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# IDF Research Grant Program

Now Accepting Applications for 2019!

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will also be given to studies that contribute to the body of medical knowledge in PI.

The program is open to applicants currently based in the U.S. and will consist of one-year grants. Award Value: \$25,000 - \$50,000 (a somewhat higher level of support is available for a few exceptional proposals).

## IDF Research Committee

Each research proposal is reviewed by IDF's Research Committee, which recommends funding for the highest scored proposals. The grant review process is based on the peer review system utilized by the National Institutes of Health (NIH). Scoring of grants is done individually by each member of the Research Committee and scores are added together to determine the final ranking of each proposal. Any reviewer with a conflict of interest is excused from voting on a specific proposal. The grant selection process is unbiased and independent; awards are based solely on scientific merit.

## Application Process

1. Download and complete the IDF Research Grant Guidelines and Application Form below.
2. Attach the following: CVs and or biosketches, Letters of support/recommendation, and Letters from collaborators agreeing to participate
3. Submit the completed application with attachments to:  
[IDFGrant@primaryimmune.org \(mailto:IDFGrant@primaryimmune.org\)](mailto:IDFGrant@primaryimmune.org).

**The application, along with the attachments must be assembled and submitted electronically as a single PDF to [IDFGrant@primaryimmune.org](mailto:IDFGrant@primaryimmune.org)**

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## MORE ABOUT THE IDF RESEARCH GRANT PROGRAM

Since 2013, IDF Walk for Primary Immunodeficiency has raised more than \$3 million. Because of the dollars raised by IDF walk teams and their supporters, IDF has established the IDF Research Grant Program, awarding grants to help improve the diagnosis, treatment and quality of life of people living with PI.

Ranging from research on Common Variable Immune Deficiency (CVID), Severe Combined Immune Deficiency (SCID), XMEN and Selective IgA, these researchers are identifying and understanding the underlying issues of these chronic diseases. With research comes better diagnoses, lives and potentially cures for those with PI.



### The Immune Deficiency Foundation Research Grant Program



#### Background

In 2016 the IDF Board of Trustees decided to allocate a portion of the funds raised from the successful IDF Walks Program to an IDF Research Grant Program designed to encourage and support patient-oriented research on primary immunodeficiency diseases (PI). The idea is to award seed grants that will support well-defined research projects that have a specified benefit for improving the treatment, health, disease management or diagnosis of persons with PI. The program is open to applicants based in the U.S. and consists of one-year grants. Award values are from \$25,000- \$50,000, and a somewhat higher level of support is available for exceptional proposals.

#### IDF Research Committee

The program is a competitive process with a formal committee to read and score grants. The committee is composed of clinicians, community member representatives and IDF staff. The committee members considered each application individually and scored each individually on the following metrics:

- Significance
- Approach
- Qualifications
- Facilities

Those applications that scored the highest received detailed discussion among the committee members for consideration for funding. IDF was fortunate in that we received many high quality grant proposals. Unfortunately, IDF was not able to fund all applications.

#### Committee Members

Clinicians	Community Representatives	IDF Staff
Mike Blasse, MD	Richard Low	Kathy Arilla
Mark Ballou, MD	Felicia Morton	Marcia Boyle
Carla Duff, CGCP, MSN, CCRP	Brian Rath	Christopher Scalchunes
Adina Kay Knight, MD	Yvette Shorten	
Elena Perez, MD PhD		

#### Deliverables

Each grantee is required to submit to IDF an interim progress report covering the first 6 months of the grant. At the end of the grant period, the grantee must submit to IDF a final report and scientific summary of accomplishments. Ultimately, grantees are required to produce a final work product that may be one of the following:

- A manuscript for publication in a peer reviewed journal
- An abstract presented at a national scientific assembly, meeting or congress

#### Grants Awarded

##### Identification of T-cell receptor clonotypes important in the pathogenesis of CVID

Sara Barnmiller, MD  
Allergy Immunology Fellow  
Department of Allergy and Immunology  
Massachusetts General Hospital

We are interested in looking at the T-cells in patients with CVID to try to determine if there are differences between the T-cells of CVID patients compared to healthy patients. We also want to investigate if there are specific T-cells that are causing the disease.

Some patients with CVID have gastrointestinal complications, including problems with absorbing nutrients, diarrhea, and weight loss. We will evaluate patients who have gastrointestinal complications with CVID (called CVID-associated enteropathy) to see if we can evaluate their T-cells to better understand why this disease occurs. We will compare the T-cells in the gastrointestinal (GI) system to the T-cells in the blood. We will also be evaluating the T-cells in patients with the GI complications in CVID to those without GI complications.

Our hope is that if we could identify specific T-cells that are causing this disease then we could potentially target these T-cells to protect patients. We would try to prevent the disease from occurring or getting worse if we can identify them early on in the course of the disease.

##### Prevalence of Fatigue in Common Variable Immunodeficiency

Joud Hajar, MD  
Assistant Professor  
Pediatrics, Allergy and Immunology  
Baylor College of Medicine

This research project was born in our clinic, where our patients with CVID often reported having fatigue. Fatigue is not often discussed or treated by providers. We believe that reporting lower quality of life compared to patients who do not have fatigue. This is especially relevant to patients with CVID, in which studies have shown that having a poor quality of life predicted shorter life span compared to patients who reported a good quality of life.

Persons with CVID have more fatigue compared to the general population and that some of the complications that result from having CVID such as lung or gastrointestinal damage were associated with having fatigue. Our goal is to determine how common fatigue is in CVID, who is at risk to develop fatigue, and what are the effects of immunoglobulin therapy on fatigue. The results from this proposed research have the potential to significantly alter how doctors evaluate fatigue in subjects with CVID, and how they prescribe immunoglobulin therapy in a way that notably decreases fatigue, improves the quality of life and potentially survival in the CVID population. Importantly, identifying the risk factors to develop fatigue will allow health care providers to recommend specific therapies to treat it.

##### Characterizing T-cell lymphopenic infants identified on routine newborn screening

Artemio Jorgoso III, MD, PhD, MPH  
Assistant Professor  
Pediatrics and Medicine  
Allergy & Immunology  
Fennell Institute for Medical Research

Severe combined immunodeficiency (SCID), a life threatening but curable PI, belongs to a category of conditions where early detection and treatment can significantly improve morbidity and/or mortality.

SCID patients have decreased number and function of T cells, which play a crucial role in the immune system, such as fighting viruses and fungi. All but three states in the US screen or plan to screen for SCID. All these programs rely on quantifying T cell receptor excision circles (TREC), circular pieces of DNA that are produced during normal T cell development and maturation. Individuals with abnormal T cell development resulting in reduced T cell numbers, will have low TREC counts. These programs identify infants with other non-SCID PIs that primarily affect T cells, such as DiGeorge Syndrome. They also detect infants with low T cell counts (known as T cell lymphopenia (TCL)) for whom the underlying cause is unknown and evidence-based management guidelines are lacking.

A better understanding of TCL is desirable because it will improve our knowledge and facilitate the development of evidence-based guidelines for diagnosis and management of this condition. Thus, these studies can guide the development of best practices to maximize patient outcomes in a cost-effective manner.

##### A new pathobiological basis for the treatment of XMEN primary immunodeficiency

Sonia Sharma, PhD, BS  
Assistant Professor  
Cellular Biology  
La Jolla Institute for Allergy & Immunology

Immunodeficiency is a state in which the immune system's ability to fight infectious diseases or cancer is compromised or entirely absent. The study of these rare diseases has been instrumental in providing valuable insight into the biology underlying the proper function of the immune system, and in the search for developing specific therapies to treat these debilitating diseases.

In this proposal, we are exploring a new molecular basis for a linked immunodeficiency with Magesium defect (Epstein-Barr Virus (EBV) infection and Neoplasia (XMEN) disease, which is caused by genetic mutations in the gene encoding magnesium transporter 1 (MACT1). We propose to shed valuable new insight into the role of MACT1 and magnesium as a regulator of the early or innate immune response to bacteria, viruses and tumor cells.

Our new insight into the pathobiological basis of XMEN immunodeficiency could be therapeutically beneficial for the treatment of XMEN disease, as XMEN patients may benefit from anti-biotic, anti-viral or interferon-based therapies to treat their early immune deficiency.

##### IKAROS deficiency and CVID: Disease biology and specific therapeutic approaches

Sergio Rosenzweig, MD, PhD  
Chief Immunology Service  
Department of Laboratory Medicine  
National Institutes of Health

Common variable immune deficiency is the most common, treatable primary immune deficiency. Treatment is usually focused on replacement of deficient immunoglobulin. Approximately half of the patients with common variable lymphoproliferative disease and this further dictates therapy with an emphasis on immune suppression or anti-inflammatory treatment.

Over the past 10 years, it has been revealed that 10-20% of patients with common variable immune deficiency have single gene defects that contribute to the evolution of their immune deficiency. IKAROS deficiency is one of the most recently described single gene defects. The goal of this proposal is to understand how IKAROS deficiency impacts the patient's DNA and to pilot treatments that would uniquely be able to treat IKAROS deficiency.

##### Deciphering function of HLA encoded microRNA in primary immunodeficiency

Nilesh Chitrin, PhD  
Postdoctoral Fellow  
Pathology and Laboratory Medicine  
Children's Hospital of Philadelphia

Selective IgA deficiency patients suffer from increased incidence of respiratory diseases and are at greater risk of anaphylactic shock during blood transfusions. Our data indicate that approximately half of the patients with selective IgA deficiency have elevated miR-4891-5p increased IgA secretion. We propose to confirm these observations in primary B-cells obtained from blood of selective IgA deficiency patients. We will decrease the expression of miR-4891-5p in patient cells and determine whether IgA secretion can be restored. Many responses targeting microRNAs are already in clinical trials. This research may uncover miR-6891-5p as a target for therapeutic intervention.

<https://primaryimmune.org/resource/idf-research-grant-recipients>  
Click to see the grants awarded in 2016-2017



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quality of life of persons with PI through advocacy, education, and research.

There are approximately 250,000 people who are diagnosed with PI in the U.S.,  
and thousands more go undetected.



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