

# ***Stronger Together: Tackling Rare Diseases Through Team Science***

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Mini Med School 2025

Exploring Rare Diseases: Discovery to Treatment

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**Kelly Brown, PhD**

UBC Department of Pediatrics and  
Division of Rheumatology at the  
BC Children's Hospital Research Institute









# Objectives of this presentation

- Introduce you to the family of **rheumatic diseases**
- Focus on childhood-onset **vasculitis** and the **Pediatric Vasculitis Initiative**
- Show how **collaboration can accelerate research** for an ultra rare disease
- Leave you **curious and inspired** about pediatric rheumatology

# From basic to translational scientist



Video from the NIH National  
Centre for Advancing  
Translational Sciences

## CHARACTERISTICS OF A TRANSLATIONAL SCIENTIST

Translation is the process of turning observations in the laboratory, clinic and community into interventions that improve the health of individuals and the public – from diagnostics and therapeutics to medical procedures and behavioral changes. The professionals involved in this process, either developing interventions or improving the process itself, are *TRANSLATIONAL SCIENTISTS*.

### RIGOROUS RESEARCHER

Conducts research at the highest levels of rigor and transparency, possesses strong statistical analysis skills, and designs research projects to maximize reproducibility.

### BOUNDARY CROSSER

Breaks down disciplinary silos and collaborates with others across research areas and professions to collectively advance the development of a medical intervention.

### TEAM PLAYER

Practices a team science approach by leveraging the strengths and expertise and valuing the contributions of all players on the translational science team.

### PROCESS INNOVATOR

Seeks to better understand the scientific and operational principles underlying the translational process, and innovates to overcome bottlenecks and accelerate that process.

### DOMAIN EXPERT

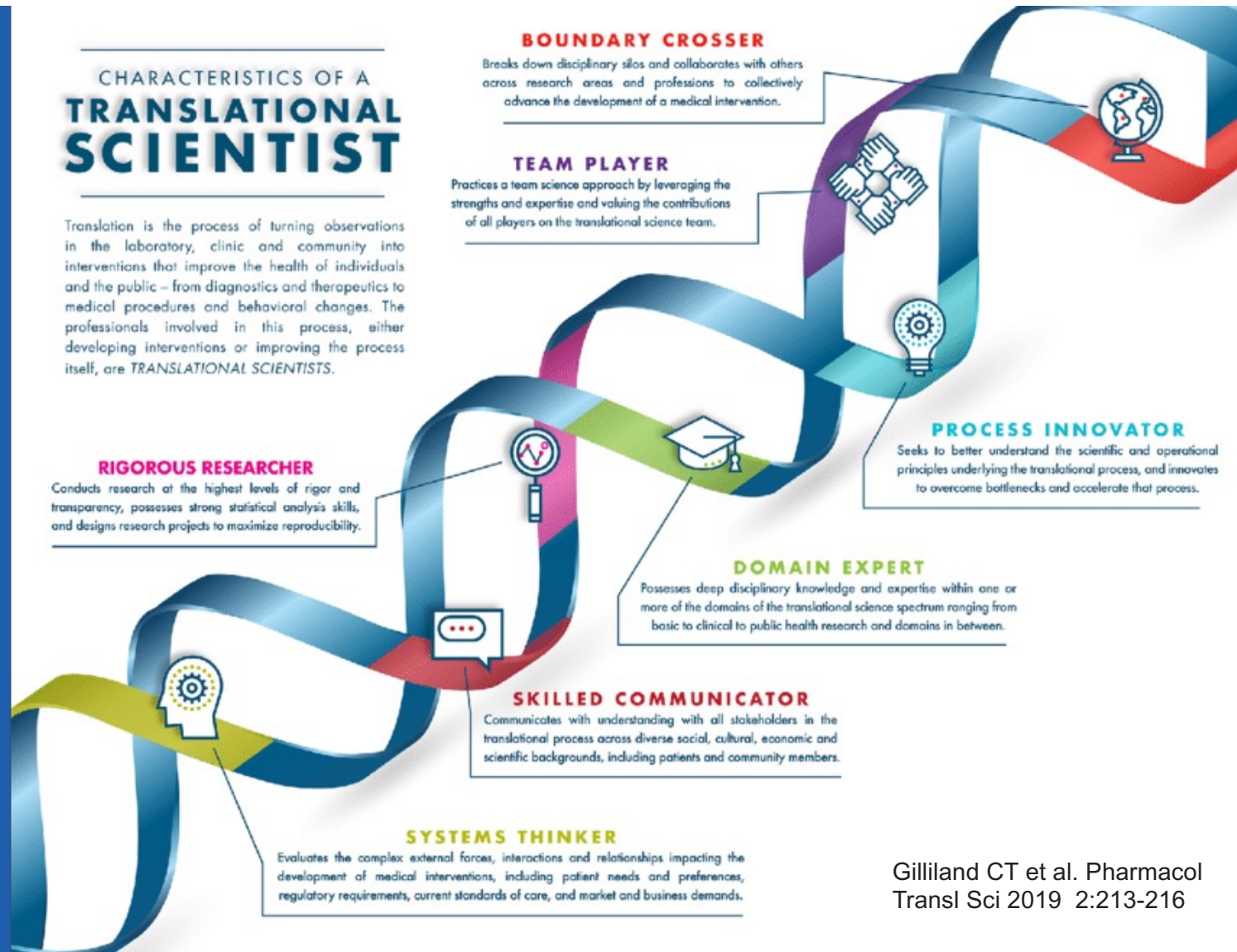
Possesses deep disciplinary knowledge and expertise within one or more of the domains of the translational science spectrum ranging from basic to clinical to public health research and domains in between.

### SKILLED COMMUNICATOR

Communicates with understanding with all stakeholders in the translational process across diverse social, cultural, economic and scientific backgrounds, including patients and community members.

### SYSTEMS THINKER

Evaluates the complex external forces, interactions and relationships impacting the development of medical interventions, including patient needs and preferences, regulatory requirements, current standards of care, and market and business demands.

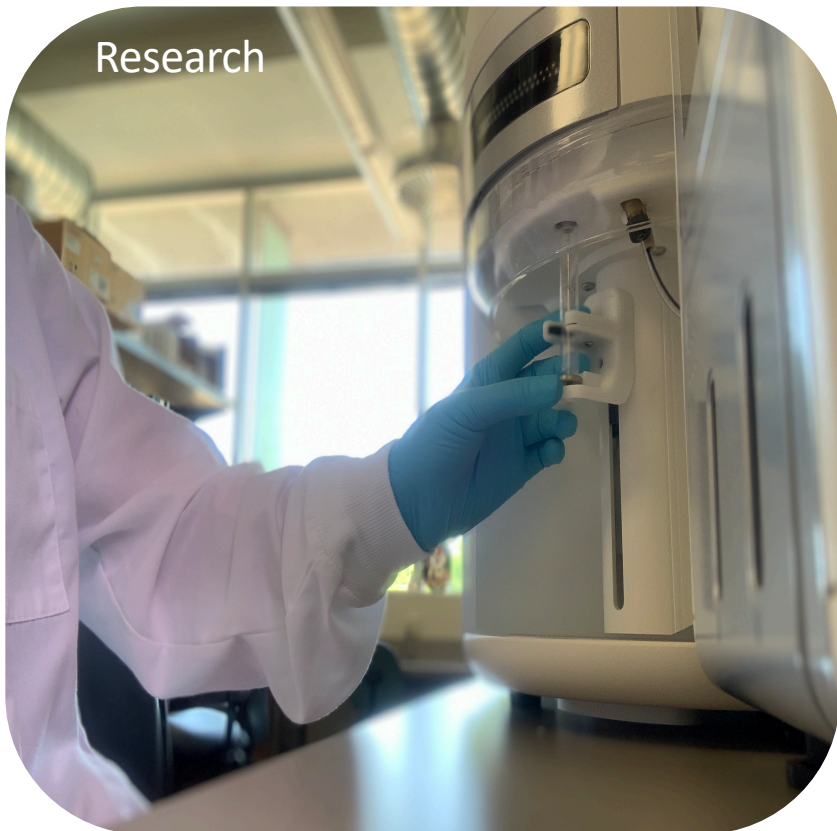


Gilliland CT et al. Pharmacol  
Transl Sci 2019 2:213-216



**BROWN LAB**  
TOGETHER IN DISCOVERY

Research



Testing



Training

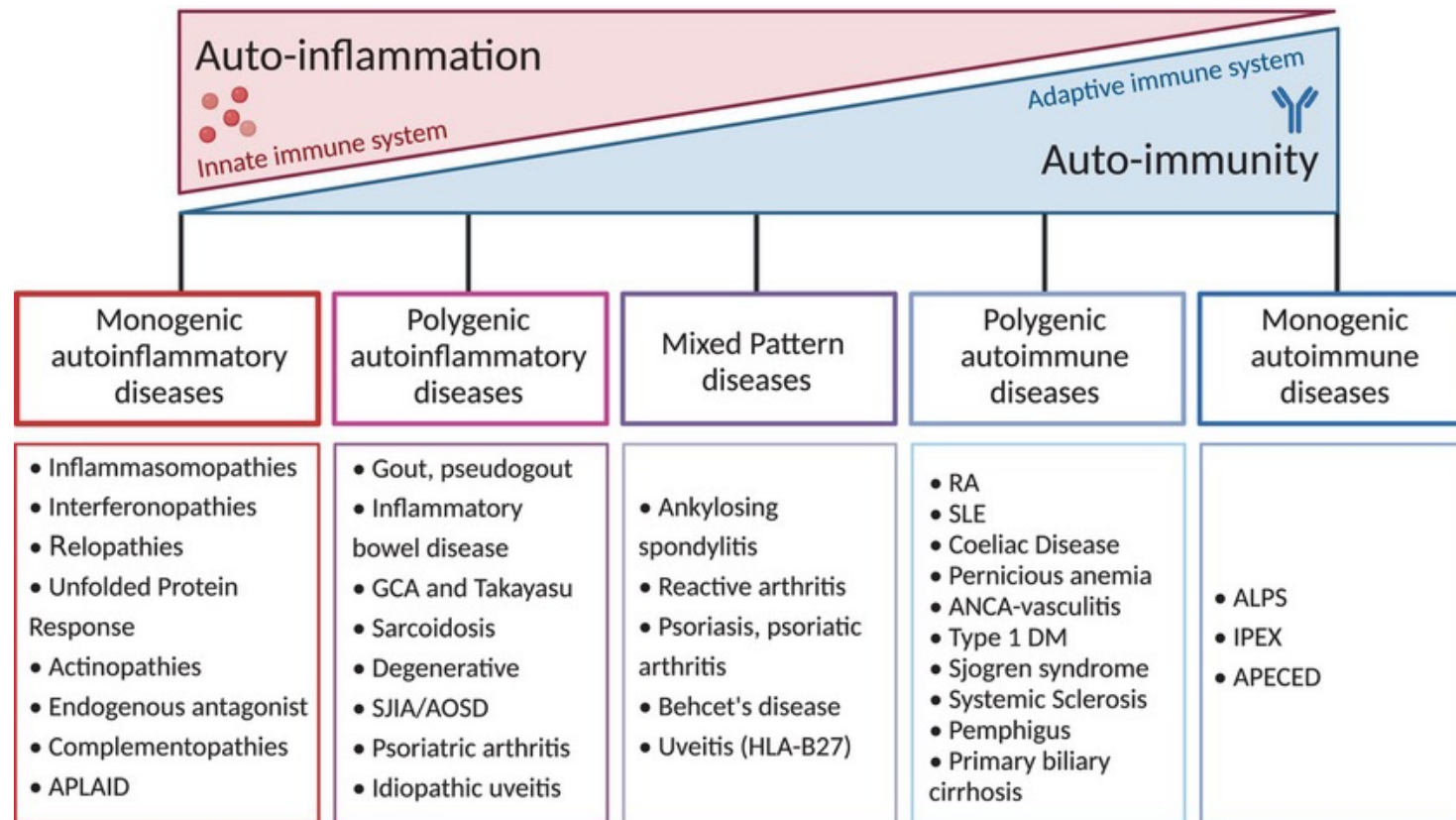


Advocacy



[Learn more here](#)

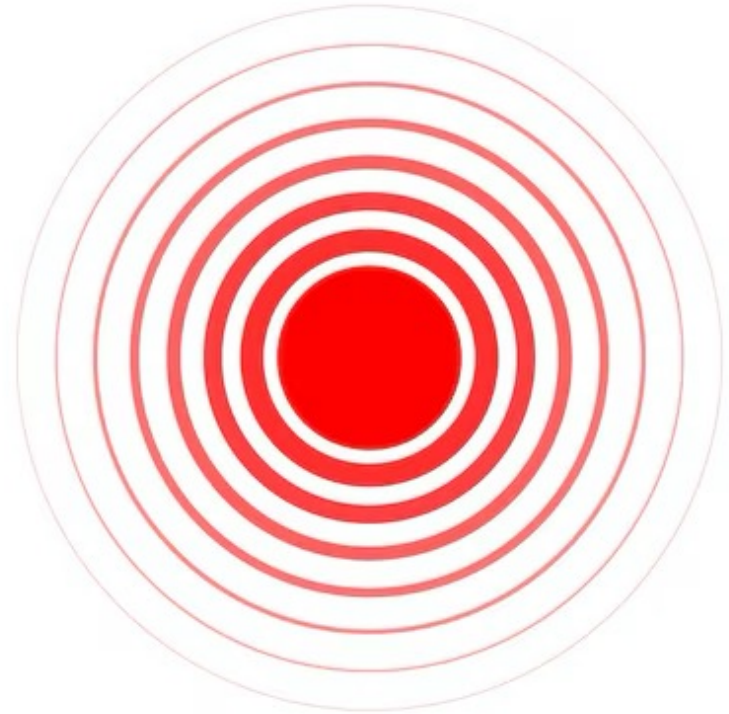
# What are Rheumatic Diseases?

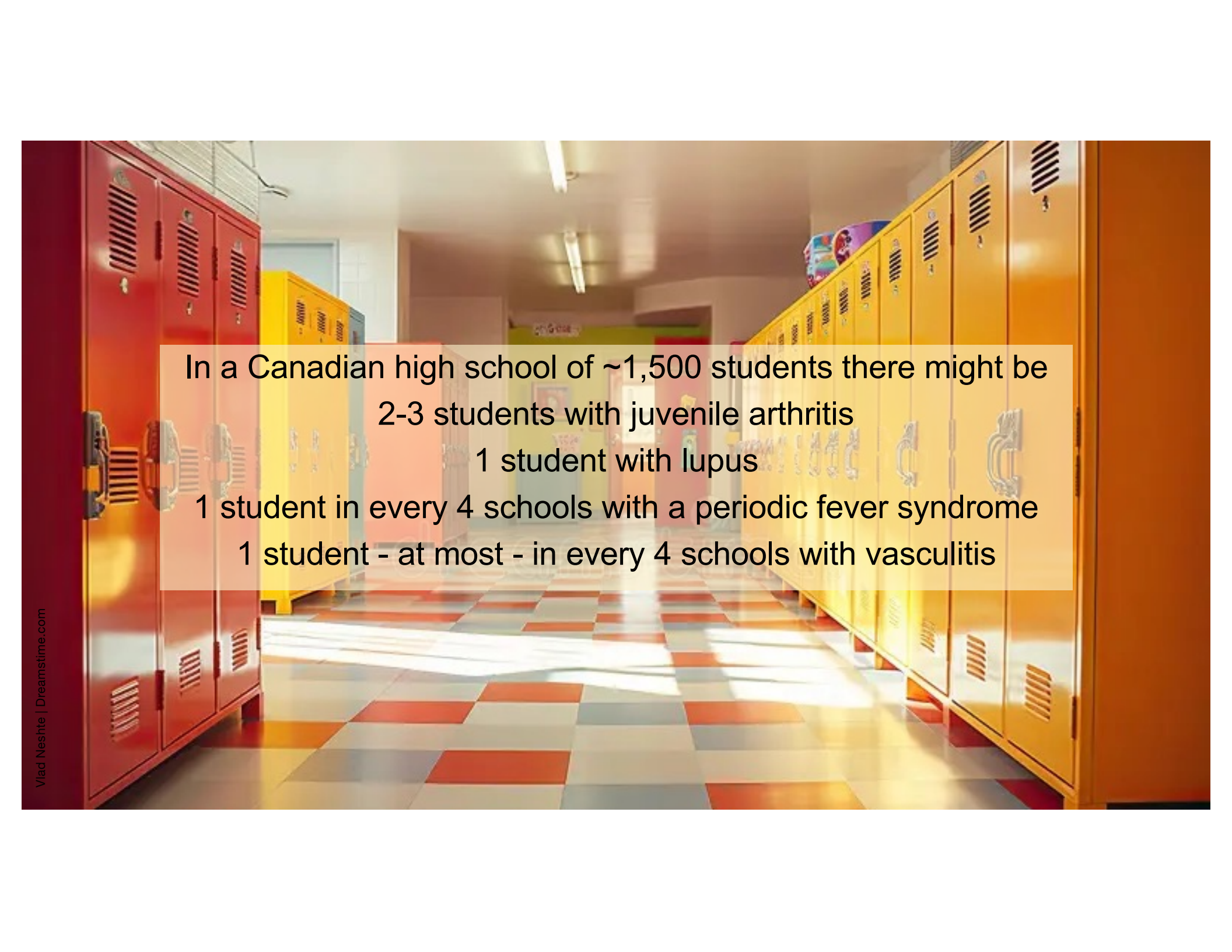


Merlo Pich LM et al. FEBS J. 2024 291(20):4414-4432.



Rheumatic diseases  
have in common  
*uncontrolled inflammation*  
in joints, muscles,  
tissues, and major  
organs.

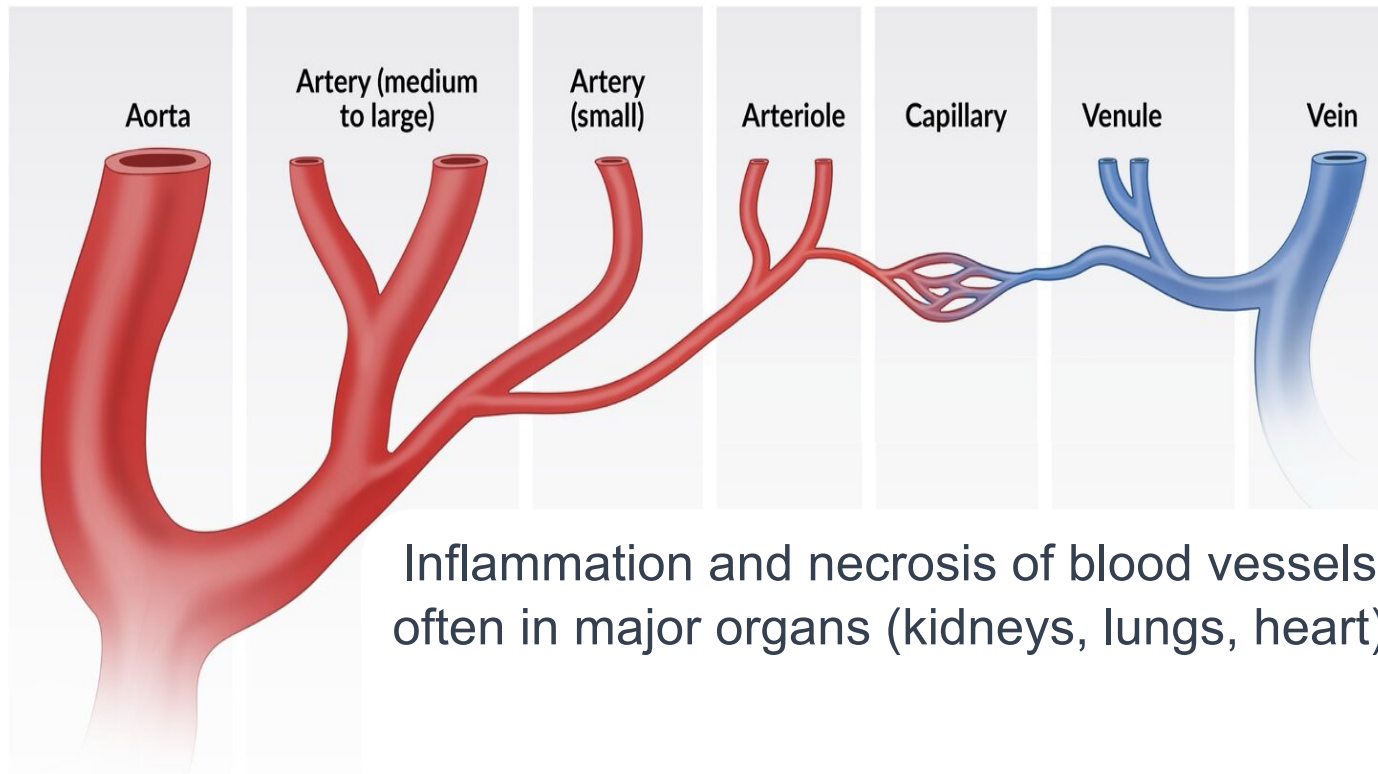


A photograph of a school hallway. On the left, there are red lockers. On the right, there are yellow lockers. The floor is made of large, square tiles in shades of red, orange, and grey, arranged in a checkered pattern. Sunlight streams in from a window on the right, creating bright patches on the floor and lockers. The hallway leads into the distance, where more lockers and a doorway are visible.

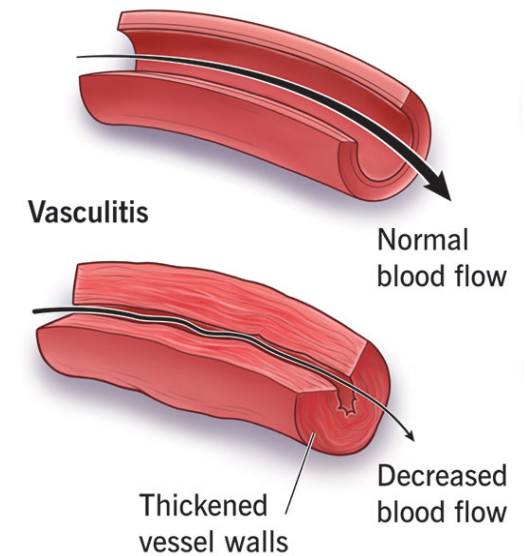
In a Canadian high school of ~1,500 students there might be  
2-3 students with juvenile arthritis  
1 student with lupus  
1 student in every 4 schools with a periodic fever syndrome  
1 student - at most - in every 4 schools with vasculitis



# What is Vasculitis?



Healthy blood vessel



For a review of 2022 ACR/EULAR classification criteria, see Koster MJ and Warrington KJ Nat Rev Rheumatol 2022 18:309-310  
Modified images from the Cleveland Clinic

# Pediatric vasculitis – the beginning

*1950s*

First reported case in a child.

*1970*

15 patients treated with cyclophosphamide.  
12 (80%) achieved remission.

Fauci et al. Medicine 1973 52:533-561

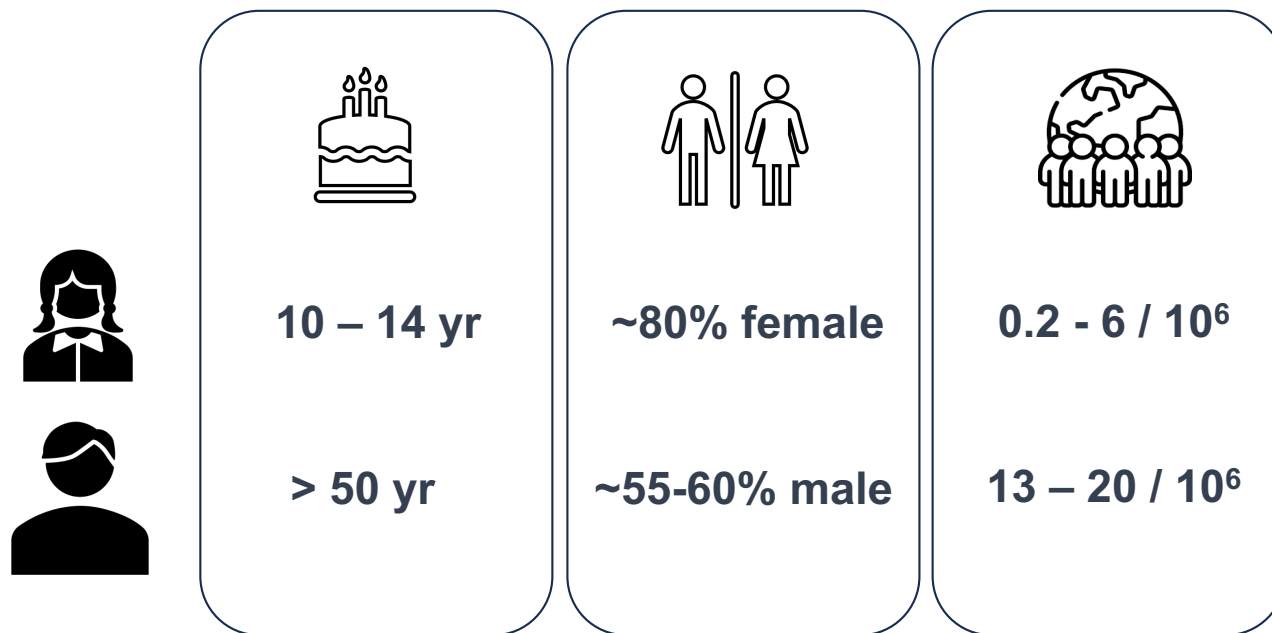
*1990s*

150 patients (23 children) treated with cyclophosphamide.  
89% achieved remission, but at a high cost with 86% experiencing morbidity.

Hoffan et al Ann Intern Med 1992 116:488-498.



# Childhood *vs* Adult onset Vasculitis



Pediatric: Annual incidence from Geetha D et al Am J Kidney Dis 2020 75(1):124–37; **Sacri A-S** et al Nephrol Dial Transplant 2015 30(Suppl 1):i104-112, **Kouri AM** et al Pediatr Nephrol 2017 32(3):449-455, **Morishita KA** et al Arth Rheumatol 2017 69:1470-1479, **Calatroni M** et al Clin J Am Soc Nephrol 2021 16:1043–1051, **Yang J** et al Front Immunol 2022 13:857813, **Marlais M** et al Am J Kidney Dis 2023 18:119-122, **Tan L-W** et al World J Pediatrics 2024 20:506-516, **Toor KK** et al Arth Rheumatol 2025 77:606-614

# Pediatric Vasculitis Initiative



CIHR IRSC



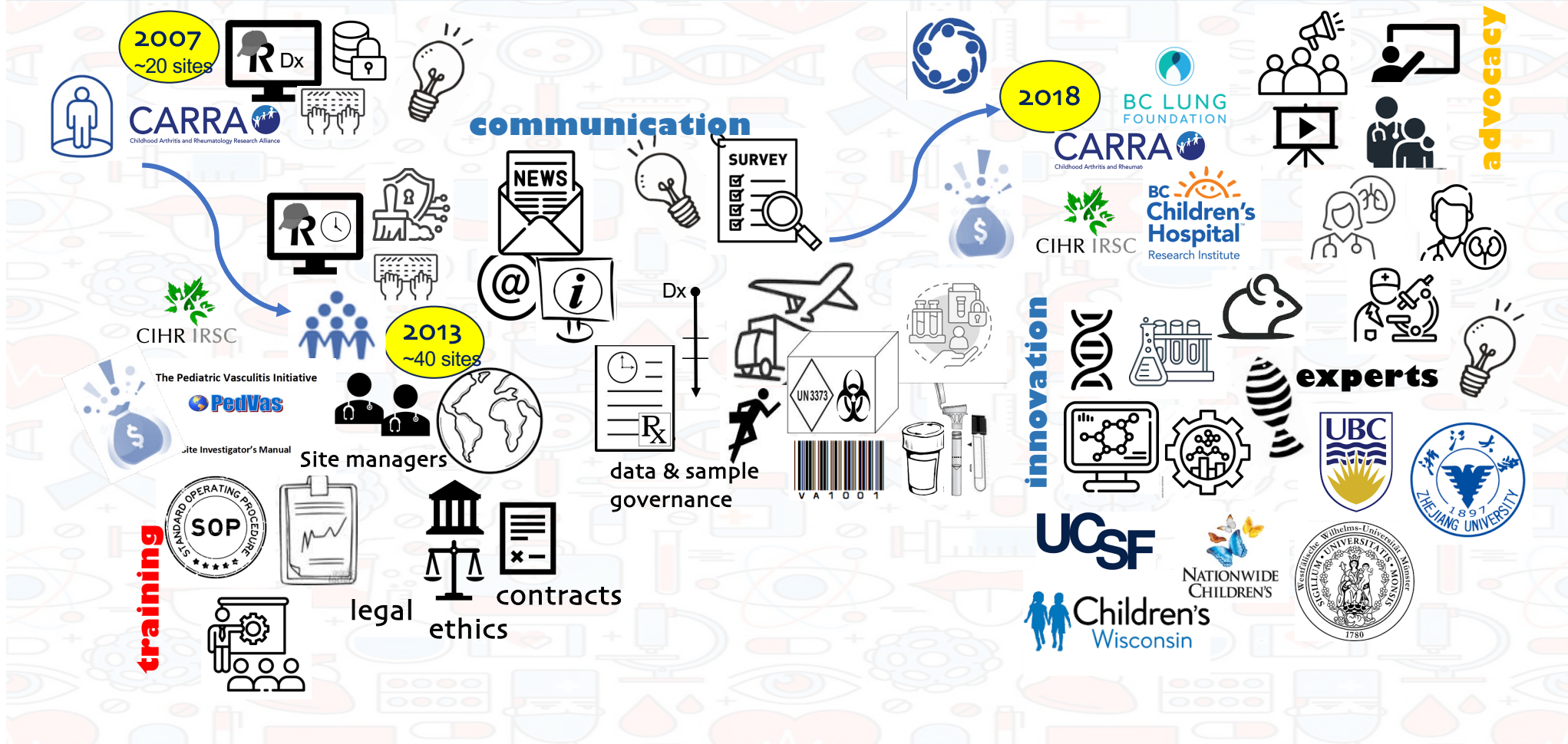
Dr. David Cabral



Dr. Kim Morishita



# Pediatric Vasculitis Initiative



# World's Largest Pediatric Vasculitis Resource

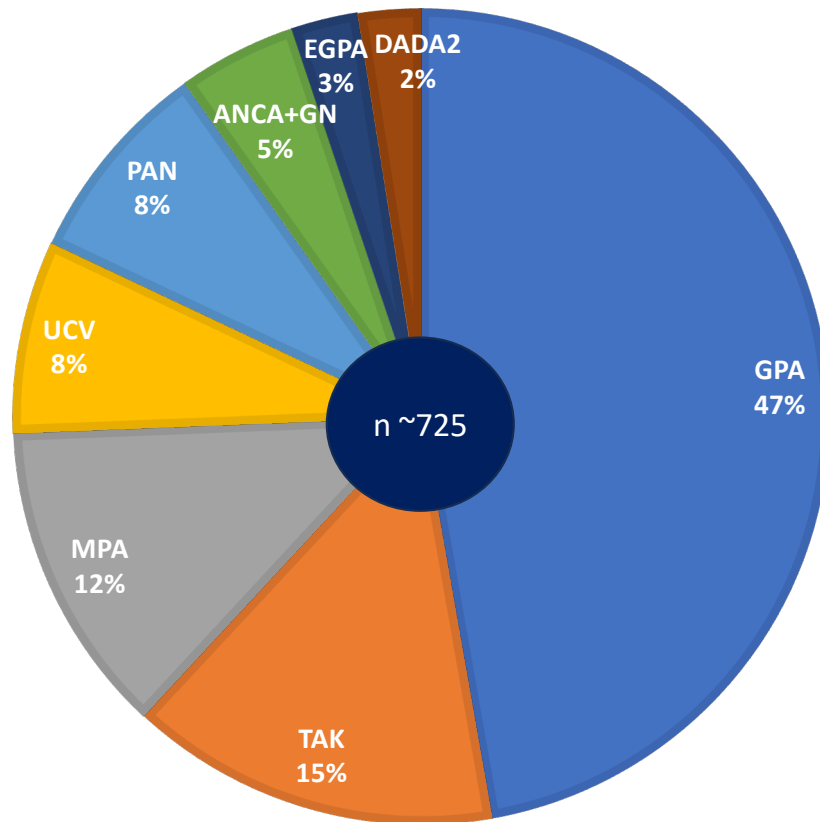


Image from BC Children's Hospital Biobank [www.bcchr.ca/biobank](http://www.bcchr.ca/biobank)