ECD GLOBAL ALLIANCE
Supporting those affected by
Erdheim-Chester Disease worldwide.

P.O. Box 775
DeRidder, LA 70634
support@erdheim-chester.org
+1-337-515-6987

www.erdheim-chester.org

2021 Grant Award Announcement

The Erdheim-Chester Disease Global Alliance awards Young Investigator Research Grant to Italy doctor to explore the genetic landscape of ECD.

The ECD Global Alliance (ECDGA) has awarded the 2021 ECD Research Grant to Dr. Francesco Pegoraro, with Meyer University Children's Hospital, Florence, Italy. The winning proposal, *Exploring the genetic landscape of* Erdheim-Chester disease by integrating GWAS and -omic data, was selected for funding for $50,000. The ECDGA is honored to be funding this groundbreaking work and believes it will result in a brighter future for ECD patients and a better understanding of histiocytosis.

Dr. Pegoraro's study is designed to clarify the genetic predisposition to ECD. It is hoped that the information learned through this study will provide direct benefits to future patients. The discovery of the genetic patterns of ECD predisposition and the possible definition of novel pathomechanisms might **help identify potentially targetable pathways and introduce novel treatments**. The study includes a network of clinicians who significantly contributed to ECD pathogenesis and the introduction of targeted approaches. If a genetic predisposition is found, the involved clinicians will further validate at preclinical and clinical levels, the pathways or biological mechanisms identified by the genetic study. In this case, clinicians will also verify through international databases if there are drugs that can potentially target the identified pathways and proceed to clinical investigation.

Dr. Pegoraro's synopsis: "We expect to identify differently methylated and/or expressed genes in ECD patients, compared to controls. The patients' clinical data will be collected from medical charts and matched with genetic results. We expect to identify if there is an association between the genetic variants or the epigenetic profiles of affected patients and the ECD-specific clinical manifestations or somatic mutations. We will integrate GWAS data with methylation and transcriptomic data to identify polymorphisms influencing methylation and/or gene expression. Outputs might also include the description of involved biological pathways and the identification of potential therapeutic targets and disease biomarkers."

The grant review team has confidence that Dr. Pegoraro's study has merit and recognizes the expertise of his multi-institutional team to carry out this study. "The investigator is a post-doctoral fellow and has published as a co-author related to Langerhans Cell Histiocytosis (LCH), a disease related to ECD..

Research is a mission-critical goal for the ECDGA. The organization has previously awarded ten (10) medical research grants, devoting over $800K to medical research. Funding for grants comes from generous, private donations. ECD research efforts, backed in part by the non-profit, have recently led to an FDA approval of a drug called vemurafenib, originally developed to treat other types of cancers, such as melanoma skin cancer, to treat ECD patients with the *BRAF*V600E mutation.

Erdheim-Chester Disease is considered a histiocytic neoplasm (type of blood cancer), an ultra-rare condition with no known cause, and is considered under-diagnosed. The illness is characterized by the accumulation of histiocytes, cells that normally fight infections, in tissue and organs. The tissue and organs become dense and fibrotic due to the infiltration of the histiocytes and can lead to organ failure unless a successful treatment is found.

The ECD Global Alliance is a 501(c)(3) non-profit organization dedicated to the awareness, support, education, and research related to Erdheim-Chester Disease.

Contact: Jessica Corkran | jessica.corkran@erdheim-chester.org

###