Bose, MD

Title: Pediatric Gastroenterology Fellow
Affiliation: Riley Hospital for Children, Indiana University School of Medicine

Dr. Bose is in her final year of her pediatric gastroenterology fellowship. Her research during fellowship includes investigation into the frequency of endoscopy utilization in pediatric EoE and mechanisms for variability in PPI response in pediatric EoE. She is also interested in characterizing patterns of dysmotility in EGIDs.

Joy Chang, MD

Title: Clinical Lecturer of Internal Medicine
Affiliation: University of Michigan

Dr. Chang is an adult gastroenterologist with clinical and research interests in improving the care, quality of life, and patient-provider communications for patients with EoE. Dr. Chang's research focuses on shared decision making, behavioral interventions in EoE care, and patient-centered collaborative research methods.

Pooja Mehta, MD, MSCS

Title: Assistant Professor of Pediatrics
Affiliation: University of Colorado School of Medicine

Dr. Mehta is a pediatric gastroenterologist. Her clinical and research interests include finding innovative methods of improving treatment adherence in adolescents and young adults with EoE. She is interested in the application of health psychology and in the use of digital health tools to reach patients outside of the traditional clinical setting. Her goals are to perform multidisciplinary and pragmatic research with patient input to improve outcomes in EoE.

Melanie Ruffner, MD, PhD

Title: Assistant Professor of Pediatrics
Affiliation: University of Pennsylvania and Children's Hospital of Philadelphia

Dr. Ruffner is a physician-scientist who cares for patients with EGIDs and other allergic conditions. Her research focuses on the role of the esophageal epithelial barrier in EoE, and her translational research studies focus on strategies to improve function of the mucosal barrier in EoE. Dr. Ruffner's project with CEGIR involves examining how proton pump inhibitor medications affect the function of the esophageal epithelial barrier and esophageal epithelial cells behave as a mucosal barrier, bind to each other, and express important membrane proteins.

Justin T. Schwartz, MD, PhD

Title: Assistant Professor
Affiliation: Cincinnati Children's Hospital Medical Center

Dr. Schwartz is a physician-scientist with clinical training in Allergy and Clinical Immunology. He provides clinical care for patients with eosinophilic disease. Dr. Schwartz's project with CEGIR is focused on further evaluating the potential role of eosinophil progenitor cells as a peripheral blood biomarker for disease activity and better understanding which these cells are recruited into the inflamed esophageal tissue and contribute to local inflammation. His research into how immunologic progenitor cells participate in the pathophysiology of EGID will help us better understand disease mechanisms to identify potential new therapeutic targets and biomarkers for disease monitoring.

Benjamin Wright, MD

Title: Assistant Professor
Affiliation: Mayo Clinic Arizona and Phoenix Children's Hospital

Dr. Wright is an allergist/immunologist whose research focuses on mechanisms of oral immunotherapy, mouse models of food allergy, and the role of IgG4 in EoE. Dr. Wright's projects with CEGIR are to examine the relationship between IgE-mediated food allergy and EoE by studying patients undergoing milk oral immunotherapy in order to better understand the pathogenesis of EoE, and to validate a tool that uses computer image analysis to quantify eosinophilic inflammation in tissue biopsies.

Learn more about CEGIR Trainee Scholars on CEGIR's website.

Highlights of Journal Publications from CEGIR in 2020

Molecular, endoscopic, histologic, and circulating biomarker-based diagnosis of eosinophilic gastritis: Multi-site study. *J Allergy Clin Immunol*. Jan. 2020 Volume 145, Issue 1, Pages 255–269: This paper describes new testing platforms for blood and tissue that may help guide clinicians to a diagnosis of eosinophilic gastritis (EG) in the future. The samples analyzed by the CEGIR investigators enabled them to develop a molecular profile that correlated with findings from endoscopy and histology. Levels of eotaxin-3 in the blood were strongly associated with expression of gastric cytokine CCL26. Additional studies are needed to validate these findings before these new testing platforms could be considered for clinical use.

Eosinophilic gastrointestinal disease below the belt. *J Allergy Clin Immunol*. Jan. 2020 Volume 145, Issue 1, Pages 87–89: This article focuses on unmet needs, barriers, and future directions in patients with non-esophageal EGIDs, such as: 1. Clear diagnostic criteria for EG, EGE, and EC must be established; 2. outcome measures, such as patient-reported outcomes and endoscopic and histologic assessments, need to be developed; 3. More work is required to understand pathogenesis; and 4. Longitudinal trials are needed to better understand disease mechanisms and long-term outcomes.

High Patient Disease Burden in a Cross-Sectional, Multicenter Contact Registry Study of Eosinophilic Gastrointestinal Diseases. *J Pediatr Gastroenterol Nutr.* Jun 2020; PMID: 32541201. Using data collected through its contact registry, CEGIR researchers characterized and contrasted patient-reported symptoms and comorbidities in non-EoE EGIDs (eosinophilic gastritis, gastroenteritis, and colitis) compared with EoE. The researchers found patients with non-EoE EGID reported more frequent symptoms of nausea, abdominal pain, diarrhea, constipation, and bloating, and were more likely to report higher frequency of fatigue, isolation, and deep muscle or joint pain. More research is needed to better understand factors that may contribute to the high disease burden of these conditions.

To access additional publications from CEGIR, visit rdcrn.org/cegir.

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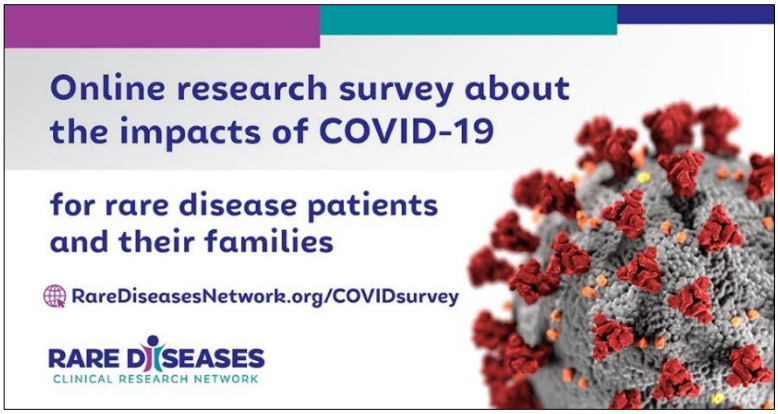
CEGIR Vision Statement: CEGIR actively promotes a culture of diversity and inclusivity in its membership, in its innovative research studies and its educational initiatives.

Survey on the Impacts of COVID-19 on Rare Diseases Patients and Families

Rare disease patients and caregivers: How are you being impacted by the novel coronavirus pandemic? Please complete a 20-minute online research survey from home to share your experiences. This study is being conducted by the NIH's Rare Diseases Clinical Research Network. Is access to care changing? Can you get needed medical and nutritional supplies? Are stress and anxiety impacting you and your family? Your responses may help researchers understand the impacts of COVID-19 on the rare disease community. Complete the survey or learn more at https://RareDiseasesNetwork.org/COVIDsurvey #COVID19



View the interim results of this survey.

The Rare Diseases Clinical Research Network will make every effort to enroll all the patients we can, but we cannot make any guarantees that we will be able to enroll everyone in a study who wants to participate. Participation in research studies is voluntary. Deciding not to participate in a research study does not affect your ability to receive care at any of our Clinical Centers or from other physicians. The Rare Diseases Clinical Research Network (RDCRN) was established by the Office of Rare Diseases Research, NCATS, National Institutes of Health (NIH) to develop research studies for rare diseases, and to encourage cooperative partnerships among researchers at over 150 clinical centers around the world. This increased cooperation may lead to discoveries that will help treat and perhaps prevent these rare diseases, as well as produce medical advances that will benefit the population in general. The Rare Diseases Clinical Research Network is comprised of a Data Management and Coordinating Center and 22 consortia studying over 200 rare diseases. CEGIR (U54AI117804) is a part of the NCATS Rare Diseases Clinical Research Network (RDCRN). RDCRN is an initiative of the Office of Rare Diseases Research (ORDR), NCATS, funded through a collaboration between the NCATS, the NIAID and the NIDDK. CEGIR is also supported by patient advocacy groups including APFED, CURED and EFC. The National Institutes of Health does not endorse or recommend any commercial products, processes, or services. The views expressed in written materials or publications do not necessarily reflect the official policies of the Department of Health and Human Services; nor does mention by trade names, commercial practices, or organizations imply endorsement by the U.S. Government.
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