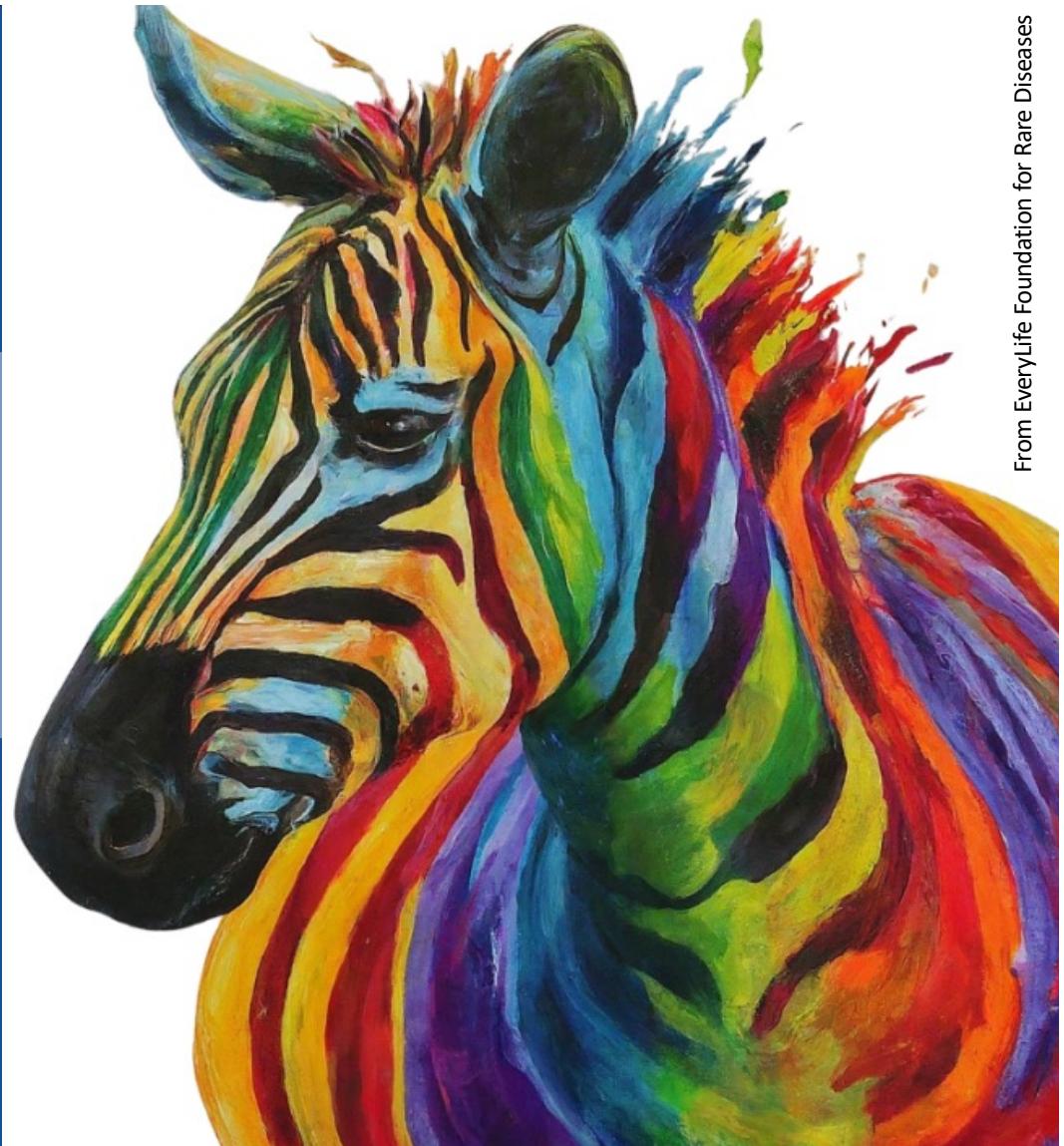


## Story #2

### Hide and Seek: finding the rare within the ultra-rare!



# Deficiency of adenosine deaminase 2 (DADA2)

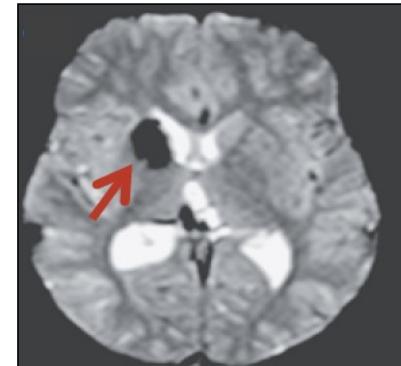
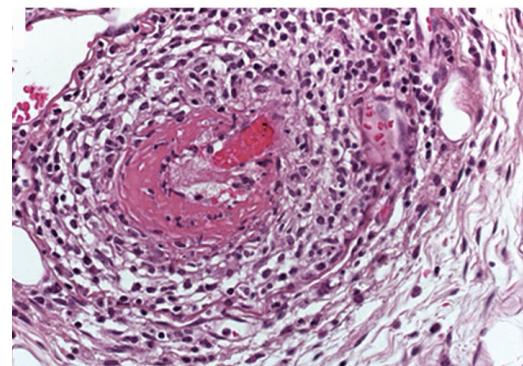
The NEW ENGLAND JOURNAL of MEDICINE

The NEW ENGLAND JOURNAL of MEDICINE

ORIGINAL ARTICLE

Elkan et al. NEJM 370 921-931 (2014)

Mutant Adenosine Deaminase 2  
in a Polyarteritis Nodosa Vasculopathy



ORIGINAL ARTICLE

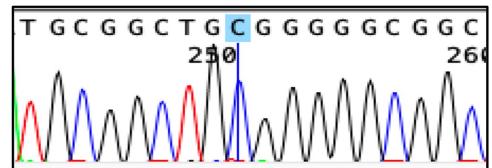
Zhou et al. NEJM 370 911-920 (2014)

Early-Onset Stroke and Vasculopathy  
Associated with Mutations in ADA2

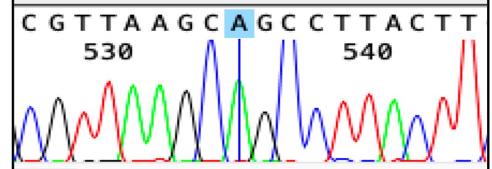
Figures from Zhou et al. NEJM 2014

# DADA2 in the Pediatric Vasculitis Initiative

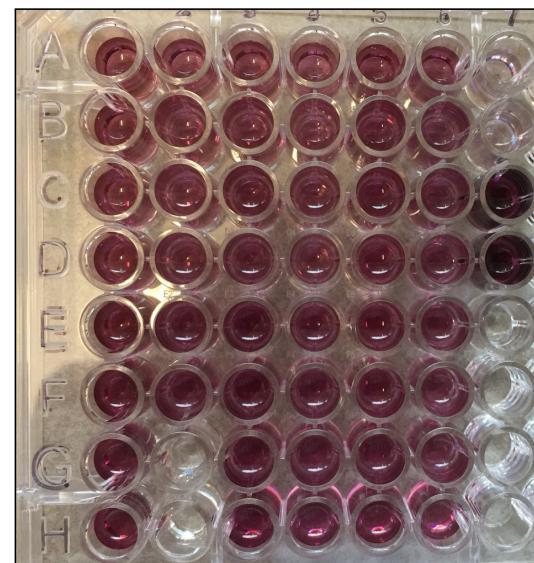
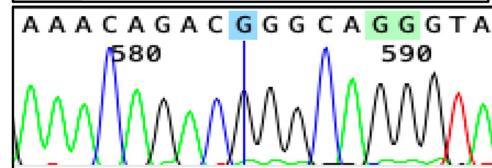
c.139G>C  
(p.G47R)



c.1052T>A  
(p.L351Q)



c.1084T>G  
(p.W362G)

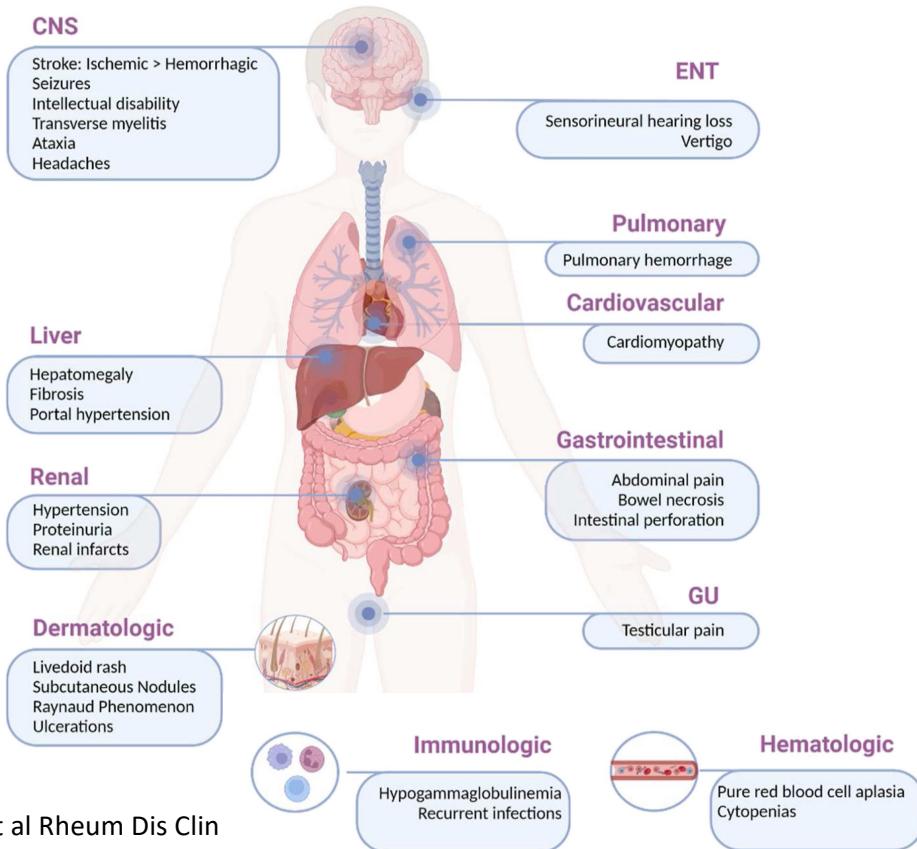


9 patients with  
DADA2!

DADA2 is best  
treated with a  
specific biologic  
(anti-TNF) or  
HSCT

Gibson et al. Arthr Rheumat 2019 71:1747-55  
Bowers et al. Ped Rheumatol. 2020 18:54

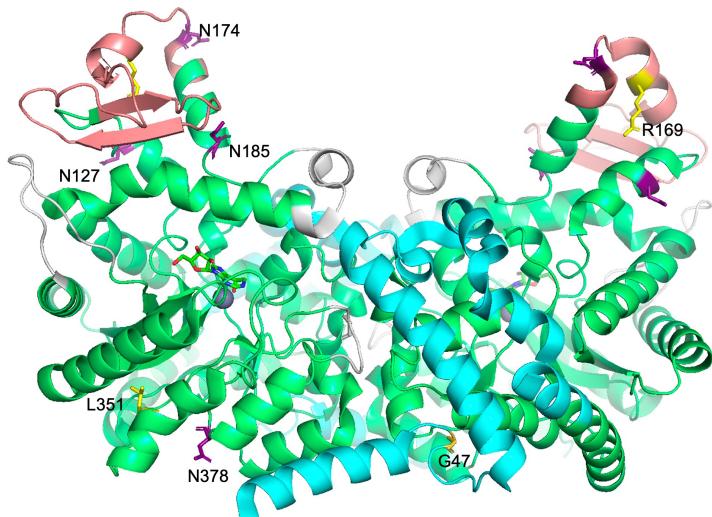
# Beyond vasculitis



Gram et al *Rheum Dis Clin*  
2023 49:773-787

- ~600 patients
- 165+ mutations in the *ADA2* gene
- ~25% of cases diagnosed in first year of life and 77% before 10 years of age
- explains ~25% of cases of 'atypical' vasculitis in medium vessels
- autoinflammatory disease

# Ongoing research



# Advocacy



## More Discoveries in ADA2

A Conversation with

**Kelly L. Brown, Ph.D., Principal Investigator**  
**Sarah M. Bowers, M.Sc., Ph.D.candidate**

University of British Columbia & BC Children's Hospital Research Institute  
Vancouver, Canada

**dada2**  
FOUNDATION  
[www.dada2.org](http://www.dada2.org)

**cassie+  
friends**



My Journey with Rare Disease

With August being Auto-inflammatory Awareness Month, we connected with Nicole Pereira to share her journey with a very rare form of Vasculitis called Adenosine Deaminase 2 Deficiency. After wondering why her legs were covered in bruises from simply watching TV, Nicole realized something was seriously wrong. Through the challenges, she's learned to curate a positive mindset and use her experiences as tools to achieve her goals. Read her powerful words below. I suggest choosing the window seat when you book a flight. Just before an airplane makes its descent into what we all hope [...]



**Brown Lab – Research Grade Testing**  
ADA2 enzyme activity and gene sequencing

# Understanding Vasculitis

## Essential Information for Educators

### What is Vasculitis?

Vasculitis is a general term that refers to **INFLAMMATION OF THE BLOOD VESSELS**. It describes a family of more than 20 rare autoimmune diseases, characterized by narrowing, weakening or scarring of the blood vessels, which can restrict blood flow and damage tissues and vital organs such as the lungs, kidneys, heart, and brain. With treatment, it can be well managed, but there is currently no cure.

Vasculitis symptoms can vary from person to person depending on the organs and tissues affected but may include:

- Fatigue/weakness
- Muscle and/or joint pain
- Lack of appetite/weight loss
- Rashes or skin lesions
- Eye pain and redness/blurred vision
- Chronic nasal, ear and/or sinus problems
- Cough & shortness of breath
- Abdominal pain
- Severe headaches
- Nerve problems (numbness, weakness, pain)



**TIP** Your student and their parents are likely feeling overwhelmed. Help connect them with the people in your school and education system who can help.

Microsoft Pow



### A Special Note for Coaches and Physical Education Instructors

Vasculitis causes extreme fatigue and negatively impacts strength and endurance. Vasculitis treatments can weaken bones and muscles. As a result, a child with vasculitis may need to avoid or modify certain types of physical activity. However, children with vasculitis need to maintain as much physical activity as possible to help lessen bone and muscle loss, preserve strength and cardiovascular endurance, and enjoy the physical and mental health benefits of being active.

It can be very hard for a child who has excelled in athletics to suddenly find themselves unable to compete and perform at their previous level. Remind them how incredibly hard their body is working to fight this disease. Help them to see that, for now, more of their body's energy and other resources need to be devoted to healing. Reassure them that in time they will have more energy for their athletic pursuits.



### How You Can Help

1. Suggest physical activities they can enjoy even with limited strength and stamina.
2. Find ways to allow them to safely participate.
3. Provide non-physical ways for them to be a leader and contribute to the team.

### RESEARCH ARTICLE

Open Access



## Clinical practice variation and need for pediatric-specific treatment guidelines among rheumatologists caring for children with ANCA-associated vasculitis: an international clinician survey

Clara Westwell-Roper<sup>1</sup>, Joanna M. Lubieniecka<sup>2</sup>, Kelly L. Brown<sup>1</sup>, Kimberly A. Morishita<sup>1</sup>, Cherry Mammen<sup>1</sup>, Linda Wagner-Weiner<sup>3</sup>, Eric Yen<sup>4</sup>, Suzanne C. Li<sup>5</sup>, Kathleen M. O'Neill<sup>6</sup>, Sivia K. Lapidus<sup>7</sup>, Paul Brogan<sup>8</sup>, Rolando Cimaz<sup>9</sup>, David A. Cabral<sup>1\*</sup> and for ARChiVe Investigators Network within the PedVas initiative



Cassieandfriends.org

ABOUT US GET INVOLVED

CLASSRHEUM HUB

Newly Diagnosed

Injection Support

JIA – Juvenile Idiopathic Arthritis

sJIA

Vasculitis is a rare rheumatic disease affecting approximately 1 in one

Uveitis Information

Genetics + Childhood Rheumatic Diseases

What are 'rare' rheumatic diseases?

Gut Microbiome + JIA Connection

Managing Pain

Medications: Methotrexate, Biologics + more!

Mental Health

Nutrition + Rheumatic Disease

Physical Activity

Sleep + JA

School Support + Toolkit

Transition from Pediatric to Adult Care + Beyond



### Vasculitis

Another Autoimmune Rare Rheumatic Disease

David Cabral

Clinical Professor, Head  
Division of Rheumatology  
BC Children's Hospital &  
University of British Columbia

Thank you for  
listening!



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TOGETHER IN DISCOVERY

