Issue 05 – 2025/1

AUTOINFLAMMATORY MAGAZINE

ImmunAID Special Edition

ImmunAID
Immunome Consortium for AutoInflammatory Disorders

RARE DISEASE DAY

Streamathon, Podcasts & Educational Brochures

PATIENT STORIES

Patient stories from around the globe

ARTICLES

Ongoing AID struggles, A Path to Motherhood

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

For more information visit fmfandaid.org

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In this edition

Editorial Team

Disclaimer: The patient stories included in this e-magazine, have been provided to the FMF & AID Global Association voluntarily and are being published with the consent and permission of the patients/parents.

Dear Readers,

FMF & AID is excited to publish the first issue of our magazine for 2025. This issue details our participation in ImmunAID, a groundbreaking project, that began in 2018 and aimed to improve the diagnosis and understanding of systemic autoinflammatory diseases. After six years of dedicated work, this initiative has now concluded, and we are incredibly grateful to have collaborated with this project. I would like to extend my sincere thanks to Prof. Bruno Fautrel and INSERM for bringing us on board and recognizing the importance of patient engagement in autoinflammatory research. I also would like to acknowledge the outstanding support received from Emna Chabaane and Frédéric Peyrane.

In this issue, we provide a further look into the ImmunAID initiative, highlighting its impact. We also spotlight our podcast series, which continues to amplify patient experiences and expert insights. We reflect on our Streamathon hosted for Rare Disease Day, connecting patients, caregivers, and the public about rare autoinflammatory diseases.

Autoinflammatory patients still face immense struggles—long diagnostic delays, lack of access to specialists, and limited treatment options. We address these critical issues with our educational brochures, which serve as a vital resource to bridge the knowledge gap and empower both patients and healthcare providers. These brochures are just one of the many ways we continue to raise awareness and advocate for better care.

This edition also features the journeys of several patients offering personal perspectives on the realities of living with an autoinflammatory disease. Additionally, we present an overview of recent research and publications, ensuring that our community stays informed about the latest scientific advancements.

We remain committed to making a difference. Through collaboration, education, and advocacy, we strive to improve the lives of those affected by these rare conditions. Thank you for being part of this journey with us—together, we are stronger.

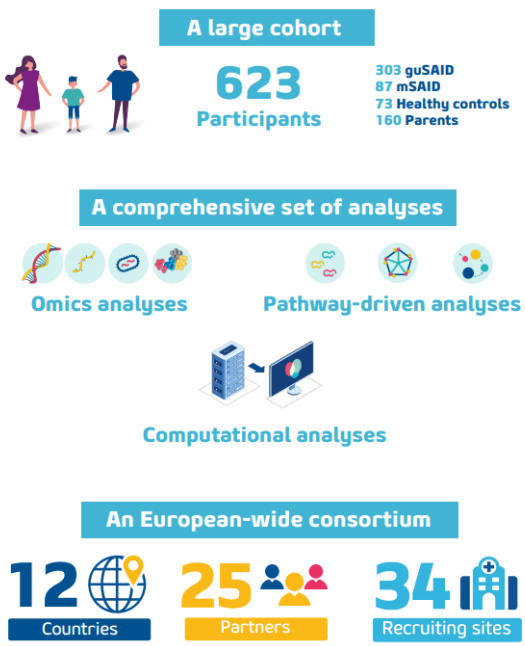
With warm regards,

Malena Vetterli
Founder & Executive Director
FMF & AID Global Association

ImmunAID: A Research Project on Autoinflammatory Diseases

FMF & AID is pleased to announce, after six years of dedicated research, that the ImmunAID project (immunaid.fr/), funded by the EU and led by INSERM (a public research organization in France), has officially concluded. This ambitious initiative focused on learning more about systemic autoinflammatory diseases (SAIDs), which are often misunderstood and misdiagnosed.

FMF & AID was the only patient organization partnering with this project. Our role included dissemination of information, communications, and recruitment of patients across 11 European sites. Our collaboration helped to ensure that this broad European study recruited a diverse segment of autoinflammatory patients to enroll in this program.



Unmet Needs

Autoinflammatory patients often have numerous symptoms that can impact any part of the body contributing to complexity of these disorders. Unlike autoimmune conditions, where autoantibodies are used to diagnose and guide treatment, autoinflammatory diseases have few to no defining biomarkers. The typical patient may be tested for inflammation indicators such as CRP, ESR, SAA and cytokines, however many lack elevations based on time of testing, genetic variants, etc., thus making specificity of an autoinflammatory diagnosis complicated.

Instead, the physician must use a clinical approach, relying on a detailed history to fully understand the pattern and cycles of symptoms associated with disease flares, and a thorough review of extended family history. This data collection can be challenging for doctors to record, due to time constraints, and lack of understanding of rare diseases of the innate immune system.

Often patients will be presumed to have infections and on average are provided with up to 5 inadequate treatments before a correct diagnosis is made. This delay may cause SAID patients, who are seriously ill, tremendous impact to their long-term health, trust of medical systems and quality of life.

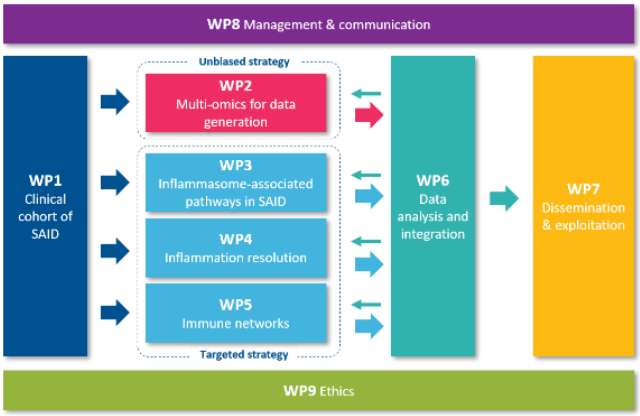
Cont. ImmunAID: A Research Project on Autoinflammatory Diseases

Thus, the overarching goal of the project, in an effort to help both the medical community and autoinflammatory patients, was to bridge the gap between basic research discoveries and clinical applications in autoinflammatory disorders. All while using a wide spectrum of innovative techniques with the following concepts undertaken during discovery and embedded into each of the project's work packages:

- Characterize potential pathogenic pathways at different steps of the inflammation process from generation to its resolution.
- Observe the inflammatory pathways at different biological scales from the molecular level to cells, tissues, and whole organism systems.
- Enable modelling techniques using bioinformatics, biostatistics and mathematical developments (big data approach) to be used to derive and identify novel diagnostic biomarkers and disease classification.

ImmunAID Framework

To ensure that these central goals were adhered to, the ImmunAID initiative was designed with a robust framework to capture a variety of scientific details for best outcomes. Work was divided into several key work packages with the objectives described below.



Package 1: Clinical Cohort

Objective - to establish a large collection of samples and associated clinical data from undiagnosed SAID patients (and their parents), monogenic SAID patients and healthy donors following a rigorously identical process, as a pre-requisite for further biological and computational analyses.

Package 2: Multi-omics

Objective - to generate omic dataset, and possibly identify novel biomarkers, which might be targets for future drug intervention.

Package 3: Inflammasome-associated pathways

Objective - to assess the range of functions, dysfunction and biomarker potential of the inflammasome complexes in SAIDs. By correlating with omics data from WP2, further inflammasome-associated signatures will be sought.

Cont. ImmunAID: A Research Project on Autoinflammatory Diseases

Package 4: Inflammation resolution

Objective - to characterize a network of lipid mediators, biosynthetic pathways and functional effects in SAID patients and identify potential abnormalities in the inflammation resolution process.

Package 5: Immune networks

Objective - to gain knowledge on individual inflammation factors through the profiling of soluble factors (e.g. cytokines, alarmins, etc.) and specific immune cells (e.g. NK cells, Tregs) and the characterization of protein structures and modifications.

Package 6: Data analysis and integration

Objective – to use the latest methodologies in bioinformatics, statistical modelling, machine learning, and deep learning will be applied to the ImmunAID datasets (throughout WP1, WP2 & WP5).

Package 7: Dissemination and exploitation

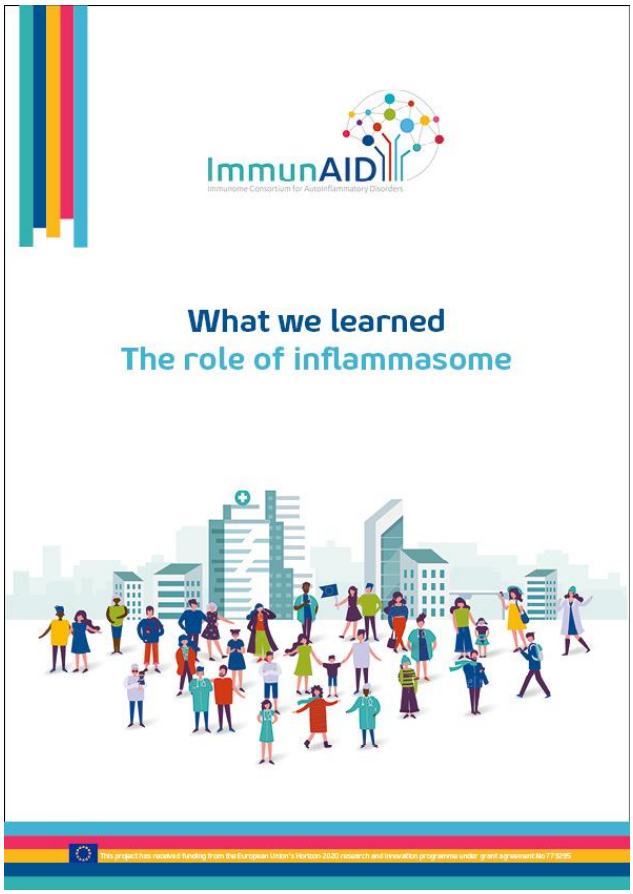
Objective - to translate research results appropriately for patients with a focus on the impact of the inflammasomes in SAIDs.

Package 8: Management and communication

Objective – to implement rigorous management procedures which will be set up to help steer project advances, anticipate risks and ensure the achievement of the project's objectives.

ImmunAID Impact

While the project has closed, the final data and results will be published over the next several months of 2025. FMF & AID looks forward to providing this information to our patient communities.



ImmunAID - Review of what we have learned

What are SAIDs?

Systemic Autoinflammatory diseases (SAIDs) are a group of rare disorders resulting in recurrent episodes of inflammation.

Symptoms are different and unique to each patient: fever, abdominal pain and other gastro-intestinal issues, headaches, skin rashes, eye issues, joint pain and swelling, myalgias, fatigue, peritonitis, pericarditis, etc.

Autoinflammation can affect all ages, genders and ethnicities and due to ancient and modern migration patterns, patients are found globally. While treatment exists, there are NO cures for these diseases.

Why are SAIDs difficult to diagnose?

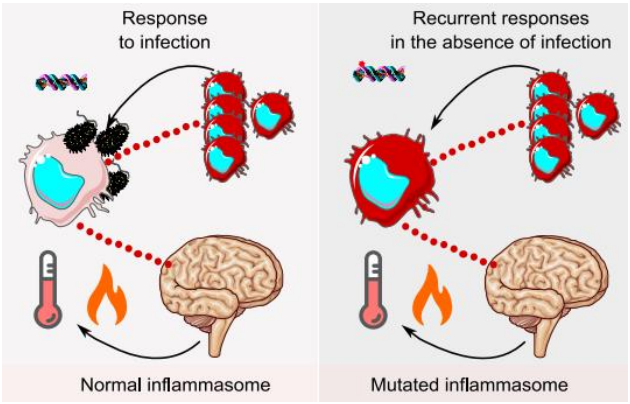
- Complex clinical presentation is unique to each patient
- Disease can evolve over time and present differently during aging
- Family members, with the same disease have varying symptom presentations
- Patients may or may not have elevations in inflammatory markers
- Monogenic mutations may or may not be detected during genetic testing

Most common autoinflammatory genes include MEFV (FMF), NLRP3 (CAPS), NLRP12 (FCAS2), NOD2 (Yao and Blau syndromes), MVK (MKD), TNFRSF1A (TRAPS), TNFAIP3 (HA20), etc.

Average time to diagnosis: 3 years for pediatrics and 14 years for adult patients.

What is the inflammasome?

The inflammasome, which is dysregulated in certain autoinflammatory diseases, maintains the body's complex surveillance and sensor response system activated during infection or by cancer cells. It is responsible for the activation of inflammatory responses and cell death. Genes encoding these sensors cause Familial Mediterranean Fever (FMF), Cryopyrin-Associated Periodic Syndrome (CAPS), and PAPA syndrome. See more at immunaid.fr/images/PDF/Legacy_inflammasome_v1.pdf



Source/credit: ImmunAID for the work and graphics.

FMF & AID Streamathon, New Merch, and Raising Awareness

Rare Disease Day ensures the voices of those around the world who live with a rare disease are heard. From February 27 to March 2, the FMF & AID Streamathon returned, streaming live 24/7, to raise funds and awareness for our autoinflammatory cause.

Chris Walker, our Fundraising and Community Outreach Officer, and an FMF patient himself, knows firsthand what it is like to grow up without support or understanding. He is committed to helping others through advocacy, and every year, he puts on an unforgettable show. He hosts a variety of activities to engage viewers—singing karaoke, playing games, interviewing patients, and even taking on crazy challenges—all to raise awareness and funds for FMF & AID. The fundraiser linked to the Streamathon allowed supporters to donate directly to help rare disease children.

This year, we amplified the message with our new Rare Disease Day merch, featuring the bold statement “I’m Rare” alongside with our two FMF & AID mascots:

JJ, the super dog, who uplifts the children’s community with his inspiring stories.

Dr. Sharpie, the Shar-Pei doctor, who represents Familial Shar-Pei Fever, the canine version of FMF, reminding us that Familial Mediterranean Fever affects more than just humans.

Every purchase of our “I’m Rare” merch helps support our advocacy efforts and spreads awareness. Whether you tuned in to the Streamathon, purchased some merch, or shared our posts, we appreciate you making a difference.

Rare Disease Day, February 28th, 2025, once again brought everyone together to support our cause, show resilience, and demonstrate that rare is not invisible.

You can continue to support our fundraising and awareness efforts by:

✅ Watch the recorded Streamathon patient interviews (see pp. 10-12)

✅ Donate to our fundraiser: streamlabscharity.com/@reds.../rare-disease-day-2025

✅ Shop our merch www.zazzle.ch/kollektionen/rare_disease_awareness-119156308160487760

✅ Spread awareness on social media

New Educational Brochures Available Online

At FMF & AID, we understand that knowledge is power—especially for those living with autoinflammatory diseases. Access to clear, reliable, and patient-friendly information can make a significant difference in understanding and managing these conditions. We are pleased to announce the addition of several new brochures to our online resource library. These brochures provide essential information on various autoinflammatory diseases and key aspects of treatment management not only for patients and parents, but also for healthcare professionals.

The information provided has been carefully reviewed and endorsed by Prof. Dr. Jürgen Rech, Head of the specialized Autoinflammatory Reference Center in Erlangen, Germany. His expertise ensures these brochures provide accurate, up to date, and valuable life-impacting information for patient care. New additions include:

Familial Mediterranean Fever (FMF) – A guide to the most common autoinflammatory disease, covering symptoms, diagnosis, and treatment options.

Cryopyrin-Associated Periodic Syndromes (CAPS) – An overview of this rare condition, which spans FCAS, Muckle-Wells syndrome, and NOMID/CINCA.

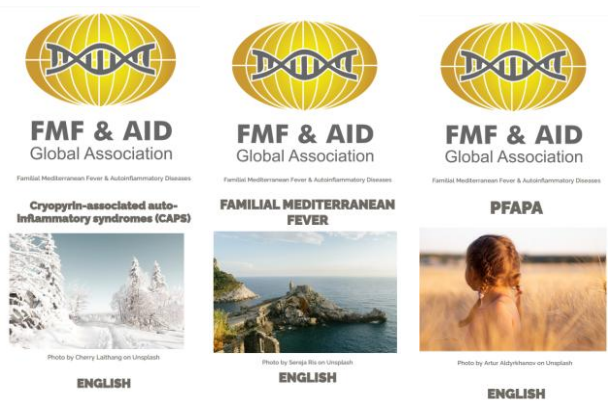
TNF Receptor-Associated Periodic Syndrome (TRAPS) – A resource for understanding this complex condition, which presents with long-lasting recurrent flares.

Periodic Fever, Aphthous Stomatitis, Pharyngitis, and Adenitis (PFAPA) – A common periodic fever syndrome mainly affecting young children.

Pain Management – A crucial resource for autoinflammatory patients dealing with pain, offering information and available treatments.

FMF & AID is currently working on additional brochures, further expanding our collection of educational materials. All brochures are available for free and can be found on our website under the Publications section: www.fmfandaid.org.

We encourage everyone in our community—patients, caregivers, and medical professionals—to take advantage of these resources and share them with others. By increasing awareness and education, we hope to increase the knowledge regarding these diseases and support those affected by them.



Patient interviews

During the FMF & AID Streamathon, Chris had the opportunity to interview several patients and parents who shared their personal journeys with autoinflammatory diseases. Their stories highlight the challenges of diagnosis, access to treatment, and their medical experiences.

Sara, a young patient with FMF

Sara was diagnosed with Familial Mediterranean Fever (FMF) as a child through genetic testing. One of the biggest challenges she faced was transitioning from pediatric to adult care. She went through a long process of "doctor shopping" before finally finding a rheumatologist who supported and advocated for her. Additionally, being a Caucasian American with FMF, rather than being of Mediterranean descent, has made her diagnostic journey particularly difficult. Despite these obstacles, one of the most rewarding aspects of her experience has been meeting her best friend through the FMF & AID support groups.

Watch Sara's interview:
www.youtube.com/watch?v=vGqpQZDWvhk

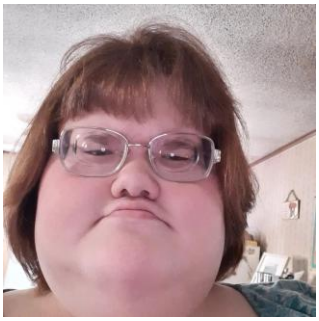


Photo provided by the patient

Ludmila, mother to Oleksii, a patient with MKD

Ludmila and her son Oleksii are from Ukraine and, due to the ongoing war, were forced to relocate abroad. They now live in Hungary. Oleksii, now 35 years old, was not diagnosed with MKD until the age of 30. Unfortunately, due to years of untreated disease, he suffered two strokes that left him mentally disabled. Since starting biological treatment, his symptoms have been well controlled. However, his well-being is now at serious risk as they are running out of medication (Kineret/anakinra). Ludmila fears the devastating and potentially fatal consequences this could have on her son. If anyone can provide assistance, please contact: info@fmfandaid.org.

Watch Ludmila's interview:
www.youtube.com/watch?v=PPzc6ekdTNE

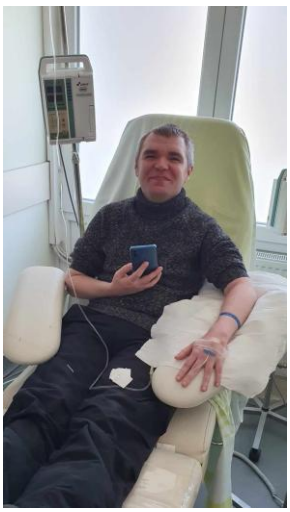


Photo provided by the parent

Cont. Patient interviews

Gaby, a patient with uSAID

Gaby, a healthcare worker, was diagnosed with uSAID (undifferentiated systemic autoinflammatory disease). She first came into contact with FMF & AID through the helpline, where she received invaluable guidance on living with her condition. Through the support groups, she found a sense of community and no longer felt alone. She was surprised to learn that all the resources provided by FMF & AID were free, and this access has been mentally and emotionally uplifting for her.

Watch Gaby's interview:
www.youtube.com/watch?v=sjn_Nc_VSdM

Photo provided by the patient

Monica, parent to a child with FMF

Monica is the mother of a five-year-old child diagnosed with FMF. She described the struggles she faced as a parent advocating for her son and the many hurdles she encountered before he was finally taken seriously, correctly diagnosed, and treated. Recently, another one of her children began showing symptoms and has since been diagnosed with FMF as well. Her journey as a mother inspired her to write a book, Searching for a Zebra, which recounts her experiences navigating the medical system and fighting for answers.

Watch Monica's interview:
youtu.be/QFMl4lmFPy8

Photo provided by the parent

Cont. Patient interviews

Rachel, a patient with MKD

Rachel is a patient with MKD and also runs the RACC-UK patient association in the United Kingdom. She shared her experience growing up with the disease and enduring multiple misdiagnoses. Although an autoinflammatory disease was first suspected when she was eight years old, genetic testing and treatment options were not available at the time. It wasn't until she was 16 that she was finally able to undergo genetic testing in the Netherlands and receive a proper diagnosis.

Watch Rachel's interview:
www.youtube.com/watch?v=fQEREbgOmXI

Photo provided by the patient

Debbie, a patient with FMF

Despite experiencing symptoms since the age of 16, Debbie was only diagnosed correctly with Familial Mediterranean Fever in adulthood. Her journey was complicated by additional health concerns, and sadly, many doctors dismissed her symptoms or attributed them to other conditions. She spoke about the frustration of not being taken seriously and the challenges of getting proper care.

Watch Debbie's interview:
www.youtube.com/watch?v=VI3axHwIoo0

Photo provided by the patient

These powerful stories remind us why advocacy, awareness, and support are crucial for the autoinflammatory community. Through shared experiences, we continue to push for better understanding, faster diagnoses, and improved access to treatment for all patients.

Patient journey: Matías with NOMID (Colombia)

My son Matías was born in Medellín Colombia. His story began when he was 13 months old and had a fever of 39°C/102.2°F degrees for 6 days. I took my baby to the hospital, and we were sent home because his fever was not accompanied by other symptoms.

Ten days later, he started crying desperately and began running a fever, which would not resolve with any OTC medications. I took him to the hospital where the doctors did bloodwork. The results indicated that his CRP and sedrate were elevated. They then decided to refer him to a specialty hospital, where he was hospitalized for a whole month. Luckily, the doctors managed to control his fevers after doing a lumbar puncture and discovering that he had aseptic meningitis.

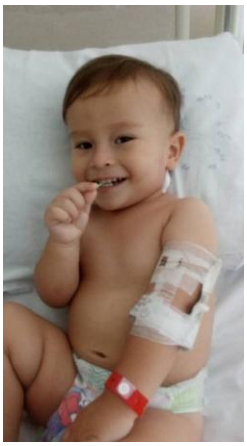


Photo provided by the parents

After that long month, we were finally able to go home. However, within a month and a half, my son started to have leg pain and run a fever again, so we returned to the hospital.

His rheumatologist and neurologist decided to undertake further studies in search of a diagnosis. However, this investigation was delayed due to new symptoms, as he began to exhibit including skin rash, joint inflammation, and irritability. Once again, he had meningitis, and the doctors could not figure out the reason and decided to treat him with daily steroids. For many months, his inflammation was well controlled with this treatment.

Two years later, we were asked to return to the hospital's rheumatology department, so that they could take a blood sample and send it to Spain, where a board of doctors would review my son's case.

After waiting a long time for the results, it was concluded he was a patient with an orphan disease called NOMID. At the same time, they informed us that the medication that could help my son was not yet available in the Invima registry in Colombia.

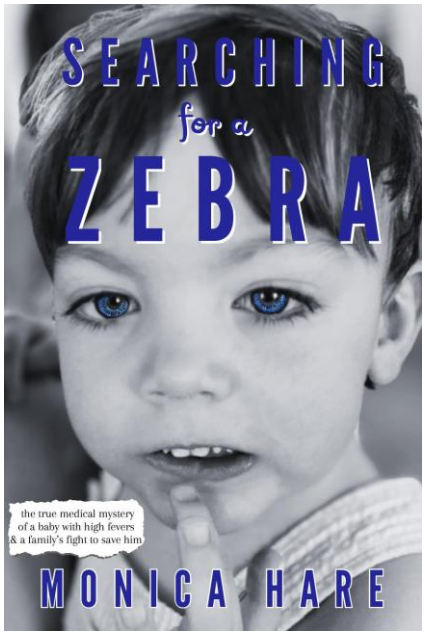
We contacted a health management representative for legal protection, who helped us file a comprehensive request to access this medication, and after 3 months, we achieved success. When Matías began Canakinumab, the doctors discovered that the dose they were giving him was not high enough to control his inflammation, as he required a blood transfusion and another lumbar puncture.

Cont. Matías with NOMID (Colombia)

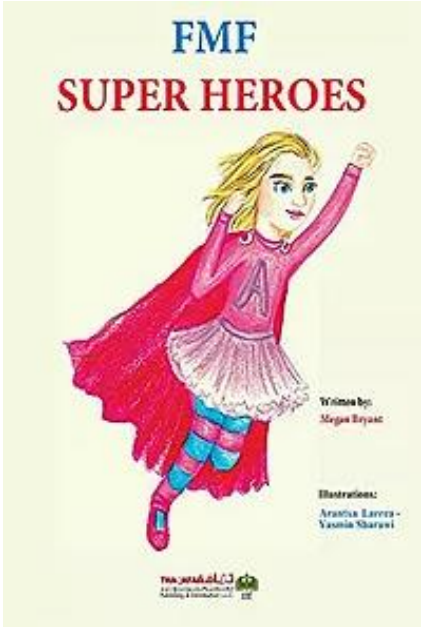
It was then decided to increase his dose. Today he is 8 years old and has not needed to go to the hospital for over a year. We are so thankful that his illness is finally well controlled.

I am grateful to FMF & AID for all the support and guidance they have provided for the past four years. This has allowed our family to have a better understanding of our child's disease. I also deeply appreciate FMF & AID for facilitating genetic testing and covering the cost, which ultimately allowed for Matías' NOMID diagnosis.

Photo provided by the parents



The book can be purchased on Amazon. 20% percent of all proceeds from the sale of the book are generously being donated to the FMF & AID Global Association medical fund.



The educational children's e-book "FMF Superheroes" is available in English, German, and Arabic. This book is great for children of all ages. Purchasing a copy also supports the FMF & AID Global Association's work.

Patient journey: Tamara with FMF (Argentina)

My daughter Tamara was born at 35 weeks. She is the youngest of three sisters and from her first month of life, she would cry continuously, and we did not know why. At eight months, she began to have episodes of fever without any sign of infection. Her pediatrician claimed this was normal as she was building her immunity. At the age of four, she had a serious fever episode that led to her first hospitalization. She was vomiting, lost strength in her arms and legs, could not explain verbally what was happening to her, and she just wanted to sleep. The lab results from the hospital were inconclusive and the doctors said there were too many symptoms to diagnose a single disease.

Going forward, every three months, she kept having the same type of recurring episode accompanied by fever, vomiting, lack of strength, and leg pain which prevented her from walking. Her doctors, who did not understand her symptoms, told us that she needed further investigation with other specialists. We visited many hospitals, saw many doctors, had multiple tests done, but there were no answers for her flares. We desperately began to search online as our family went through a very hard time throughout this diagnostic odyssey.

Tamara had been hospitalized more than 15 times, and despite this, the doctor kept questioning her symptoms saying that she must be making up her illness. The same doctor began accusing me of feeding my child with lies, which made me doubt for a whole year if what was happening to her was indeed real. She was so delicate that I did not want to continue exposing her to more medical investigations. Despite her symptoms, we kept her away from the doctors and did not seek medical attention during sick times to protect her.

Unfortunately, she suffered another severe episode requiring hospitalization. During her stay, she was given pain and fever reducing medications, along with corticosteroids via IV, despite not knowing what disease she was being treated for.

Disappointingly, social services would not support genetic testing, but luckily, we came across ALAPA (the Argentine Alliance of Patients with Rare Diseases), and with their help, we were able to obtain the first genetic test for our daughter.

Photos provided by the parents

Cont. Tamara with FMF (Argentina)

Testing confirmed that Tamy had Familial Mediterranean Fever (FMF), but even with her new diagnosis, there was no doctor who would treat her. Additionally, the doctors did not know how to interpret the genetic results, nor did they know anything about FMF. Once again, we felt abandoned, and I feared for my daughter's well-being and was concerned that one day she would be unable to permanently walk.

Once again, we turned to social media for information and support, and this time we found the FMF & AID Global Association. I contacted them and from the first discussion, we finally felt understood as to what was happening to our daughter medically. We were advised to seek a physician specialist outside of our city, approximately two hours away, so that Tamy could receive expert care.

Since she failed colchicine, a year ago she began treatment with ILARIS. Getting her on this medication was not an easy task as social services did not want to cover the cost. We resorted to hiring lawyers to defend the rights of our daughter to have access to this life-saving biologic medication.

Although we know that the disease has no cure, her flares have become milder on this treatment. We are grateful to Malena from FMF & AID for her support in making our diagnostic journey shorter by recommending such wonderful autoinflammatory experts who care about my daughter's health.

Although the bad experiences have left painful memories, we are so happy that in the end, our daughter was finally able to receive proper diagnosis and treatment.

Photos provided by the parents

Autoinflammation Decoded & Beyond

FMF & AID Global Association is excited to introduce their brand-new podcast series, Autoinflammation Decoded & Beyond moderated by Ellen Cohen. This series dives into the world of autoinflammatory diseases and other related topics, featuring conversations with leading research experts, physicians, patient advocates, patients, etc.

Professor Seth Masters

In our debut episode, FMF & AID has the honor of speaking with Prof. Seth Masters, Head of the Centre for Innate Immunity & Infectious Diseases at the Hudson Institute in Australia. He shares his expertise on innate immunity and its connection to autoinflammatory diseases.

Spotify: podcasters.spotify.com/pod/show/fmf--aid

Apple: https://podcasts.apple.com/us/podcast/autoinflammation-decoded-and-beyond/id1767156698

YouTube: youtu.be/1FEwyc2oDCY?si=H_t_wpS0lplj0Qpl

Sharon Kensell

In our second episode, FMF & AID features Sharon Kensell, Founder & Chair of the FMF & AID Australian Association and a passionate patient advocate. She discusses her journey with FMF, her dedication to raising awareness and supporting the Australian community.

Spotify: podcasters.spotify.com/pod/show/fmf--aid

Apple: podcasts.apple.com/us/podcast/autoinflammation-decoded-and-beyond/id1767156698

YouTube: youtu.be/tI5LyVkFLkE

Rachel Rimmer

In our third episode, FMF & AID speaks with Rachel Rimmer, Founder & Chair of RACC-UK (Rare Autoinflammatory Conditions Community - UK). She shares her experiences as an advocate and discusses the challenges and successes of building a supportive community for those affected by rare autoinflammatory diseases.

Spotify: podcasters.spotify.com/pod/show/fmf--aid

Apple: podcasts.apple.com/us/podcast/autoinflammation-decoded-and-beyond/id1767156698

YouTube:www.youtube.com/watch?v=UgCG6GYAPcg&t=6s

🎙️ Stay tuned for more insightful conversations by listening to Autoinflammation Decoded & Beyond on your favorite platform and join us as we work together to support and educate the autoinflammatory community worldwide!

The Ongoing Struggle: Finding a Doctor Who Understands Autoinflammatory Diseases

For patients living with systemic autoinflammatory diseases (SAIDs), receiving a proper diagnosis and care is often a long, frustrating journey. Unlike more well-known conditions, these diseases remain largely unfamiliar to many healthcare providers, making it difficult for patients to find a knowledgeable specialist or general practitioner with basic understanding of these rare and complex genetic conditions.

A Widespread Challenge

Many patients report years of misdiagnosis, delayed treatment, and uncertainty before finally being correctly diagnosed. Unfortunately, SAIDs are not included in standard medical curriculum; thus, specialists struggle to identify these patients, often mistaking their symptoms with infections, autoimmune disorders, etc.

As a result, patients are left doctor-shopping, undergoing unnecessary tests and procedures, and facing skepticism. For those with severe disease manifestations, this delay in care can cause irreversible organ damage, chronic pain, and a significant impact on quality of life.

Raising Awareness: Our Contribution

At FMF & AID, we recognize the diagnostic urgency of all SAIDs and as a result, we are committed to educating both the medical community and patients about these diseases.

As part of our global efforts, we have developed a series of brochures reviewed and endorsed by a recognized autoinflammatory specialist.

- FMF
- CAPS
- PFAPA
- TRAPS
- MKD
- Kindergarten & Schools
- Colchicine
- Pregnancy & Menstruation
- Pain management
- Antibiotics & Steroids
- Biological medications

www.fmfandaid.org/publications

Awareness Videos

FMF & AID has also produced a series of autoinflammatory related videos available in several languages at the FMF & AID YouTube channel. These can be used as an effective tool to educate family, friends, caregivers, healthcare professionals and the general public. We strive to unravel many myths and misconceptions regarding these rare diseases.

- Recognizing Autoinflammatory Diseases
- Myths and Facts in Autoinflammatory Diseases
- PFAPA
- Familial Mediterranean Fever
- Colchicine

www.youtube.com/@FMFandAID/videos

FMF & AID Helpline

We continue to assist patients and parents via our WhatsApp helpline. By providing reliable data and information, and support to empower those living with autoinflammatory diseases.

Helpline: +41 77 265 2644

or through Facebook/Messenger

An FMF Patient's Path to Motherhood

I want to share my fertility journey with other women who have Familial Mediterranean Fever (FMF), so they don't have to face the same challenges I did while trying to conceive and maintain a pregnancy. I want you to know that, in the end, my husband and I were blessed with twin boys born in 2024.

Ten years ago, I married a wonderful man, and together, we were hopeful about starting a new life and family. Unfortunately, my first year of marriage was marked by medical complications that, unknowingly, would affect my ability to have children.

A terrifying emergency room visit left me fearing I had meningitis. Earlier that week, I had gone to a private hospital with chest pain while breathing, but I was sent home without answers. That same night, I vomited and lost consciousness, prompting my husband to call an ambulance.

I was rushed to the hospital, where doctors discovered I had an enlarged heart, fluid in my lungs, and systemic edema. It was then that I was diagnosed with FMF and prescribed daily colchicine. My symptoms improved, and thankfully, I started to feel better. However, my doctor never fully explained the medication's effects on my body.

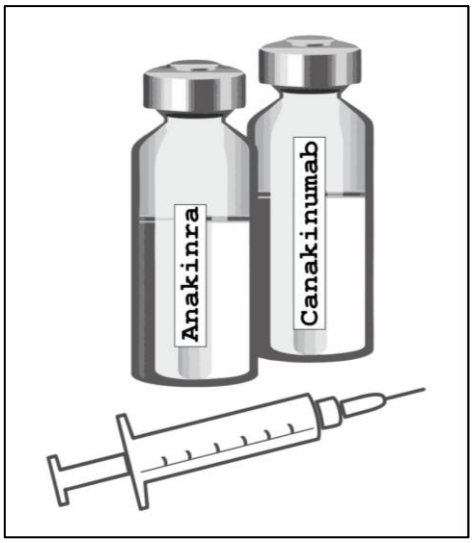
My fertility journey began with disappointment as I spent a year trying to conceive after my FMF diagnosis, only to face repeated failure. I then pursued IVF in my country, but it was unsuccessful. My husband and I traveled abroad to undergo IVF in several other countries, yet each attempt ended in failure. After enduring numerous procedures and devastating miscarriages, I was heartbroken.

Determined to find answers, I consulted with multiple physicians. Finally, one doctor realized that the colchicine, I had been taking daily for FMF, was likely impacting my ability to become pregnant. He explained that colchicine can halt cell division, and in a small number of women, and its daily use may impair conception and pregnancy. I was beyond shocked and upset—after more than a decade of searching for answers, not a single doctor had ever mentioned this risk.

Photo credit: Amina Filkins on Pexels.com

Cont. An FMF Patient's Path to Motherhood

Fortunately, I reached out to FMF & AID Global Association, where I was provided with scientific articles and information explaining that while some FMF female patients must stop colchicine to conceive, others do not. In cases where stopping colchicine is necessary, anakinra (anti-IL-1 biologic) can be used as an alternative.



Unfortunately, this medication was not available in my country except through a lengthy approval process or privately at a high cost. This was not an option for me, leaving me with a difficult dilemma: stop colchicine and risk a flare-up that could endanger the embryos, or continue taking it despite the uncertainty.

My last two embryos were in a freezer, and at 43, this was my final chance to a baby, so I decided to stop colchicine a few days before the embryo transfer and wait to resume it until I had a positive pregnancy test.

To reduce the risk of an FMF flare, I was put on prednisone during this time, which worked remarkably well. Once implantation was confirmed, I was able to restart colchicine without any issues, and thankfully, the rest of my pregnancy went smoothly.

Photo credit: Nik for Unsplash.com

I am incredibly grateful for the support of FMF & AID and feel beyond blessed to have two healthy baby boys. My journey serves as an important lesson for women who may need to temporarily stop colchicine to conceive and a reminder to always ask critical questions about how medications affect the body. Every one of us responds differently to life-saving colchicine.

Photo credit: Greta Fotografía on Pexels.com

2025 Noteworthy Autoinflammatory Medical Abstracts/Papers

Patients with Adult-Onset Still's Disease in Germany: A Retrospective Analysis of Clinical Characteristics and Treatment Practices Ahead of the Release of the German Recommendations

By Schoenau, V.; Wendel, S.; Tascilar, K.; Henes, J.; Feist, E.; Baerlecken, N.T.; Popp, F.; Schmidt-Haendle, M.; Hellmich, B.; Kötter, I.; et al. J. Clin. Med. 2025, 14, 981.

Abstract

Background/Objectives: Adult-onset Still's disease (AOSD) is an autoinflammatory disorder that can be challenging to diagnose and manage. The aim of this study was to analyze retrospective data to provide insights into the clinical presentation, disease activity, and treatment patterns and outcomes of AOSD during routine clinical care prior to the release of new AOSD guidelines.

Methods: This retrospective database analysis evaluated adult patients (≥18 years) with a diagnosis of AOSD who engaged in a clinical visit between 1 January 2010 and 31 December 2020. The evaluated outcomes included demographic characteristics, symptoms, disease activity, and treatment.

Results: Our study included 120 patients (67 [55.8%] of whom were female) diagnosed with AOSD according to the Yamaguchi criteria at ten German rheumatology centers. The median (quartile [Q] 1, Q3) age was 51 (36, 62) years, and the median (Q1, Q3) time from diagnosis was 9 (4, 11) years.

Approximately half (66 [55.0%]) had a polycyclic disease course.

The most frequent symptoms at initial diagnosis were arthralgia (105 [87.5%]) and fever (86 [71.7%]), and these symptoms continued for a substantial proportion of patients at the current visit (35 [29.2%] and 22 [18.3%], respectively). High neutrophil and ferritin levels were also common. The mean Still Activity Score, a measure of disease activity, improved from 4.66 at initial diagnosis to 1.97 at the most recent visit. The treatments most frequently used at some point in the disease course were glucocorticoids (118 [98.3%]), interleukin (IL)-1 inhibitors (89 [74.2%]), and methotrexate (85 [70.8%]). The most common current treatments were IL-1 inhibitors (55 [45.8%]), followed by methotrexate (29 [24.2%}) and glucocorticoids (28 [23.3%]).

Conclusions: Our cohort of patients with AOSD seen at German rheumatology clinics showed strong improvements in symptoms and disease activity from initial diagnosis, but a high symptom burden remained for some patients. Future studies may be able to build on our data to document the impact of new guidelines on treatment patterns.

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Cont. 2025 Noteworthy Autoinflammatory Medical Abstracts/Papers

Current landscape of monogenic autoinflammatory actinopathies: A literature review.

By Mertz P, Hentgen V, Boursier G, Delon J, Georgin-Lavialle S. Autoimmun Rev. 2025 Jan 31;24(2):103715. Epub 2024 Dec 5.

Abstract

Autoinflammatory diseases (AID) are conditions leading to a hyperactivation of innate immunity without any underlying infection, and may be poly- (e.g. Still's disease) or monogenic. The number of monogenic AID is continuously expanding, with the discovery of novel pathologies and pathophysiological mechanisms, facilitated in part by easier access to pangenomic sequencing. Actinopathies with autoinflammatory manifestations represent a newly emerging subgroup of AID, associated with defects in the regulation of actin cytoskeleton dynamics. These diseases typically manifest in the neonatal period and variably combine a primary immunodeficiency of varying severity, cytopenia (particularly thrombocytopenia), autoinflammatory manifestations primarily affecting the skin and digestive system, as well as atopic and autoimmune features. Diagnosis should be considered primarily when encountering an early-onset autoinflammatory skin and digestive disorder, along with a primary immunodeficiency and either thrombocytopenia or a bleeding tendency. Some of these diseases exhibit specific features, such as a risk of macrophage activation syndrome (MAS) or a predisposition to atopy or lymphoproliferation. The complete pathophysiology of these diseases is not yet fully understood, and further studies are required to elucidate the underlying mechanisms, which could guide therapeutic choices. In most cases, the severity of the conditions necessitates allogeneic marrow transplantation as a treatment option. In this review, we discuss these novel diseases, providing a practical approach based on the main associated biological abnormalities and specific clinical characteristics, with a special focus on the newly described actinopathies DOCK11 and ARPC5 deficiency. Nonetheless, genetic testing remains essential for definitive diagnosis, and various differential diagnoses must be considered. pubmed.ncbi.nlm.nih.gov/39644982/

Yao syndrome: a novel systemic autoinflammatory disease with cutaneous manifestations

By Shakhashiro M, Sadeghian S, et al. Int J Dermatol. 2025 Jan;64(1):44-50.

Abstract

Yao syndrome (YAOS) is a novel systemic autoinflammatory disease linked to the nucleotide-binding oligomerization domain (NOD2) gene.

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It is characterized by periodic fevers, gastrointestinal (GI) symptoms, arthritis, and dermatitis, among other symptoms. A sparse literature exists on this disease, and little is known about its dermatological manifestations.

A review of available literature was performed to characterize the cutaneous manifestations of Yao syndrome. Cutaneous manifestations were documented in 85.7% of patients, with common characteristic descriptions of erythematous patches and plaques involving the face, trunk, abdomen, and extremities. Based on our review of treatment modalities employed for Yao syndrome, prednisone is an appropriate initial approach, with oral sulfasalazine and other disease-modifying antirheumatic drugs serving as appropriate secondary options. YAOS should be considered in the differential diagnosis of patients presenting with a dermatitic rash, especially in the context of concurrent articular symptoms, periodic fever, and GI symptoms. pubmed.ncbi.nlm.nih.gov/38965064/

VEXAS syndrome: an adult-onset autoinflammatory disorder with underlying somatic mutation

By Kötter I, Krusche M. Curr Opin Rheumatol. 2025 Jan 1;37(1):21-31.

Abstract

Purpose of review: VEXAS syndrome (Vacuoles, E1 enzyme, X-linked, Autoinflammatory, Somatic) was first described in 2020, where in a cohort of adults with unexplained fever or inflammation, systematic genetic testing was performed and 25 men with a median age of 64 years and somatic mutations in the UBA1 gene were identified. In the current review, we aim to discuss the relevant literature from January 2023 until July 2024 to give new insights into the pathophysiology, epidemiology, diagnosis and treatment of VEXAS.

Recent findings: VEXAS affects 1 : 4269 in men over the age of 50. Janus-Kinase-inhibitors (JAKi) and IL-6-inhibitors are more effective immunosuppressants against hyper-inflammation. Ruxolitinib is more effective than other JAKi. Azacitidine induces remission in many patients, but only few MDS-associated patients were treated. Allogeneic stem cell transplantation is feasible for selected cases. Infections are the major cause of death. Prognosis is still poor with a 5-year mortality rate of 18-40%.

Summary: In the current review, we discuss the novelties for VEXAS, including pathogenic pathways, epidemiological data, diagnostic criteria and algorithms, treatment options and complications. We hope that this review may improve rheumatologists' understanding of VEXAS.

Cont. 2025 Noteworthy Autoinflammatory Medical Abstracts/Papers

We strongly recommend enrolling VEXAS patients in registries and clinical trials, to improve prognosis of VEXAS in the future.

pubmed.ncbi.nlm.nih.gov/39470174/

Colitis in a patient with familial Mediterranean fever: Is it Crohn's disease or ulcerative colitis?

By Hoshi A, Shimodate Y, et al. 2024 Sep 18;5(1):e70013.

Abstract

A 24-year-old woman was referred to our hospital with joint pain, fever, abdominal pain, and diarrhea. A colonoscopy revealed longitudinal ulcers with a cobblestone appearance throughout the entire colon, suggestive of Crohn's disease. However, treatment with 5-aminosalicylic acid, azathioprine, and infliximab failed to achieve clinical remission. A colonoscopy 5 months later revealed a diffusely spreading granular mucosa without visible vasculature, compatible with active ulcerative colitis. Based on these serial changes in colonic lesions, we tested the patient for MEFV gene mutations and found variants E148Q and L110P in exon 2. Administration of colchicine resulted in complete clinical remission. Our experience suggests that drastic changes in the features of colonic inflammation may be a clue to the diagnosis of enterocolitis associated with familial Mediterranean fever. pubmed.ncbi.nlm.nih.gov/39295638/

Increasing Importance of Genotype-Phenotype Correlations Associated with Common and Rare MEFV Gene Mutations in FMF Patients in the Last Thirty Years

By Yildirim, S.; Bekis Bozkurt, H.; Erguven, M. J. Clin. Med. 2025, 14, 712.

Abstract

Background/Objectives: Studies have shown that some mutations, especially M694V, are correlated with renal RI and/or AA. There is limited data about rare mutations on severity of the disease and RI. Today, evaluating genotype-phenotype correlations in rare mutations is important to better understand FMF. We aimed to evaluate clinical, demographic and genetic changes and genotype-phenotype correlations in pediatric patients with FMF over thirty years as well as the importance of the rare mutations. Methods: A total of 2765 pediatric patients with FMF were included in this study. Results: There was a significant increase in compound heterozygous mutations, E148Q het/hom, R202Q het/hom, complex mutations and rare mutations in the last decade.

Conclusions: It may be misleading for clinicians that mutations which have increased in frequency over the years are clinically mild. RI and AA rates in rare mutations are not less than the related rates in common mutations.

doi.org/10.3390/jcm14030712

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

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