

Matthew Vesely, MD PhD  
Yale University

Edward Cowen, MD MHSc  
National Institutes of Health

## MDS Mentorship Summary Report

The Medical Dermatology Society (MDS) mentorship award allowed me to become immersed in an incredible clinical program dedicated to complex medical dermatology at the National Institutes of Health (NIH). My mentor, Dr. Edward Cowen who leads the dermatology consultation service at the NIH, provided with one of the most rewarding clinical experiences in residency.

Every day, I had the great pleasure of joining Dr. Cowen and his consult team in seeing patients with a wide variety of rare diseases from monogenic autoinflammatory diseases, primary immunodeficiencies, genodermatoses and adverse drug reactions from novel targeted chemotherapeutics. Some of the most memorable patients were those with autoinflammatory diseases treated with targeted therapies. For example, one patient with neonatal onset multisystem inflammatory disease (NOMID) was currently well controlled on anakinra. Another patient with STING-associated vasculopathy with onset in infancy (SAVI) was being treated with Janus kinase (JAK) inhibitors since these patients have elevated levels of type I interferons. Additionally, numerous patients with primary immunodeficiencies were examined including, Hyper IgE Syndrome, GATA2 deficiency, chronic granulomatous disease, deficiency of ADA2, and WHIM (warts, hypogammaglobulinemia, infections and myelokathexis). Again, many of these patients were enrolled in clinical trials targeting the underlying pathogenesis such as blocking CXCR4 for WHIM syndrome. Many of the diseases above were characterized at the NIH and thus leading experts in infectious disease, endocrinology, pediatrics, hematology and immunology were often in dermatology clinic to provide multi-disciplinary care.

Perhaps more thrilling than the opportunity to see rarely encountered dermatological diseases, were the new diseases being characterized at the NIH. On two separate occasions, we had patients with profound skin findings whose constellation of signs, symptoms, immune profiling and genetic mutations do not fit into previously known diseases. I presented one of these patients at the NIH Dermatology Grand Rounds, allowing me to better comprehend the translational clinical research that is required for disease discovery. For me, this highlights the limitations of our knowledge in medicine and inspires me to delve deep into disease pathogenesis, whether rare or common.

In addition to Dr. Cowen, I was fortunate enough to interact and learn from the other incredible faculty at the NIH Dermatology branch. The first time I saw a patient with xeroderma pigmentosum was with Drs. John DiGiovanna and Kenneth Kraemer. With my interest in basic science and translational science, I was able to attend journal club with Drs. Heidi Kong, Isaac Brownell and Chris Nagao.

Dr. Cowen and other mentors at the NIH will undoubtedly shape my career. My hope is to ultimately become a player in the collaborative network of academic dermatologists, basic scientists and translational scientists involved in developing emerging therapeutics. I am deeply grateful to the MDS and to Dr. Cowen for this exceptional opportunity to help transform my clinical interests into a career focused on academic dermatology.