

Emil Skamene (1941–2024)

By Philippe Gros & Erwin Schurr

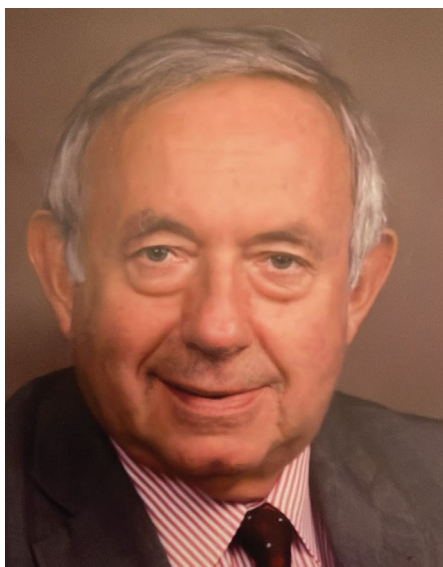
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It is with much sadness that we mark the passing of Emil Skamene, professor emeritus, researcher and clinician at the Research Institute of the McGill University Health Centre. Emil is remembered for his contributions to the field of genetic studies of susceptibility to infections, which he developed and popularized over three decades.

Emil was born in Poland in the town of Buczacz (now located in Ukraine) on 27 August 1941. He miraculously escaped the Nazi death camps as an infant but lost many family members. He was raised by his adoptive Skamene family in the Czech Republic. He obtained his MD and PhD degrees from Charles University in Prague; he then undertook post-doctoral work at Harvard Medical School in Boston and completed his specialty in allergy and immunology at McGill, where he remained.

Emil was a pioneer of, and internationally recognized for, the study of the host genetic basis of susceptibility to infections. His work rejected the then dominating microorganism-centric view of infectious disease and was a watershed for infectious diseases research. He was inspired by the landmark studies of Lurie and Dannenberg, who from the 1930s onward bred inbred strains of rabbits that were inherently resistant and susceptible to aerosol infection with *Mycobacterium tuberculosis*. He would often refer to similar work on *M. tuberculosis* by Wright and Lewis in hamsters and Lynch and Dubos in mice as planting the seeds for his own work¹.

When Emil started his career mice were typically considered just mice, with little attention to their genetic background and research focused on the use of outbred stocks that were inexpensive and easy to breed. Emil insisted on using multiple strains of mice to study biological traits, arguing that this would represent an opportunity to study natural modulating alleles that are fixed to homozygosity by inbreeding². Using such combinations, Emil championed the use of mouse models of experimental infections with several bacterial (*Mycobacterium* species, *Salmonella*, *Pseudomonas*, *Legionella* and *Listeria*), parasitic (*Leishmania*, *Toxoplasma* and *Plasmodium*), viral (Influenza, Cytomegalovirus and Coronavirus) and fungal (*Candida*) pathogens. His group was able to map and identify major gene



effects imparting susceptibility and resistance to these infections.

His work in inbred strains and the mapping of the *Bcg* locus, modulating susceptibility to mycobacterial infections, led to the breakthrough of the positional cloning of *Nramp1* (also known as *Slc11a1*)³. He discovered single gene effects for susceptibility to many other infections and adapted the use of recombinant inbred strains for mapping of genes affecting susceptibility to tuberculosis (*Icsbp*, also known as *Irf8*), salmonellosis (*Ity*, also known as *Slc11a1*), leishmaniasis (*Lsh*, also known as *Slc11a1*), Legionaire's disease (*Lgn1*, also known as *Naip5*), cytomegalovirus infection (*Cmv1*, also known as *Klra8* or *Ly49h*) and candidiasis (*Hc*, also known as *C5a*), among others.

He developed genetic tools in the form of AcB/BcA recombinant congenic strains of mice to deconvolute complex genetic effects. This led to the demonstration that pyruvate kinase (*Pklr*) deficiency causes resistance to malaria in mice and humans. These strains led to the discovery that the metabolic product (cysteamine) of the enzyme vanin-1 can protect against blood-stage and cerebral malaria. Emil applied genetic analyses in mice to study pathogenesis of other disorders, including cardiovascular, metabolic, neurodegenerative and inflammatory diseases, establishing the broad applicability of mouse genetics to understand human disease.

In 1988, Emil founded the Centre for the Study of Host Resistance while continuing his own research and clinical duties at the Montreal General Hospital. He later served as the scientific director of the McGill University Health Centre, where his leadership impacted clinical practices and the future of healthcare. Emil was a co-founder of the Canadian Genetic Diseases Network, a Distinguished Scientist of the Canadian Society for Clinical Investigations, a member of the Royal Society of Canada and a recipient of the Killam Prize from the Canada Council of the Arts and of the G. Mendel Award from the Czech Academy of Sciences.

With his boundless energy and enthusiasm, he nurtured the career of numerous trainees and junior colleagues whom he recruited to the internationally recognized Centre for the Study of Host Resistance; many became leading researchers in this field. He inspired the careers of many who embraced genetics to study rare sporadic or familial susceptibility to infections in humans, providing a major impetus for the mapping and cloning of genes underlying inborn errors of immunity.

We join our voices to those of many who grieve the loss of a giant, a generous leader, a gentle soul and a consummate scientist.

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Competing interests

The authors declare no competing interests.