Issue 04 – 2024

AUTOINFLAMMATORY MAGAZINE

AWARENESS MONTH

Streamathon, Podcasts & New Storefront

PATIENTS' STORIES

Real stories from patients around the globe

PUBLICATIONS

FMF & AID publication Searching for a Zebra

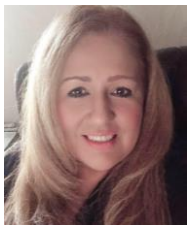
Rhyker with HIDS (USA)

This magazine is presented by the FMF & AID Global Association.

For more information visit fmfandaid.org

IN THIS EDITION

Editorial Team



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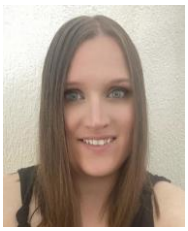

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Dear Readers,

Welcome to the 4th edition of the FMF & AID Magazine! September is finally here, and we are excited to present this year's theme of "No patient left behind", to highlight the many patients who are facing diagnostic challenges and remain without treatment.

FMF & AID has arranged a comprehensive set of educational tools for September 2024 aimed at empowering both the patient community and the general public. They include our publication "Patient Experiences and Challenges in the Management of Autoinflammatory Diseases—Data from the International FMF & AID Global Association Survey", the release of new educational videos, topic-specific brochures on managing autoinflammatory diseases, a podcast series in English, German and French featuring autoinflammatory topics, etc. Collectively, these efforts will ensure that every age patient receives a timely diagnosis and is not be left behind.

A fundraising campaign will be launched to raise money for the FMF & AID medical assistance program and will be undertaken in English and in German. We are also excited to offer new autoinflammatory disease-themed merchandise to support our work.

Patients unfortunately continue to face delayed diagnoses, limited access to necessary treatments, and a lack of able physicians with expertise to manage these rare conditions. Widespread misconceptions and a shortage or non availability of biological medications in many countries further compound these issues. This September, we urge all stakeholders to come together to address these global challenges.

FMF & AID is excited to offer all age patients and their families some fun products through our new storefront. All funds raised will go to support our various projects.

As a mother of an autoinflammatory child and as a patient myself, I continue to believe that FMF & AID plays a vital role in the lives of patients we are able to assist in fighting for diagnosis and access to treatment. I appreciate everyone, who donates to support our global medical assistance program.

Our team looks forward to continuing work on our many initiatives and we appreciate everyone for helping us make this 2024 September awareness campaign visible and successful.

Malena Vetterli
Founder & Executive Director

2024 FMF & AID Fundraiser

FMF & AID is again hosting our Streamathon hosted by Chris Walker on September 13-17, 2024. This Streamathon is available here.

Chris hosts this event twice yearly with the other fundraiser held in February for Rare Disease Day. We encourage everyone to participate and know that all donations raised will be used directly for the FMF & AID Medical Assistance Program.

Photo credit: Chris Walker

Meet Chris Walker – Our fundraiser host

Chris is our dedicated Fundraising Officer and Community Outreach, for the USA at FMF & AID. As a patient himself, Chris has battled Familial Mediterranean Fever (FMF) from a young age. Growing up without any form of support made his journey particularly challenging, but the experience fueled his passion for helping others.

As an adult, he is determined to ensure that no child faces the same isolation and hardship he endured. This determination led him to join the FMF & AID team, where he plays a crucial role in fundraising efforts. Chris knows firsthand the importance of having access to both medical and psychological support and is committed to making these resources available to other autoinflammatory patients.

One of Chris's most impactful contributions are his biannual Streamathon events. The funds raised, along with other donations, allow FMF & AID to provide essential support for children and adults with autoinflammatory diseases worldwide.

Through his tireless efforts, Chris not only raises funds but also raises awareness, building a stronger, more connected community so that no patient faces these rare disease challenges alone.

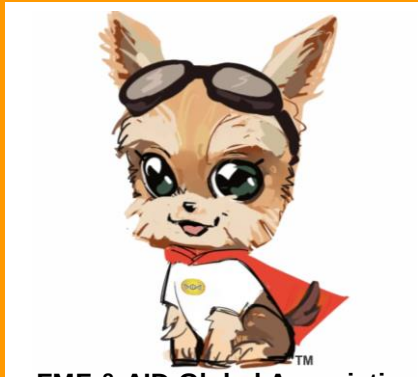
His work is a testament to the power of resilience and represents the difference that one person can make, when driven by empathy and a desire to create positive change.

Chris Walker

FMF & AID's SuperDog Mascot JJ

"My Supportive **Start"**

After I was born, I went to live with an awesome and loving family who treated me with super special care. They blessed me with fantastic toys, beautiful sweaters, and even special dog brothers. However, in the shadow of living in a beautiful setting with the best outside garden to play in, my human sibling Marky was often ill with fevers, joint issues and severe GI pain. While it was so hard to see him suffer, I knew he needed me by his side, which always helped him feel better and be less scared of his life-long impacting symptoms. My presence nearby always reassured him with my woof healing powers, that he would recover soon. Since we also loved to go on adventures and travel when he was well, we became inseparable partners in both sickness and in good health.



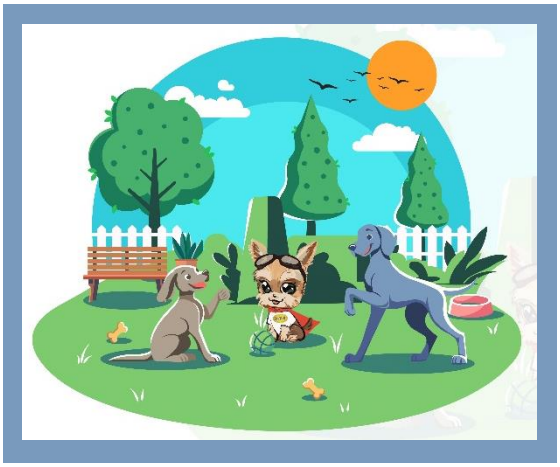
FMF & AID Global Association

"Trading My Sweater for a SuperDog **Cape"**

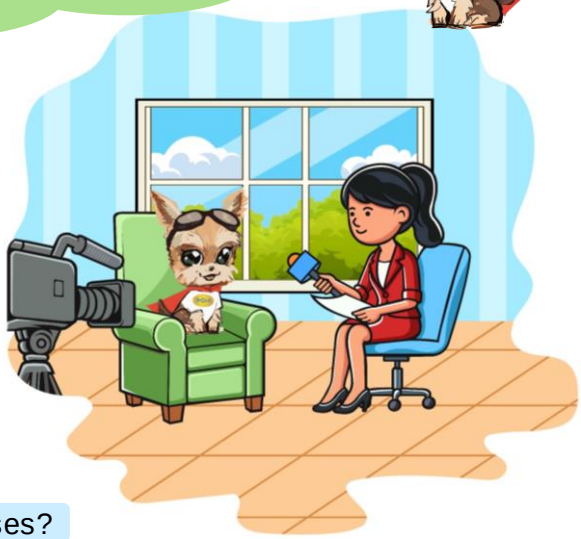
FMF & AID invited me several years ago to join the free therapy sessions offered to autoinflammatory children every month, as they knew how much I helped Marky. During the sessions with our fabulous therapist, we play games, make drawings, read stories and talk about the challenges living with autoinflammatory diseases. Since I love supporting these special rare disease children, who are like my Marky, I was asked by FMF & AID to trade in my sweater for a cape. A big honor for my tiny dog self! I am honored and excited to become the official support SuperDog mascot to ensure that important autoinflammatory awareness is raised globally.

"Other SuperDog **Friends"**

A great thing about my newfound work is that I have been able to help FMF & AID place service animals where other children, like Marky, received a support dog of their own to help them manage their disease with a four-legged friend. Last year some of my dog friends were chosen to go to amazing autoinflammatory families where their woof magic helps daily. Visit our website and see those stories in the FMF & AID 2023 e-magazine edition.



Interview with FMF & AID SuperDog Mascot JJ



JJ, how can you define in simple words what are autoinflammatory diseases and how are they different from autoimmune diseases?

Autoinflammatory diseases are a result of the innate immune system overreacting and causing hyper-inflammation that does not turn off - kind of like a leaky faucet. Most of these diseases are caused by genetic mutations. Autoimmune diseases are a result of the adaptive immune system mistakenly attacking itself.

JJ, what are the most common autoinflammatory diseases?

The most common inherited autoinflammatory disease is Familial Mediterranean Fever, and PFAPA is the most common non-inherited condition. Other diseases include CAPS, TRAPS, HIDS, etc.

JJ, what are the symptoms of autoinflammatory diseases?

The symptoms are fever, GI and abdominal pain, rash/urticaria, joint swelling, leg pain, headaches, mouth ulcers, eye problems, pericarditis, fatigue, etc.

JJ, what types of doctors diagnose autoinflammatory diseases?

Rheumatologists, immunologists, infectious disease, and any other doctor who has knowledge of autoinflammatory diseases.

JJ, where are your support services offered?

I provide support to patients globally. I am a multilingual dog and speak English, German, Spanish, French, Italian, and Portuguese.

Dr. Sharpie

JJ, do any of your dog friends have an autoinflammatory disease?

My dog friend Dr. Sharpie (aka SharPei) suffers from Familial Shar-Pei Fever and has symptoms similar to our human friends with Familial Mediterranean Fever (FMF).

JJ, how are you raising funds to support the FMF & AID medical program?

Because and I am the most photogenic mascot, I'm available on t-shirts, mugs, and other items, to provide woof support to everyone. Please keep reading the e-magazine to find me in the new FMF & AID storefront.

JJ, where can the general public find more information on autoinflammatory diseases?

I highly recommend people to visit the FMF & AID website.

Exciting News: Our New FMF & AID Merchandise Storefront is Here!

We are thrilled to announce the launch of our official storefront, a new and exciting way for everyone to support the FMF & AID Global Association. Our shop will offer a wide range of products including t-shirts, mugs, cups, magnets, stickers, and more. Each item is a symbol of hope and solidarity in our fight against autoinflammatory diseases. We also feature our SuperDog Mascot JJ.

Shop with Purpose

When you shop at our storefront, you are making a difference as every item sold helps raise vital funds for our medical assistance program. This ensures that patients in need receive the care and support for their medical condition.

Our Products: More than Just Merchandise

Our products are designed exclusively to spread awareness and positive messaging for our autoinflammatory community.

Our designs reflect the resilience of our patients from our youngest to our mature members. Whether it is a comfortable t-shirt that you can wear with pride, a sturdy mug for your morning coffee, or a colorful sticker to decorate your laptop, each item is a testament to the strength of those living with autoinflammatory diseases and the dedication of those who support them.

Get Involved and Spread the Word

We invite you to visit our new storefront and explore the variety of products offered. By purchasing these items, you are directly contributing to our cause. Share the love by gifting these items to friends & family and encourage them to join the movement as well.

Thank You for Your Support

As always, we are deeply grateful for your continued support. Together, we are making a tangible difference in the lives of those affected by autoinflammatory diseases. Let's keep the momentum going—shop our storefront today and help us continue our mission of raising awareness and providing essential medical assistance.

We can't wait to see our community proudly displaying these symbols of hope! To the shop.

FMF & AID Publication

FMF & AID is proud to report that our paper Patient Experiences and Challenges in the Management of Autoinflammatory Diseases—Data from the International FMF & AID Global Association Survey was published in the Journal of Clinical Medicine in 2024.

This paper would not have been possible without the collaboration and effort with our fabulous group of expert authors: Jürgen Rech, Georg Schett, Abdurrahman Tufan, Jasmin B. Kümmerle-Deschner, Seza Özen, Koray Tascilar, Leonie Geck, Tobias Krickau, Tatjana Welzel and Marcus Kühn.

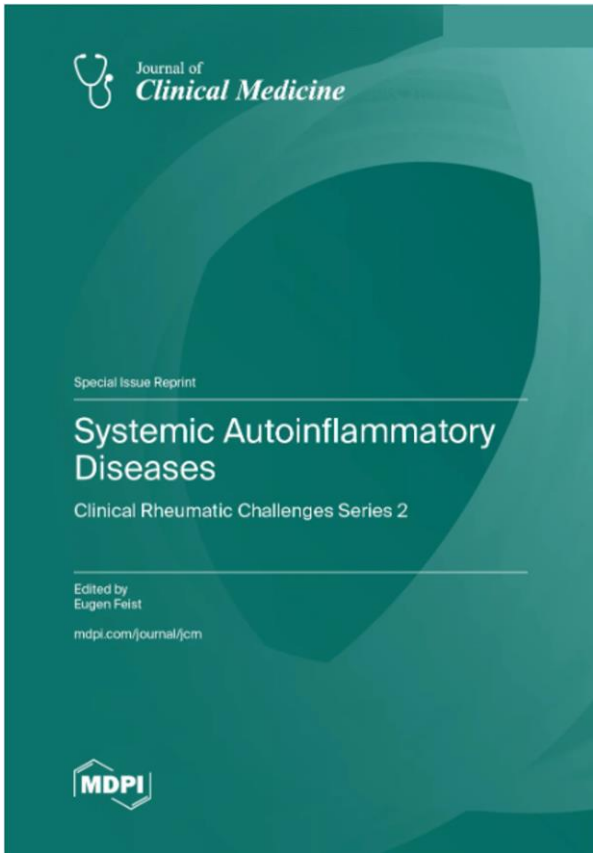
We are also grateful to all the patients who participated in the survey. The information collected allowed for authorship to recognize important issues faced by all age patients.

FMF & AID's paper concluded that autoinflammatory patients, particularly adults, suffer from significant delays in diagnosis, misdiagnosis, and a variety of symptoms, including pain and fatigue. Based on the results presented, raising awareness of these diseases in the wider medical community is crucial to improving patient care and quality of life.

The article, which is part of a special issue on Systemic Autoinflammatory Diseases —Clinical Rheumatic Challenges Series 2, has been very well received with the highest number of views, currently at 5,491.

Abstract

Background: Autoinflammatory diseases (AIDs) are rare, mostly genetic diseases that affect the innate immune system and are associated with inflammatory symptoms. Both paediatric and adult patients face daily challenges related to their disease, diagnosis and treatment. For this reason, a survey was developed in collaboration between the FMF & AID Global Association and the Erlangen Center for Periodic Systemic Autoinflammatory Diseases. Methods: The aim of the survey was to collect the personal assessment of affected patients with regard to their current status in terms of diagnostic timeframes, the interpretation of genetic tests, the number of misdiagnoses, and pain and fatigue despite treatment.



cont. FMF & AID Publication

Results: In total, data from 1043 AID patients (829 adults and 214 children/adolescents) from 52 countries were collected and analyzed. Familial Mediterranean fever (FMF) (521/50%) and Behçet's disease (311/30%) were the most frequently reported diseases. The average time to diagnosis was 3 years for children/adolescents and 14 years for adults. Prior to the diagnosis of autoinflammatory disease, patients received several misdiagnoses, including psychosomatic disorders. The vast majority of patients reported that genetic testing was available (92%), but only 69% were tested. A total of 217 patients reported that no increase in acute-phase reactants was detected during their disease episodes. The intensity of pain and fatigue was measured in AID patients and found to be high. A total of 88% of respondents received treatment, while 8% reported no treatment. Conclusions: AID patients, particularly adults, suffer from significant delays in diagnosis, misdiagnosis, and a variety of symptoms, including pain and fatigue. Based on the results presented, raising awareness of these diseases in the wider medical community is crucial to improving patient care and quality of life.

Key findings are summarized:

- The average timeframe for diagnosis: 3 years in children/adolescents and 14 years for adults.
- 20% of patients reported no elevation of acute-phase reactants found during flare-ups.
- Adults with autoinflammatory disease present with fewer fever episodes than children or none.
- Misdiagnosed conditions reported included: psychosomatic disorders, fibromyalgia, osteoarthritis, IBS, and asthma.
- Patients were not taken seriously with regard to their symptoms in the following: they were refused appropriate and timely blood testing during flares, encountered physicians unwilling to consult with experts in the field, patient cases were rejected or discharged from doctor care.
- 52% of adult patients reported IL-1 biologics did not provide enough efficacy for QoL, only 34% regained full functionality and 13% reported no improvement with biologic use.
- Despite treatment, 468 out of 1043 respondents reported having a mean pain score of 5+ or above in the last 30 days.
- Only 206 out of 415 FMF patients reported having an improved QoL, 164 had a partial response, while 45 patients had no QoL improvement, despite being treated.
- The location of patients responding suggests that the disease manifests more globally than current medical literature suggests.

Marathoner Steven Scalora runs for FMF & AID

Last October, Steven Scalora, a patient with RARE Familial Mediterranean Fever (FMF) created a fundraising event to support the work undertaken by FMF & AID Global Association. He successfully completed the HARTFORD Marathon on October 14th. He created a 99-day donation drive countdown campaign to ensure that patients, parents, and the broader community had time to share the fundraising campaign across social media.

Steven received his FMF diagnosis and colchicine treatment over 7 years ago, after which he began to focus on his diet and exercise routine. He did so to improve his health, which had deteriorated for several years prior to him being diagnosed. With hard work and dedication, he received IRONMAN status and then went on to have a family with two lovely children. He completed the 26.2-mile (42.1km) HARTFORD race in 3:54:17 and raised $655 for the FMF & AID medical assistance program.

We congratulate Steven's achievement and thank the many donors who sponsored him. He is an inspiration to all autoinflammatory patients, and he wishes everyone good health and his motto is to never give up.

Photo credit: Steven Scalora

Photo credit: Steven Scalora

Zoom support group for children with autoinflammatory diseases by Karin Purugganan

For the fourth year in a row, I have been moderating free psychological support sessions in English to children with autoinflammatory diseases, generously funded by the FMF & AID Global Association.

There are very few things worse than watching your child suffer. When working with parents of chronically ill children, there is an almost ever-present feeling of guilt and worry. This can be compounded by the fact that children with rare genetic disorders are often late receiving a diagnosis and often do not know other children who suffer with the same condition. There is power in connection.

Every month a group of children with autoinflammatory diseases meet over Zoom to make connections with each other. Their locations are global, from Hawaii to the Middle East. It is empowering to have shared experiences with others close to your age. The children's ages range from 4-17 years-old, with the younger participants having a parent close by for technical assistance. The sessions tend to start out the same each month, sharing updates and any news since our last meeting: adopting a pet, enduring a flare, recent theatrical participation, or who has been to a doctor's appointment or to the hospital. While the kids tend to focus on the positive, we do not sugarcoat the fact that bumps in the road occur. I often find that clinically, children are more adept at these fluctuations compared to their parents. There is nothing more rewarding than watching the growth of these young warriors. Kids are really quite resilient!

Another part of the session includes a mini lesson from which topics are often provided by the parents or feedback from the group members. There are some standard topics that are covered a few times a year such as returning to school (always tricky with acknowledging some of the limitations within a school building), handling a disease flare, and my favorite lesson I like to call "becoming an expert of my own body." This lesson is always a hit with the kids because they know so much about their own body, but often in clinic or hospital settings they do not feel like they have the autonomy to express specific requests. Things like shots and blood draws will be a constant in their lives and giving them age-appropriate information about how to handle these procedures is imperative.

cont. Zoom support group for children with autoinflammatory diseases

Frequently the group will discuss members who have recently been in the hospital, lost a pet, latest travel adventure, fun video games, or had a birthday! The connections that have developed over the last few years are palpable. Every year the group makes a card for one group member who unfortunately has to miss the November/December sessions because she is performing in The Nutcracker. Recently, we made a lovely card for a group member who had lost their beloved dog. The kids are quite sweet sharing their lovely drawings. We use a collective white board, and all the kids have access to write or draw what they like! The finished card is then emailed to the child and their family.

I have run therapeutic groups for children for over 7 years and this is the longest running and most fun! I will leave parents and caregivers with the following advice for managing a child or family member with a chronic illness:

1. Cultivate the ability to recognize your loved one's strengths and struggles. Focusing on one or the other can be unhelpful, but growing your ability to recognize each of these personality traits is important. Share these openly and honestly.
2. Recognize and manage your own guilt. This can be hard because as parents we feel like we should give our children everything. Parents who feel an overwhelming sense of guilt can tend to overcompensate in other areas of their parenting.
3. Teach your loved one or child to self-advocate. This can be as simple as stating that your IV sticks work better in your left arm, asking for ice or freezy spray before an injection, or asking the medical team to request input from them directly prior to any procedures. We want to work with and not against our medical team members.
4. Incorporate a strong self-care regimen for yourself and teach your child or loved one the same! True self-care can look like limiting commitments, meditating and creating a calm space for yourself every day, eliminating negative interactions with others, and using a journal to note moments of gratitude.

Karin Purugganan NCC, LPC, Counselor & therapist with over 20 years of experience working with medically fragile kids and supporting those with rare and chronic diseases. Her private practice, Wonderologie, is located in Alexandra, VA, and serves this population along with children who struggle with ADHD, anxiety and depression. Please contact her at www.wonderologie.com for more information.

Karin Purugganan

New Ambassadors

FMF & AID Global Association is excited to announce the addition of a new ambassador to our team, Maria De Marco.

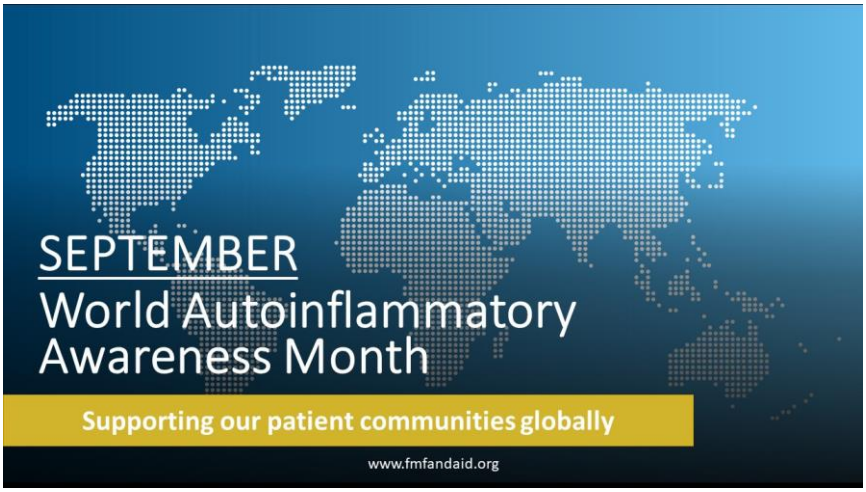
As a patient and advocate, she will be helping us raise awareness about autoinflammatory diseases within the German and Italian speaking communities.

She will share insights into the many facets of these diseases from a patient's perspective and also launch a series of podcasts to delve deeper into these patient-centric topics. We are thrilled to have her support and warmly welcome her as an ambassador for our organization.

FMF & AID Global Association is delighted to introduce our new ambassador, Catherine Pasquier. As both a patient and a passionate advocate, Catherine will play a crucial role in raising awareness about autoinflammatory diseases within the French-speaking communities. She brings a wealth of experience and insight from a patient's perspective, which she will share through a dedicated series of podcasts in French, focusing on the challenges and nuances of living with these conditions. We are pleased to welcome Catherine to our team and look forward to her contributions in amplifying the voices of those affected by autoinflammatory diseases.

Maria De Marco

Catherine Pasquier



Highlighting New Autoinflammatory Diseases by FMF & AID

Several new autoinflammatory diseases have been uncovered due to better genetic reporting globally and an increase in research initiatives searching for new gene causing mutations impacting the innate immune function.

CAPE Syndrome stands for CARD14-Associated Papulosquamous Eruption. The disease is caused by mutations in the CARD14 gene, which is closely associated with causation of psoriasis and familial pityriasis rubra pilaris. The gene also functions as an activator of NF-κB. Patients have clinical features of both disorders, but do not respond to traditional therapies of methotrexate, oral retinoids or TNF-α inhibitors. This can then lead to diagnostic confusion. IL-23 blockers are the appropriate treatments for this genetic disorder.

COPA Syndrome is named for the gene and stands for Coatomer Complex-I. The gene is responsible for transiting molecular cargo from the Golgi complex to the endoplasmic reticulum. The disease crosses categories of autoinflammatory, autoimmunity and immune deficiency. Clinical features include arthritis, renal issues, and interstitial lung disease with possible pulmonary hemorrhage. Treatment is based on symptoms and may include cyclophosphamide or rituximab (during pulmonary exacerbations), systemic corticosteroids, methotrexate, azathioprine, hydroxychloroquine, etanercept, and IVIG.

CRIA Syndrome stands for Cleavage-resistant RIPK1-Induced Autoinflammatory syndrome. The disease is caused by mutations in the RIPK1 gene, which controls the body's response to inflammation and programmed cell death. Clinical features which present from birth include persistent episodic fevers (every 2 to 4 weeks), lymphadenopathy, severe abdominal pains, gastrointestinal problems, diarrhea, headaches, mouth ulcers, tonsillitis, enlarged liver and spleen (hepatomegaly & splenomegaly). Some patients may have joint pain. TNF-α inhibitors are first line treatments.

LAVLI Syndrome stands for Lyn kinase-Associated Vasculopathy and Liver Fibrosis. The disease is caused by mutations in the LYN gene which is a regulator of immune response. Clinical features are variable and may include purpuric skin rash, periorbital erythema, testicular swelling, and hepatosplenomegaly. C-reactive protein, anemia, thrombocytopenia, elevated liver enzymes, leukocytosis, and circulating autoantibodies also present with abnormalities. The disease is seen in infancy/early childhood and treatment includes TNF-α inhibitors and Dasatinib.

LIRSA Syndrome stands for Loss of IL-1R1 Sensitivity to IL-1RA. The disease is caused by a mutation in IL1R1, which causes a defective hydrogen-bond-dependent interaction.

cont. Highlighting New Autoinflammatory Diseases by FMF & AID

This results in over production of IL-1 signaling, which activates NF-κB leading to a dysregulated production of inflammatory cytokines and chemokines. Clinical symptoms include erosive arthritis/osteomyelitis and developmental delay. Treatment used is IL-1 blocker Canakinumab.

PAMI stands for the gene PSTPIP1 (Proline-serine-threonine phosphatase-interactive protein 1)-Associated Myeloid-related proteinemia Inflammatory causing syndrome. The PSTPIP1 gene encodes a cytoskeletal protein, highly expressed in hemopoietic tissues. It functions with several proteins involved in cytoskeletal organization and in the inflammatory processes. Clinical features include: chronic systemic inflammation, cutaneous and osteoarticular manifestations, hepatosplenomegaly, fevers, lymphadenopathy, anemia, growth failure, neutropenia and elevation of zinc and MRP (s100A8). Treatments include corticosteroids, anakinra, cyclosporine A, canakinumab and anti-TNF biologics. Hematopoietic stem cell transplantation has been recently reported to be successful in five patients.

ROSAH Syndrome stands for Retinal dystrophy, Optic nerve edema, Splenomegaly, Anhidrosis, and Headache. The disease is caused by mutations in ALPK1 gene, which is an intracellular pattern-recognition receptor that senses metabolites of various bacteria. The condition has variable phenotypes, and includes clinical features of recurrent fevers, cytopenias, dental and nail abnormalities, Sicca syndrome, and brain meningeal inflammation. To date approximately 60 people have been described with the syndrome worldwide and have been treated with several drugs including anti-IL-1, anti-IL-6 and anti-TNF-α biologics.

SITRAME Syndrome stands for Systemic Inflammatory Truncular Recurrent Acute Macular Eruption. It is believed to be caused by up-regulated interferon-alpha signatures while elevated plasmatic levels of CXCL10 and CCL19 present during flares. The condition is triggered by viral infections or SARS-CoV2 mRNA vaccination. Patients had reductions of flares using the anti-IgE blocker, Omalizumab.

Please contact FMF & AID for any references needed for these newly discovered diseases.

Patient newly diagnosed with PAMI.

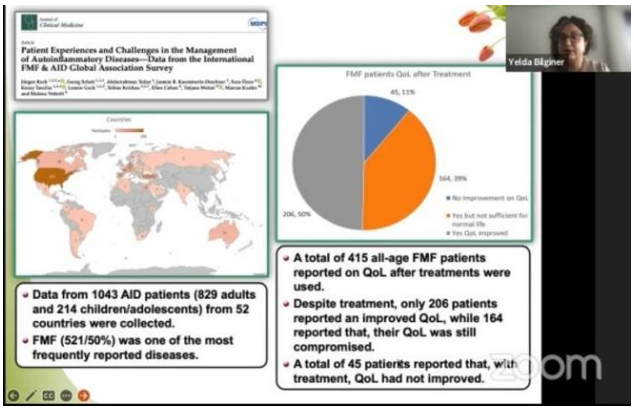
2nd Familial Mediterranean Fever Meeting

Earlier this year, the 2nd Familial Mediterranean Fever Meeting was hosted in Istanbul, Turkey in May 2024 and organized by the Cerrahpaşa Rheumatology Society in Turkey. The event caters to medical professionals, both students and physicians from around the world, who are interested in learning new concepts regarding Familial Mediterranean Fever. FMF & AID Global Association would like to thank the Co-Presidents, Dr. Huri Özdoğan, Turkey and Dr. Eldad Ben Chetrit, Israel for allowing us to virtually attend the meeting.

During the opening sessions, the latest FMF & AID publication Patient Experiences and Challenges in the Management of Autoinflammatory Diseases – Data from the International FMF & AID Global Association Survey, was presented by Dr. Yelda Bilginer, as part of a review of key FMF medical literature from the last year.

Topics of interest included discussions on the inflammasomes' role in pyrin dysregulation, FMF around the world - in Japan, Italy and Saudi Arabia, gender differences in disease presentation, use of various colchicine and IL-1 drug protocols, and transitioning FMF patients from pediatric to adult care.

As patient experts, we appreciate listening to these top experts, to learn about cutting-edge research and other factors relating to Familial Mediterranean Fever. The information discussed will allow us to better guide our FMF patient communities globally.



Dr. Yelda Bilginer's slide show

Photo credit: Cerrahpaşa Rheumatology Society, Turkey

Searching for a Zebra by Monica Hare

When I was almost 40, I found out I was pregnant with baby number five. After the initial shock, I soon settled into the idea. When Myles made his debut a few months later, the love I felt for him was indescribable. He was healthy and strong and was growing faster than I was ready for. He completed our family.

When Myles was three months old, he ran his first fever and his doctor told us, it must be a virus. He soon recovered, but about three weeks later he had another fever. We took him to the pediatrician who looked him over, did swabs and blood work, but nothing was returned as abnormal. Another virus was assumed. This routine pattern continued, as Myles would become hot, feverish, vomit and shake. I would hold him on my chest, watching his little body jerk, feeling his heartbeat race against me, and praying for God to get him through this episode.

After a few days, his fever would subside, and his dad and I would talk about how incredibly healthy he was, despite having had a recent fever of 105°F/40.5°C. It was hard to reconcile our playful child was lifeless only days ago.

We took Myles to so many doctors and ER visits that we lost count. The medical professionals did not know why he was so sick and would instruct us to give him fever reducers (which were ineffective) and to return if he did not improve. I told many of these physicians his fevers often presented in a predictable pattern. Unfortunately, they had no answers, and I did not know where else to turn.

Watching my little boy suffer was one of the worst things I have ever experienced, and knowing he was going to be ill again soon was extremely frustrating as there was nothing, I could do to stop it. Our family prayed--a lot.

We took him to yet another doctor, who asked, “Have you ever heard of PFAPA?” I responded: “P-what-a?” She then described the condition and gave me the most wonderful thing: HOPE. I started researching fever disorders and was amazed how Myles’ symptoms fit with a periodic fever diagnosis. This informed doctor knew about PFAPA as her coworker’s son had the condition. I believe we found her for a reason.

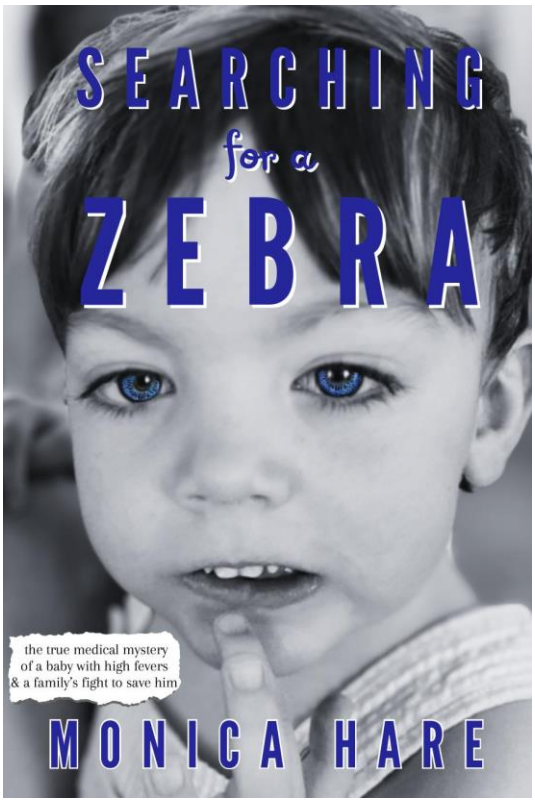


Photo provided by Monica Hare

cont. Searching for a Zebra by Monica Hare

We were then sent to an infectious disease specialist to confirm the suspicion of PFAPA. However, this new doctor, dismissed the idea that Myles had a fever disorder. She said, when you hear hoof beats, it is a horse—not a zebra.

After endless appointments with her and a total of two years of my baby having painful and extreme fever episodes, we finally found a rheumatologist with expertise in autoinflammatory diseases. After the first appointment, she immediately ordered genetic testing, which would confirm our son's FMF diagnosis and provide us with an understanding of our son's condition. She was a hero to Myles and our entire family.

With a diagnosis of Familial Mediterranean Fever, Myles is finally being treated and receives monthly Ilaris injections to control the fever episodes. While some days, I can tell he does not feel his best, and occasionally he will experience breakthrough symptoms, he is now able to play and grow like any typical five-year-old child. I will forever be thankful to the doctor who suggested a PFAPA diagnosis and to the pediatric rheumatologist, who not only listened to our concerns, but provided us with a life-changing diagnosis.

Children and adults all over the world are experiencing what my son went through. I decided I wanted to do something to support those families. While I cannot stop fever disorders, I can raise aware so other patients and families can receive more timely help.

In February of this year, I published Searching for a Zebra about Myles' journey. My hope and prayer is that it will make autoinflammatory disorders more well-known and encourage people to advocate for themselves and their children. God gave Myles and my family the strength to get through one of the hardest experiences of our lives.

FMF & AID Global Association has been a guiding light for me and continues to be there for my family. If we all work together, we will change things for the better for those who are struggling with autoinflammatory disorders.

20% percent of all proceeds from the sale of the book are generously being donated to the FMF & AID Global Association medical fund.

The book can be purchased on Amazon.

Photo provided by Monica Hare

Parent Playbook for Success in the **Doctor's** Office by Monica Hare

Doctor: "He has a cold."

Parent: "What?"

Doctor:"Your son has a cold. I know you would like for me to say he has some rare disease, but when you hear hoofbeats, it's usually a horse--not a zebra."

This is the dialogue between a mother and the specialist who had been seeing her child for several months. Despite the baby presented with 105°F/40.5°C recurring fevers, the doctor had just brushed them off for the last time. Although the child's symptoms and inflammation markers aligned perfectly with a fever disorder (no cold symptoms!), it was clear that the doctor was not going to help her child. Her story, unfortunately, is not unique. Children with autoinflammatory disorders take on average three years to get a diagnosis. So, what is a parent to do?

Respectfully advocate for your child. You are the voice for your child. Tell the doctor why you are concerned. Be specific.

Be prepared. Even if you are very familiar with your physician, doctor visits are stressful. In many cases, you are worried, sometimes intimidated, and often functioning on little to no sleep.

- Write down your questions and concerns. When the doctor rushes in and out, it is easy to forget something that has been on your mind for weeks.
- Bring any pertinent medical records regarding your child's situation on paper and provide to physician (urgent care/emergency visit, etc).
- If you have been keeping a journal or picture diary of your child's fevers and symptoms, bring it with you. (If you haven't, start today!)
- Depending on the age of your child, bring something to keep them busy. These visits are stressful for them, too. A baby can be nursed or cuddled while you talk with the doctor. For toddlers and small children, a LED drawing board or a new figit toy is a great distraction. Older children can watch videos on a phone with headphones.

Educate yourself. It is hard to find medical professionals who are experts in autoinflammatory disorders. To be fair, there are many rare diseases, and as the name implies (rare), doctors may have never seen any cases of FMF or PFAPA compared to more common disease.

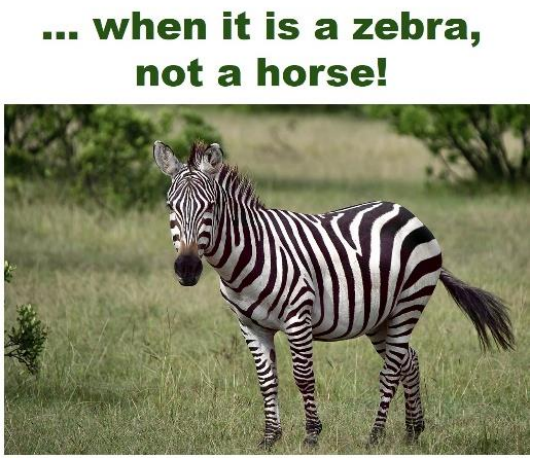


Image by Alp Cem from Pixabay.

cont. Parent Playbook for Success in the **Doctor's** Office by Monica Hare

- Research your child's symptoms.
- If you find diseases that match those symptoms, dive in and learn as much as you can.
- Being educated will not only give you the confidence to speak up, it will help you to have intelligent and productive discussions with your doctor. It may also allow you to understand better, their perspective.
- When your child is diagnosed, you will want to know as much as possible about their disease and treatment options. Your research will allow for better understanding of your child's rare disease.

Say good-bye. Doctors are humans. Some of them are the most caring, self-sacrificing individuals you will ever meet. Their goal in life is to help you, your child, and every other person they encounter who has a medical problem. However, there are others who are not a good fit for their patients or parents. What do you do if a doctor will not listen to your concerns, makes you or your child uncomfortable, or just is not doing enough to help your child's case?

- Leave. Do not beg. Do not raise your voice or use inappropriate language. Do not cause a scene that will make you go viral on social media. Do not stay for months (like many parents do), thinking the doctor will see the light or, more accurately, the test results and symptoms and say, "Hey! I was wrong.
- Your child does have a rare genetic condition!" Just leave. Quickly and politely, and before doing so, get a copy of your child's record.
- There are other doctors available. It may take several appointments, but when you find the one, you will know.
- Throwing a fit will not help your child. It will get you escorted out. It might even get you arrested or reported to child protective services, but it will not help your child get the care they need or deserve.

Ask for help. There are people who will help you find a good doctor. The FMF & AID Global Association can help you navigate the medical system and find help for your child. They have a Facebook page and various support groups in many languages where you can chat with other patients or parents. It is a great resource to find answers and to receive 24/7 support.

Photo by Pavel Danilyuk: on Pexels.com

Buzzy: The innovative medical device for pain reduction in children

“Buzzy” is a child-friendly device shaped like a bee or ladybug designed to reduce pain in all age children or adults when receiving shots. It uses cold and vibration to reduce pain and sensitivity by inhibiting the nervous system.

The applications for the device are diverse as it can be used in vaccinations, blood draws, and medication injections. Buzzy can be used at home or taken to the doctor’s office or hospital due to its compact size and portability. Parents report that Buzzy helped their children during injections by providing distraction and relieving pain.

FMF & AID is happy to report that the organization has provided several autoinflammatory children with a Buzzy. They have all had success using Buzzy to increase their tolerance to anti-IL-1 injections.

Due to the positive feedback, FMF & AID will include their availability through their global medical program in an effort to make shots less traumatic.

Disclaimer: FMF & AID does not receive financial compensation for sale or use of Buzzy.

Photo provided by the parents

Photo provided by the parents

No Patient Left Behind!

PATIENT COLLAGE

Follow-up to **Antonella's** story: a NOMID/CINCA case in Ecuador

FMF & AID continues to provide unique support to a very special patient Antonella. As previously reported in our 2023 magazine, a baby girl from Ecuador, was diagnosed with NOMID/CINCA at the age of 6 months. NOMID/CINCA is the most rare and severe form Cryopyrin Associated Periodic Syndrome (CAPS), and treatment is crucial for survival.

Photo provided by the parents

Despite CAPS being a unique and rare disease (1 in a million), the physician who was initially treating and managing Antonella's case, recognized her disease status, and due to his skills and qualifications, knew to order genetic testing immediately, which led to her diagnosis. Sadly, this wonderful treating physician was removed from his job, after he prescribed the treatment, due to the medication's high cost and unavailability.

Unfortunately, this biological treatment Kineret (Anakinra), used to treat NOMID/CINCA, was not available in Ecuador at the time of her diagnosis. Antonella's parents initiated a campaign to raise awareness in a variety of high-profile ways to bring attention to their daughter's illness, in an effort to encourage the health ministry to authorize and import the drug to Ecuador.

The parents contacted the press to ensure that Antonella's case was made public to the media, as they participated in news interviews, documentaries and other like programming to tell their daughter's medical story.

They also recruited well-known people from the entertainment, music, and sports industries to be ambassadors, who could record and share messages on their social media accounts encouraging government officials to bring this life-saving treatment to save Antonella.

Throughout this family's 2-year effort to save their child, FMF & AID provided continued and unwavering support to the parents and Antonella's medical team. Due to the seriousness of the patient's condition, FMF & AID proposed several solutions including the purchasing of medication abroad, contacting key medical institutions globally for treatment assistance, or relocating patient to another country. These efforts were considered due to the rapid progression of her disease. Unfortunately, due to the regulatory issues in Ecuador, it was impossible to import the medication and relocation of the family was not feasible. After these efforts were realized to be impossible, it was determined the only solution would be for the parents to file a lawsuit against the local government.

cont. Follow-up to **Antonella's** story: a NOMID/CINCA case in Ecuador

Fortunately, they won their case in a rapid timeframe, and the hospital was ordered to purchase the medication. After which, FMF & AID then provided critical information to the medical team regarding medication vendors and multiple bidding channels as required, to secure a drug agreement.

On 2nd April 2024, a groundbreaking official ceremony attended by the first lady of Ecuador, Lavinia Valbonesi, was hosted, where she presented the first box of Kineret to Antonella's parents on behalf of her husband, President Daniel Noboa.

The arrival of this life-saving mediation required coordinated efforts between the inter-institutional management of the Ecuadorian Social Security Institute (IESS), the National Public Contracting Service (Sercop), the National Regulation, Control and Health Surveillance (Arcsa), all of whom worked together to acquire Kineret to treat the first patient with NOMID/CINCA in Ecuador.

First lady, Lavinia Valbonesi, highlighted the coordination of these several institutions to achieve a common objective for the benefit of a minor. "We have all come together through the trenches to help. For me it is an honor that today we are together making history, setting a precedent in Antonella's case and we have shown that we must all join efforts so that more children can have a better quality of life with their families."

Photo provided by the parents
Left side: Antonella with her mother
Right side: First lady of Ecuador, Lavinia Valbonesi

Antonella's father thanked the First Lady, the authorities and the doctors who always supported his family. "The day has finally arrived for my daughter to receive the medication. It is very gratifying. I want to thank you with all my heart. This fight has taken two years to obtain this medication so that my daughter can have a better quality of life."

Photo provided by the parents

FMF & AID is happy to report that Antonella has had an excellent response to Kineret. It has lowered her pain, increased her appetite and growth, helped with her skin rashes, and improved her cognitive function.

Patient journey: Rhyker with HIDS (USA)

Our son Rhyker's first year of life was healthy and amazing but at 13 months, he became ill requiring hospitalization and was sadly diagnosed with Kawasaki's Disease. He was then referred to his first rheumatologist, cardiologist, and infectious disease specialist.

While waiting several months to be seen by the rheumatologist, Rhyker experienced two more lengthy fever episodes, which were different than his previous ones. As a first-time mom, I had no clue what to do, but I did take pictures of his rashes, mouth sores, and swollen joints. Thankfully, my son has an amazing primary care physician (PCP), who helped us in numerous ways. At each visit we would discuss his symptoms, which included fevers of 103.4°F/39.6°C or higher, joint pain and swelling, stomach pain, mouth ulcers, swollen lymph nodes, and skin rashes. Our PCP would order blood work, which would eventually aid in his future diagnosis.

After our first rheumatology/infectious disease appointment, we left discouraged, confused, and wanting more answers. During the visit, we were told about PFAPA, and the doctor said she was sure that this was his diagnosis. Afterwards, I read extensively about PFAPA and believed that Rhyker's symptoms did not match this disease. The doctor prescribed prednisolone and told us to try it at his next fever episode. We did this for the next three months with no positive results.

When we returned for the next appointment, I was prepared, as I had done further reading, had taken photographs and noted all symptoms. I pushed hard for genetic testing, but the doctors refused. I was in tears by the end of the appointment, feeling hopeless and unheard. The doctor decided to prescribe Cimetidine to give with prednisolone. Despite my disagreement with this approach, three more months of fever episodes went by without any symptom resolution on both medications. I wanted answers and if genetic testing would tell us more, I was prepared to do anything possible to get it done.

Photo provided by the parents

cont. Patient journey: Rhyker with HIDS (USA)

I had finally reached my breaking point and his next appointment and told the doctor I was not leaving until we received the paperwork ordering genetic testing.

A week later, the test kit arrived, and I swabbed his mouth and returned the sample. The results were in two weeks later and the doctor reported no abnormalities were found, but that we needed to find a new children's hospital as our needs were out of their expertise.

We were then referred to our second closest children's hospital about two and a half hours away. We made an appointment and were seen promptly. When we walked into the hospital, we were amazed and knew it was going to be a better experience.

When we met with the doctor, he was prepared with all results from Rhykers' genetic testing and labs done with our PCP. He started asking questions and actually listened to our responses and additionally had comparison documents between normal genetic testing and information on MKD/HIDS. During the consultation, we felt heard, understood, and sympathized with. The doctor then stated that Rhyker had an MVK genetic abnormality that left us shocked but not surprised. At this point we were grateful that the hospital was the right choice our son. The next month after that initial appointment we started Ilaris with amazing results and have had only one episode in the last 8 months.

Finding support is an essential need when dealing with a child who has a rare disease.

Thankfully, I found FMF & AID Global Association on Facebook. The support group has opened a whole new community of people going through similar issues as our family. I could not thank them enough for always having a listening ear and allowing us to see other children going through the same thing that we are.

Photos provided by the parents

Patient journey: Chloe with uSAID (Australia)

We live in rural Queensland approximately five hours from Brisbane which makes our journey a bit more difficult when trying to access appropriate paediatric services and treatment.

My daughter, now 11 was diagnosed with Kawasaki disease at 17 months old which was treated on day 5 with IVIG. She had quite significant arthritis following this for the next three months but then made a full recovery until she started experiencing joint and abdominal pains around the age of 3. Her paediatrician could not diagnose any issue. We then sought help with a private rheumatologist, who was not convinced her symptoms were relevant to a medical condition. Luckily, she started to improve temporarily.

Between the ages of 5 and 7, she began having intermittent joint pain, periods of fatigue and was generally unwell. We dismissed them as viral episodes until they became regular with increased intensity. Rashes would appear on her chest, abdomen, arms and legs in the evenings after her shower and her tongue would become ulcerated and get really sore. Episodes of joint pain, swollen lymph nodes, headache and fatigue would then follow but did not fall into a significant pattern for another two years. We reconnected with the private rheumatologist for further review but again, it felt as if he was dismissing her newfound symptoms by labelling them as viral and told us to return in several months for another appointment. It felt as though he was just using our daughter's case as a money-making situation. After this encounter, we began keeping a very specific diary of her symptoms, photographing rashes and monitoring all oral symptoms. We strongly felt her condition was not viral and decided we needed further medical help.

We then got a referral to immunology at our state children's hospital and felt we were finally being taken seriously regarding her case. It took another year to start a trial of steroid medication due to the extended time between appointments and her symptoms not fitting any distinct disease pattern. When we started using the prednisone for flares, it was taking 3 doses before the drug would alleviate the symptoms and additional days to fully recover. At this time, I became very frustrated and decided to investigate her symptoms by googling alternative diseases and joining Facebook groups. We then organized genetic testing through our private paediatrician.

Photo provided by the parents

cont. Patient journey: Chloe with uSAID (Australia)

The results came back with 4 variants of uncertain significance and provided little clue as to what the disease-causing issue might be.

Two years ago, our daughter finally started colchicine but unfortunately, was never able to reach a therapeutic dose before suffering gastrointestinal issues. We then tried increasing the dose in small increments over a long time period, but unfortunately, the same side effects occurred. We then returned to treating the flares with prednisone.

Immunology finally got rheumatology on board this year and have taken over as our main treating team. She started a trial of Anakinra, and while it had limited success, it did not stop her from prolonged flaring and was discontinued.

At the time of writing this update, our child has been hospitalized for 8 days at a state children's hospital that is located 5 hours away from our home. No elevated inflammatory markers ever show up in her blood tests, although she does have some cyclic neutropenia. We are currently in limbo with no standard treatment options left, as Ilaris is not available in Australia. The medical team is not keen to trial expensive biologics due to the negative findings.

It is really hard when the patient does not check one specific box to narrow in on a diagnosis and treatment. Sadly, she has missed 70 days of school this year and we are only halfway through term three.

Overall, she has probably missed 50 days in each of the last school years due to sickness and doctors' appointments. The in-patient stays are hard on my other child who is just 9 and my husband who is left to cope at home own.

Photo provided by the parents

Our daughter is missing out participating in netball and athletic competitions, which are her favourite things to do. She misses her friends but is too young to connect on social media. Living rurally means we are a 15-minute car drive from friends at the best of times.

We understand that autoinflammatory conditions can take a long time to diagnose and treat but it does not always make it easier to stay positive and that a treatment will be found.

I am so grateful for the support of FMF & AID. As a parent it is a lonely journey trying to figure out what is happening when your child is unwell each month and the medical teams brush it off as viral. The support of this organisation has enabled me to advocate for my daughter using real evidence & research to support requests for treatment and formalise a potential diagnosis.

Patient journey: Beni with HIDS (Argentina)

Our son Beni was born with epilepsy and at two months old, he presented with seizures, and shortly after, he began running fevers every two weeks. By the age of six months, he was diagnosed with motor delays and received kinesiological support. At nine months, he developed an encapsulated subdural hematoma that thankfully, did not require surgery. However, his high fevers continued and were now accompanied by multiple swollen lymph nodes, chronic diarrhea, abdominal inflammation, rash-like skin lesions, and recurrent erythematous outbreaks.

At age 3, Beni underwent surgery for appendicitis and during the procedure, the doctor noticed several unusual adhesions, which led our immunologist to suspect he might possibly have an autoinflammatory disease. To confirm his suspicion, the doctor enrolled our son in a free program that conducts genetic studies on children with suspected rare diseases. In February of this year, we were called to the hospital for a blood test, with the understanding that the results may take some time to be returned. Indeed, it took about three months for them to arrive.

Unfortunately, a few weeks later, Beni had to undergo yet another surgery, this time for intestinal obstruction. Once again, firm ties between the jejunum and the mesentery were evident.

The doctor's suspicion of an autoinflammatory disease grew stronger and treatment with colchicine was initiated, to which Beni responded positively.

In April, we finally received a diagnosis: a pathogenic variant in the MVK gene, indicating Hyper IgD Syndrome (HIDS), was confirmed by homozygosity associated with a phenotype compatible with the disease. The severe presentation of HIDS has impacted Beni's growth and there was concern by the medical team, that it could lead to complications such as, amyloidosis, and macrophage activation syndrome. Therefore, the doctors recommended starting treatment with canakinumab, that marked the beginning of our fight to secure the medication.

Beni is the only child in our province with a diagnosis of HIDS. When we first received the news, our world crumbled.

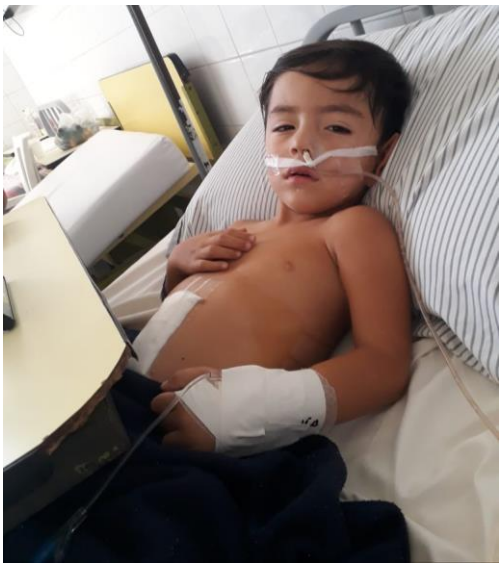


Photo provided by the parents

cont. Patient journey: Beni with HIDS (Argentina)

We were devastated, especially when we learned that the treatment our son needed came with an extraordinarily high monthly cost, far beyond our means. In my desperation, I reached out for help wherever I could, both in and outside the country. Luckily, the FMF & AID Global Association based in Switzerland, heard about our case and contacted us. That is when our outlook began to change as Malena became our pillar of strength, helping us navigate this overwhelming medical situation. She connected us with contacts in Argentina to access the medication and added me to a support group for which we are immensely grateful.

Photo provided by the parents

Today, Beni is four years old and waiting to start treatment. The support from other parents who are also fighting for a better quality of life for their autoinflammatory children has been incredibly important to our family. Knowing that we had the support and backing of an organization like FMF & AID was deeply comforting. We did not feel alone—there were many people concerned about us, and Malena continues to be in constant communication with us, always asking about Beni's health and how the process of obtaining the medication is going. We are profoundly grateful, with all our hearts, for the support FMF & AID has given us. I thank God for bringing this association into our lives and for all the help and support provided.

Photo provided by the parents

Patient journey: Malena with TRAPS (Argentina)

Malena is our 3-year-old daughter who has grown up with recurring monthly fevers. Many times, we would seek help at the local hospital, where she would be continually swabbed for virus or infection but was always negative for illness.

At one point, a kind physician suggested that our daughter's case was seemingly complex and referred her to the Garrahan Children's Hospital in Buenos Aires. Taking her to this hospital was life changing for her and our entire family, despite our fears of what her diagnosis might be.

The team we consulted with, suggested her undergoing a genetic study to determine what might be the cause of her symptoms. After waiting a good amount of time for the results, her rheumatologist finally called us to share that she had a rare autoinflammatory disease called TRAPS Syndrome, which stands for Tumor necrosis factor receptor-associated periodic syndrome, a genetic disease that had no cure.

The doctors then told me, there was a medication that would help have a better quality of life. However, the drug cost millions of Argentinian Pesos. After which, the social system, learning of her diagnosis, turned their back on our case and cancelled us without our consent or signing any paperwork.

There was a final option to receive the medication through our national ministry, but it required our family to give up its income, as my husband was then forced to quit his bank job so that our daughter could receive this life-saving medication.

Additionally, we had little information on this disease and its treatment, so I had many questions.

I searched online and found the FMF & AID Global Association. I was thankful that they supported and helped me to understand, both TRAPS and IL-1 treatment.

Photos provided by the parents

cont. Patient journey: Malena with TRAPS (Argentina)

I no longer felt alone, and it was comforting to know that I had someone to reach out to with all of my questions, whenever I needed answers or further information.

Our daughter, despite her young age, understands that she has a rare disease called TRAPS. When she feels bad, she knows we must go to the hospital to find out where her inflammation is coming from for further investigation and treatment. Even though her doctors have increased the dose of her biological medication, there are times when she is really unwell with breakthrough flares.

Currently, we are awaiting the results of her recent lung biopsy, due to discrepancies on her CT scans.

We will continue fighting for her because even though she has approval for the medication, there is continuous concern that the government will stop providing it to her. We will always advocate for our daughter and do whatever it takes to ensure she keeps receiving her life-impacting treatments.

Thanks again to the FMF & AID for all your support and kindness for our child's case. The assistance has helped us to navigate this extremely difficult journey.

Photos provided by the parents

Patient journey: Wiktor with FMF (Poland)

My son has been struggling with various ailments since the first months of his life. When he was little, he had a recurring rash and fever. Shortness of breath was also a very common symptom.

When he was 3, he developed severe abdominal pain which appeared every few days. Additionally, a lesion was found in his brain stem, which the doctors believed was a vascular malformation. Wiktor was often hospitalized for his flares and unfortunately contracted inflammatory sepsis.

Between the ages of 4 and 8 years old, he was treated at the rheumatology department for severe joint pain. Fluid collected in the joints making it difficult for him to walk. He was also treated by the pulmonology department as fluid accumulated in the pleura. This serious issue made it difficult for him to breathe.

Wiktor's condition worsened, as fluid began to appear in the peritoneum, in his scrotum. My poor son did not have a spot on his body that did not hurt him. This was a very difficult period for our family. Socializing with friends and family became impossible. His immune system was so weakened that we had to give up activities outside of the home for fear of him catching an infection or a virus.

My son was so sick that he could not attend school, so we had to switch to homeschooling. He continued to have fevers and uncontrollable pain throughout his body. The doctors could not figure out what was wrong with him and often accused him of exaggerating his pain. They tried to convince us that his pain was not that severe.

When my son was 10 years old, a doctor from the rheumatology department thought that Wiktor might have Familial Mediterranean Fever (FMF) and sent us to a specialty hospital. We then took Wiktor to the immunology department at the Children's Health Center in Warsaw, where they started the diagnostic process and determined he had FMF. He was then treated with colchicine, and unfortunately, it was not effective to control his symptoms despite using the medication for over a year.

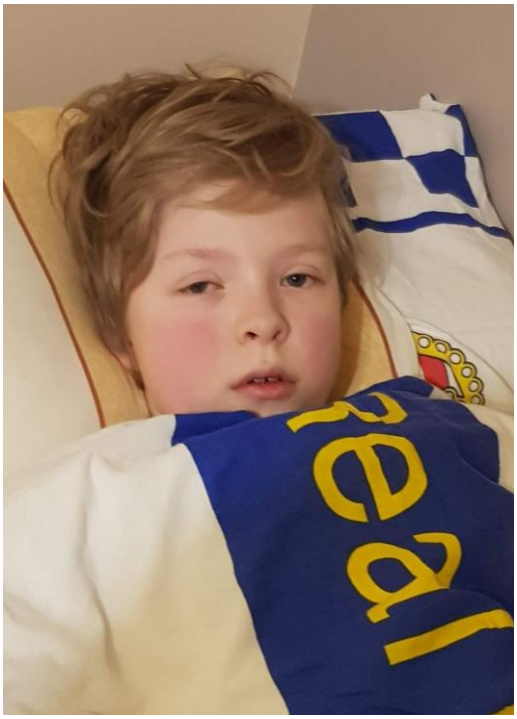


Photo provided by the parents

cont. Patient journey: Wiktor with FMF (Poland)

He continued to have high fevers with longer flares, as fluid would accumulate in his body cavities, and he suffered from severe diarrhea. My son was unable to gain weight and by the age of 11, he only weighed 27kg/59lbs. The hospital was unwilling to offer us any other treatment.

Thanks to the outstanding help of the FMF & AID Global Association, our family learned that it is possible to treat FMF with biological drugs such as Kineret and Ilaris. We are grateful for all the information provided to us and to our son's attending physician. Their support, willingness to help, and caring assistance has been invaluable for our son's case. FMF & AID helped to ensure that Wiktor received lifesaving Kineret. This miracle drug initially reduced his fevers and abdominal pain. He gained weight, grew taller, and is now a well-built young boy.

Unfortunately, the Kineret does not work 100 percent, as Wiktor continues to flare. Although the flares are less frequent, they have become more concerning, as he has had pericarditis four times in the past two years. His major issue is that fluid continues to accumulate in the pericardium often, which limits his physical capability significantly. Wiktor must be on diuretic medications daily and cannot exercise. Thanks to Kineret treatment, he was able to return to school. However, his immune system is weak, and he is often required to stay at home or be hospitalized.

FMF & AID continues to support Wiktor and will be helping us navigate other medical options for our son's case.

Photo provided by the parents

The educational children's e-book "FMF Superheroes" is available in English, German, and Arabic.
This book is highly recommended for children of all ages. You can purchase a copy and in doing so, you will also be supporting the FMF & AID Global Association's work.

Patient journey: Janek with uSAID with PID (Poland)

During our child's first years of life, there were no signs that anything was wrong with his health. However, at age 4, our son Janek was diagnosed with neurological delays and was often sick with infections and abdominal pain episodes. He had fevers with every childhood vaccination given. He was also found to be IgA-immune deficient and frequently suffered from bronchial asthma.

Since November 2021, he has been struggling with recurring fevers, accompanied by a rash on his trunk and limbs, elevated CRP accompanied by neutrophilia, joint pain, swelling, and abdominal issues. Due to these symptoms, he has been cared for by our local children's hospital, but to-date, his physicians have been unable to make a complete diagnosis.

In June 2023, our son qualified for treatment of a congenital autoinflammatory syndrome and Kineret was introduced. Unfortunately, it had little effect, as he continued to have more flares than before. The medication was discontinued, and he was then treated with colchicine and steroids, without any positive results.

Thanks to the FMF & AID Global Association, we have hope for further diagnosis for our son, as they are helping us to find a European specialty center to address his combination of PID and autoinflammatory disease. Sadly, in Poland, his doctors have informed us that further diagnostics and treatment are not possible. The support from FMF & AID through these difficult times plus their financial aid enabling us to travel will hopefully help us uncover his disease and appropriate treatment protocol.

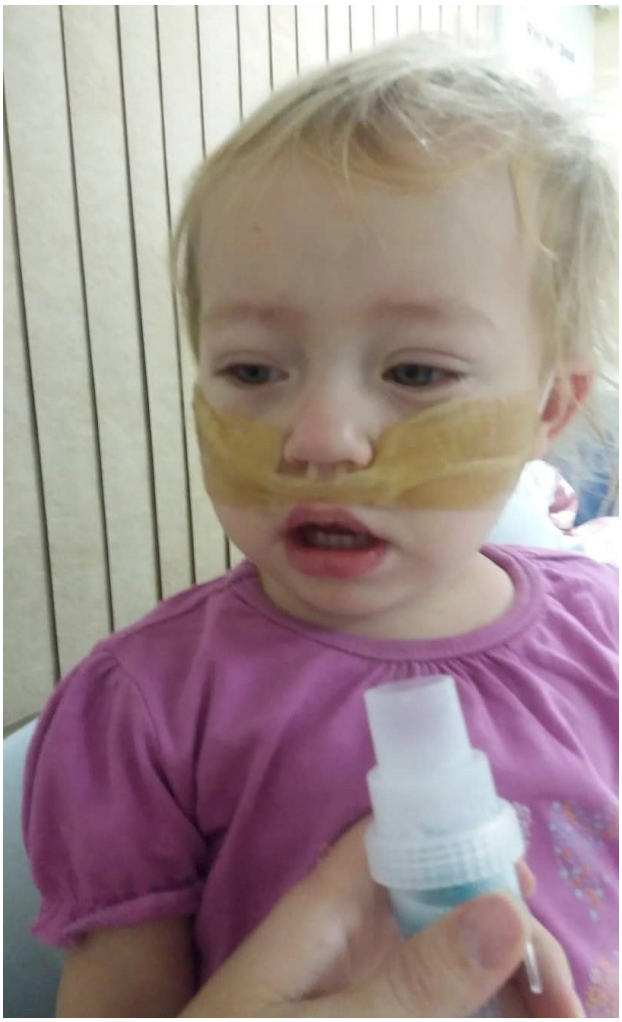
Along with FMF & AID support, our family is currently raising funds to help with our medical costs and future trips. My family and I bake cakes, sweet treats, dumplings and make preserves, to sell at various outdoor markets. We also have a Facebook group where we auction off donated items to support this initiative. We greatly appreciate all the help received from family and friends. It is wonderful for our family to know that we are not alone and thanks to the FMF & AID organization, we know there is future hope to save our child.

Photos provided by the parents

Photo provided by the parents

Patient journey: Family with uSAID and PID (South Africa)

I am a mother of two children with an undifferentiated systemic autoinflammatory disease (uSAID), as well as primary immunodeficiency (PID), and I am also affected by these conditions. My children, Shaun and Elisha, have been sick since birth, but their symptoms have manifested differently.



Shaun's first hospital admission was at two weeks old due to his high fevers. My daughter, Elisha, did not require hospitalization until she was four months old. She did not have a fever but cried continuously until she turned blue, suffering from severe gastrointestinal issues.

During flares, their inflammatory markers are often, but not always, elevated. However, their platelet counts are consistently high, both during and outside of flares. Febrile episodes, accompanied by joint pain, typically last 3 to 5 days for Elisha and up to 12 days for Shaun, often with respiratory symptoms.

Both children endure recurrent ear infections, bronchitis, pneumonia, high fevers, headaches, abdominal pain, bloating, joint pain, sore throats, mouth ulcers, swollen lymph nodes, blood in their stool, and painful red eyes. They are also anemic. Their flares can be triggered by vaccinations, physical exertion (like sports), and cold temperatures.

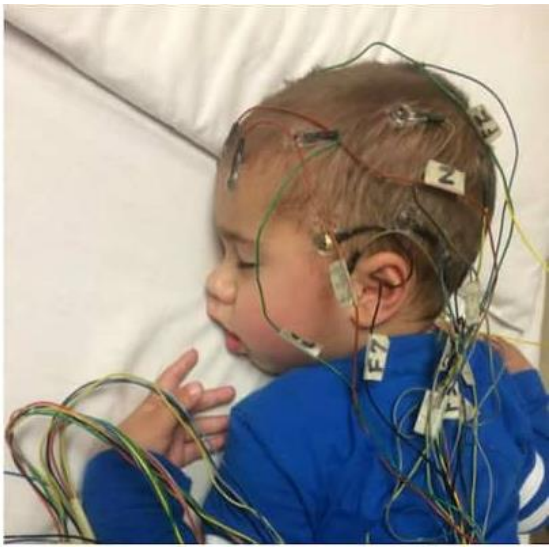
Photos provided by the parents

Patient journey: Family with uSAID and PID (South Africa)

Today, my daughter is 7 years old, and my son is 10. Unfortunately, it has not been an easy journey for my family. Over the years, we have seen countless doctors in our search for answers. The challenge is that South Africa lacks rheumatologists with specialized knowledge in autoinflammatory diseases.

It was not until recently that we found a specialist, but they were in another city, which meant travel and accommodation expenses for our whole family. FMF & AID, aware of our situation, generously stepped in to help. They covered the costs of a consultation with a specialist abroad, and flights for our entire family to see a specialist in Cape Town.

I do not know what we would have done without their support. We feel incredibly fortunate to have FMF & AID in our lives and will forever be grateful for their support, which has made us feel like we are a part of a community, despite the rarity of our conditions.



Photos provided by the parents

Patient journey: Family with PID (South Africa)

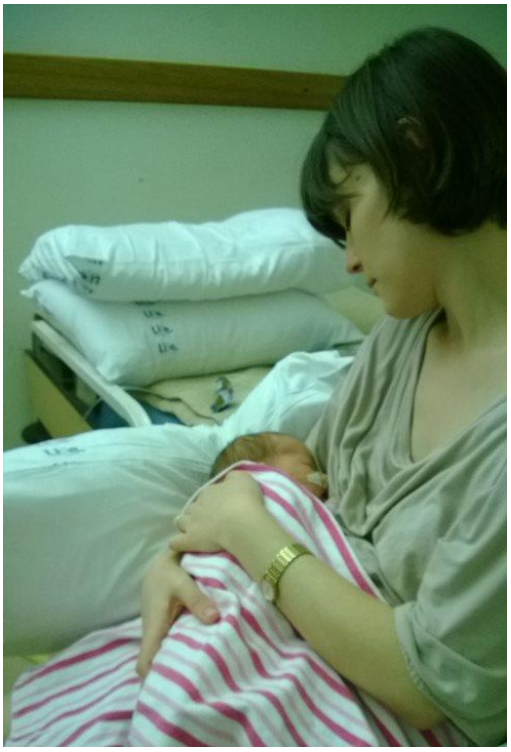
Our journey began 42 years ago. From childhood through adulthood, I was constantly sick, yet no one ever evaluated me for immunodeficiency. In our country, awareness of such conditions is still not widespread. Despite my frequent illnesses, I persevered, often studying from a hospital bed during my university years. Eventually, I graduated and became a teacher, working face-to-face with children—an environment where I was constantly exposed to bacteria and viruses. Unsurprisingly, I was frequently booked off sick, but I managed to work around it. As a chronically ill patient, you just do what you have to do.

In 2013, my eldest was born, and I decided to take a break from full-time teaching to spend more time with her. That decision turned out to be fortuitous. She was only three weeks old when she was admitted to the hospital, gravely ill. Over the next few months, she was in and out of the hospital. Although tests revealed nothing conclusive, things eventually settled down—until she started kindergarten. She was constantly sick again. At that point, an allergist tested her for IgA deficiency and allergies. She tested positive for IgA deficiency, but we were told she would outgrow it. So, we kept going.

In 2016, my youngest was born. From the start, she suffered from a variety of infections—ear infections, pneumonia, and UTIs from unusual pathogens. It was not until 2020, during the pandemic, that we saw a specialist paediatric gastroenterologist familiar with primary immunodeficiency. That marked the beginning of our long journey toward a diagnosis. Initially, tests showed IgA deficiency, but about a year later, her IgG levels began to drop, and she was diagnosed with primary immunodeficiency.

Meanwhile, my own health continued to decline, with more frequent illnesses than ever before. By the end of 2022, I insisted on being evaluated.

After another year of tests, I received an initial diagnosis based on immunophenotyping. However, many questions remained, and our doctors needed guidance as to the next steps, as they suspected our family also had an autoinflammatory condition.



Photos provided by the patient.

cont. Patient journey: Family with PID (South Africa)

During my research, I came across the FMF & AID, while learning about autoinflammatory diseases. I contacted them in search of help and luckily, we were able to undergo paid genetic testing through their medical assistance program.

The results revealed that my eldest daughter and I share a same genetic mutation, resulting in memory B-cell issues and other immune deficits. This information was crucial, as my daughter began to fall ill more often again and suffered a severe case of appendicitis. Armed with this genetic information, we went through a comprehensive testing process, and she is now receiving treatment for her immunodeficiency.

We are incredibly grateful to FMF & AID because without their support, we could not have afforded the genetic testing, which provided us with a clearer understanding of our condition. Thanks to the genetics, we now have access to appropriate treatments, and our doctors know what they are dealing with. Thank you, FMF & AID!

Photos provided by the parents.

Centre of Research Excellence (CRE)

The Hudson Institute of Medical Research in Melbourne Australia has announced the creation of a new Centre of Research Excellence (CRE) for Autoinflammatory Diseases Research and for Nucleic Acid Sensing. These programs aim to develop treatments for a variety of diseases with Prof. Seth Masters and Prof. Carl Walkley leading each. Their work will be focused on improving the quality of life of patients, their families, and ensuring that patient input is a part of the research. This initiative is supported by the National Health and Medical Research Council (NHMRC).

Autoinflammatory diseases are caused by an overactive innate immune system often as a result of changes in certain genes. Recent advances in genetics have enabled scientists to identify the genetic changes responsible for these diseases. The new Centre led by Prof Masters aims to achieve improved patient outcomes by performing basic research to:

-Implement novel biomarker detection methodology
-Validate gene variants of unknown significance
-Identify new pathways driving autoinflammation
-Develop more specifically targeted therapies.

The new Centre led by Prof Walkley will explore nucleic acid sensing pathways. He believes that these sensors can be disease causing and sharing resources and building knowledge will improve patient outcomes. Nucleic acid sensing pathways are the cells' frontline defense against infection from a range of DNA and RNA containing pathogens, particularly viruses. When activated, the body responds to fight infection. Understanding these pathways may lead to the development of new RNA treatments.

Nucleic acids research topics:
-Antiviral Defense Mechanism
-Tumor Immunotherapy and Gene Therapies
-Regulation of Gene Therapy
-Therapeutic Strategies
-Application in Therapy

The new CRE will bring together experts from prestigious institutions including WEHI, the Universities of Queensland and New South Wales, the Garvan Institute of Medical Research, St Vincent's Institute of Medical Research, and major hospitals such as Monash Health, Peter MacCallum Cancer Centre and Sydney Children's Hospital.

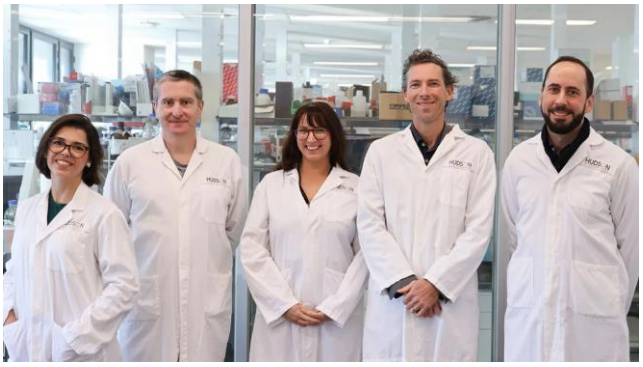


Photo credit: Hudson Institute of Medical Research

New Partner Associations

FMF & AID Global Association is glad to announce the addition of two organizations into our network:

AIPO z.s., Czech Republic

AIPO z.s. is a non-profit association registered in the Czech Republic with the aim of contributing to improving the quality of life of patients with autoinflammatory diseases and their families.

The association is led by Katerina Azim Aburas. It was founded in February 2024, following the initiative of adult patients and parents of affected children wanting to have a formal national representation and voice. The association is working on providing up-to-date information on all autoinflammatory diseases, emotional support, and guidance.

Katerina Azim Aburas, President

APMARR, Italy

APMARR, National Association of People with Rheumatic and Rare Diseases - Italy, celebrates the 40th anniversary of its foundation. Their mission is to improve the quality of patient care and life by offering help and information to people with Rheumatic and Rare Diseases. They do this through volunteering activities, providing qualified psychological support, undertaking awareness campaigns, and hosting several national and local activities, such as free yearly screenings for early diagnosis throughout Italy.

APMARR promotes research in the rheumatological field. They also advocate by organizing workshops and meetings to promote social and human rights, patient health needs for all ages. The organization also brings important attention regarding these issues to political health decision makers and the media.

APMARR also involves and provides continual assistance to patients and their families through their toll-free hotline, magazine “Morfologie” and their social media community of over 5,000 members.

Antonella Celano, President
www.apmarr.it

Disease Terminology and Awareness Dates

Disease	Autoinflammatory Syndromes	Gene	Awareness Day
TRAPS	Tumor necrosis factor- associated Periodic Fever Syndrome	TNFRSF1A	2nd September
NOD2	Blau/Yao Syndrome	NOD2 (CARD15)	3rd September
PFAPA	Periodic fever, aphthous stomatitis, pharyngitis and cervical adenitis	N/A	4th September
HA20	A20 haploinsufficiency	TNFAIP3	5th Sepember
HIDS / MKD	Hyper IgD / Mevalonate Kinase Deficiency	MVK	6th September
AOSD sJIA	Adult-onset Still's disease Systemic Juvenile Idiopathic Arthritis	N/A	7th September
CAPS	Cryopyrin-associated periodic fever syndromes (CAPS):	NLRP3	9th September
	Muckle Wells Syndrome (MWS)		
	Familial cold Autoinflammatory Syndrome (FCAS)		
	Neonatal onset multisystem inflammatory disease (NOMID) Chronic infantile neurologic cutaneous and articular syndrome (CINCA)		
FCAS2	Familial cold Autoinflammatory syndrome 2	NLRP12	10th September
PAPA	Pyogenic Arthritis, Pyoderma gangrenosum and Acne	PSTPIP1	11th September
DADA2	Deficiency of Adenosine Deaminase 2	ADA2	15th September
FMF	Familial Mediterranean Fever	MEFV	17th September
SAPHO	Synovitis-acne-pustulosis-hyperostosis-osteitis syndrome	N/A	19th September
IRAP	Idiopathic recurring acute pericarditis	N/A	25th September
uSAID	Undefined systemic autoinflammatory disease	N/A	29th September
Other autoinflammatory diseases			
HS	Hidradenitis Suppurativa	N/A	6th – 12th June
BD	Behcet's disease	N/A	20th May
CRMO CNO	Chronic recurrent multifocal osteomyelitis Chronic nonbacterial osteomyelitis	N/A	October

FMF & AID Sister and Affiliated Associations

Italy Spain Georgia Czech Republic Turkey United Kingdom Italy Australia

Italy Spain Chile Argentina El Salvador Israel Germany

Italy Brazil Thailand Morocco Italy Spain Middle East France