Issue 03 – 2023

AUTOINFLAMMATORY MAGAZINE

AWARENESS MONTH

September is Autoinflammatory Awareness Month

PATIENT STORIES

Real stories from patients around the globe

WORKING TOGETHER

News from our affiliates

This digital magazine is presented by the FMF & AID Global Association and its affiliated associations.

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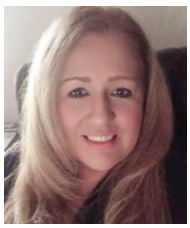
Editorial Team



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Dear Readers,

Welcome to our 3rd edition of the FMF & AID Magazine!

It is amazing how time flies, and again, ready to welcome September as the **World Autoinflammatory Month**. Many of you have probably taken some family time in August to go on vacation or to relax at home, time that our volunteers have effectively worked on this magazine and prepared this year's awareness campaign.

As an international umbrella organization, we are thrilled to share that the representatives of our European affiliates met in person in Budapest for the first time! The aim of our meeting was not only to strengthen our relationships, but also to coordinate our work and activities more efficiently. Hopefully, one day soon, we will be able to host a global meeting and meet in person with our affiliates from Australia, the Middle East, Asia, South and Central America, and the United States. Another important development is that our organization has established sister organizations in Australia and Germany. These newfound groups will help expand our abilities to reach and support more patients in the autoinflammatory community.

The progress in autoinflammatory diagnosis and treatment continues to be challenging. There is still a lack of specialists globally with sufficient knowledge and experience to recognize these diseases in both children and adults. As many of you are aware from your own patient experience, without diagnosis, there is no treatment.

In an effort to educate healthcare professionals, we will continue to organize webinars and social media campaigns. We look forward to our continued work, informing and educating all stakeholders to ensure that patients have a timely diagnosis, treatment, and a better quality of life.

As a mother of an autoinflammatory child and as a patient myself, I firmly believe the work the organization does continues to be vitally important, and I appreciate the continued support from our global patient community. Thank you for helping us make this September awareness campaign 2023 visible by sharing our magazine with friends and family.

Malena Vetterli

Founder & Executive Director

September is World Autoinflammatory Awareness Month - Endorsed by the European Reference Network RITA

This year at the FMF & AID Summit of all European autoinflammatory affiliates, it was decided unanimously that this year's theme should be patient access to genetic testing. While clinical symptoms should always lead to diagnosis and treatment plans, genetic testing can be a useful and sometimes lifesaving tool. There are more than 40 known autoinflammatory diseases.

Despite the advances in genomic sequencing, patients continue to struggle and often cannot access genetic testing. These issues can vary by country, ranging from providers unwilling to order genetic testing to genetic testing facilities being unavailable, a lack of insurance coverage, or denial per health insurance policy. Symptomatic patients who have undergone extensive genetic testing where only a heterozygous single mutation was found are often dismissed based on the genetic findings, thus being medically ignored.

uSAID classification presents another genetic hurdle for autoinflammatory patients. uSAID stands for undifferentiated systemic autoinflammatory disease, which comprises approximately 60% of the known cases where no genetic mutation could be found to explain symptom presentation. It is important for health professionals to understand that no genetic findings in a symptomatic patient should equal a uSAID diagnosis to ensure that there is not a lack of care or a psychosomatic label attributed.

It is important to understand that genetic variants are still being studied in these diseases, and not enough information is available to confirm absolutely if a mutation is pathogenic or not. Moreover, there is little consensus across various databases that a mutation's status is always the same.

On the other end of the spectrum, there are a few autoinflammatory diseases that are not yet recognized and have no treatment code; however, they should still be treated as uSAID. This includes Yao syndrome (NOD2) and the more recently discovered autoinflammatory diseases that remain only known in the medical literature. There are many genetic issues that patients face globally as we continue to advocate for timely testing, diagnosis, and treatment.

FMF & AID European Affiliates Advocacy Summit 2023

FMF & AID Global Association recently organized and held the FMF & AID European Affiliates Advocacy Summit in Budapest, Hungary. Fourteen Patient Associations and patient advocates from nine European countries (Spain, France, Germany, Italy, UK, Georgia, Switzerland, Greece, Turkey, etc) participated in the 2-day summit where autoinflammatory topics of common interest were discussed.

We are thankful to Novartis for sponsoring this important meeting and appreciate their dedication to supporting the FMF & AID Global Association and its affiliates, to ensure that patient voices are recognized.

The group had successful sessions and look forward to advocating further for patients globally. The main outcomes and results from this meeting are found here below.

We are very appreciative of the patient representatives and advocates traveling to Budapest to meet in person. Our group had a great experience collaborating and sharing ideas together. We look forward to meeting again.

Photo credit: Novartis

Key findings

- Collaboration with patient organizations at a local level and in native languages helps the global autoinflammatory community better understand and address issues faced by patients
- Autoinflammatory patients need better clinical diagnosis (based on symptoms), access to genetic testing, and appropriate and timely treatments
- Clear transition processes and implementation from pediatric to adult care must be established globally
- Acute phase reactant testing is not a single comprehensive way to diagnose autoinflammatory diseases

Patient challenges at the local level

- Patient members are often unable to support the association due to health and financial constraints
- Adult patients are often excluded, as the majority of patient organizations are pediatric-focused
- Patients are required to travel long distances to receive care, as there are few expert physicians treating autoinflammatory diseases
- If patient's disease is not recognized nationally, social services and disabilities may be inaccessible
- 60% of autoinflammatory patients are genetically diagnosed with uSAID (undifferentiated systemic autoinflammatory disease)
- Patients' autoinflammatory symptoms often do not correlate with genetic findings
- Many patients are unable to receive IL-1 biologic treatments due to their unavailability in their country

Photo credit: Malena Vetterli

Treatment and dosing issues in autoinflammatory patients

- There is a lack of experience and knowledge in treating autoinflammatory patients with biological medication
- Treatment escalation with biologics is required but not executed due to lack of knowledge
- Biological medication dosing and treatment adjustments should be based on the patient's disease activity
- There is not enough expertise on the use of combination therapies
- The halting of biologic medications during COVID, sepsis, infection, and pregnancy is incorrect based on the medical data

Common issues reported by Patient Organizations

- Patient-reported flares should not be dismissed if CRP, ESR and SAA are normal
- Positive genetic findings and elevated lab markers are often required to receive biological treatment
- Treated patients may have breakthrough symptoms or flares requiring additional medication, i.e., steroids, pain medications, etc.
- Illnesses may be overlooked in treated patients, but they may be infectious (bacterial), and timely antibiotic treatment may be necessary

Photo credit: Malena Vetterli

Announcement – Establishment of new FMF & AID associations

The FMF & AID Global Association is glad to announce the registration of its sister organizations.

FMF & AID Australian Association

Our Australian branch was officially registered at the beginning of the year. It is being led by Sharon Kensell with the support of an astonishing team of local patients, parents, and caregivers.

In August 2022, FMF & AID Global Association was invited by Prof. Seth Masters and the Walter and Eliza Hall Institute (WEHI) to attend the 2nd Australian Autoinflammatory Disease Symposium and present on the patient experience in Australia. Despite her own health issues, Sharon Kensell traveled to Melbourne and delivered a fantastic presentation! Thank you, Sharon, for your great representation on behalf of our Australian patient community. We were also invited to submit an abstract that was accepted and published as part of the brochure that was distributed at the symposium.

Sharon Kensell, Executive Support Officer

FMF u. AIE Deutschland e. V.

In June 2023, the German branch of the FMF & AID Global Association was founded and is currently being registered. Süreyya Der will be leading the association. She is not only a patient advocate but also suffers from FMF and was not diagnosed until adulthood. She strongly supports disease awareness for FMF and other autoinflammatory diseases among patients and medical professionals practicing in Germany. Additionally, she believes that daycare facilities, schools/educational institutions, and employers need to be educated on how autoinflammatory diseases impact students and workers, thus empowering German organizations and companies to play a role in supporting all ages of patients. Our goal is to collaborate with specialty centers throughout Germany and create a medical network for patient referrals. Finally, we hope to inform the German public and other German-speaking countries about autoinflammatory diseases to increase acceptance and awareness.

FMF & AID Australia

FMF u. AIE Deutschland e. V.

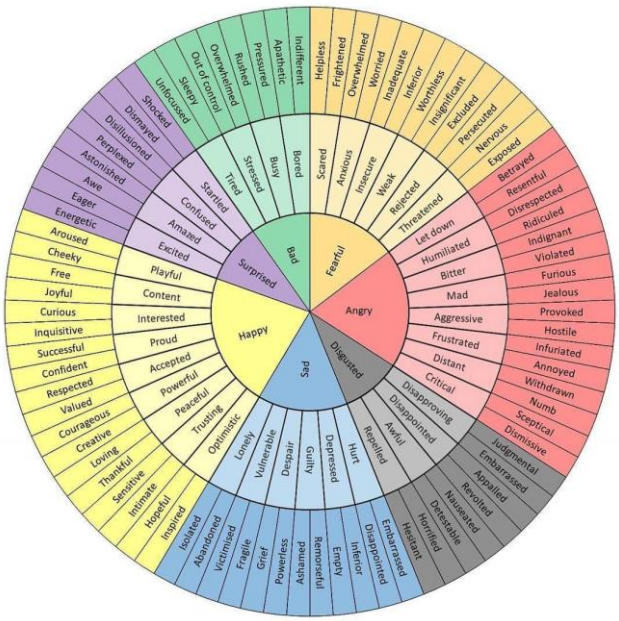
Zoom support group for children with autoinflammatory diseases

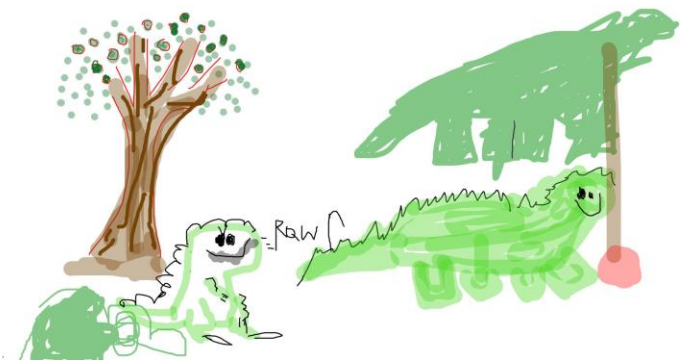
For the third year in a row, the FMF & AID Global Association has been able to offer free psychological support sessions in English to children with autoinflammatory diseases. The sessions are moderated by Karin Purugganan, Counselor and therapist from wonderologie.com, specialized in helping children with rare and chronic diseases.

The kids love her and cannot wait until the next session. According to Karin, when dealing with children, it is crucial to earn their trust but also to be relevant. The kids love to share not only what interests them, but also their worries, fears, etc., and we provide a safe platform for them to share what is important to them.

In a recent session, the children were asked what was going on in everyone's lives. Since we have children joining from multiple countries, some reported that they had already completed school were out for summer break, while others still had weeks of classes remaining. After getting an update, Karin presented two topics: the first is using a feelings-wheel to help the kids better understand their emotions, and the second is tapping or EFT used to reduce anxiety and stress. This exercise sends signals to the brain and body, restoring energy. Using this technique can be helpful before a doctor's appointment, performances, or sporting events. At the end of the session, children drew pictures of dinosaurs (see drawing).

Other session topics have included communication and response with others regarding rare disease illness, managing school and activities during a flare, coping with anger and frustration, etc. These monthly sessions have received excellent feedback, and we will continue to provide this special type of support to our pediatric community.





September 2023 Theme: Access to genetic testing

Autoinflammatory diseases are rare genetic inborn errors of immunity and can be monogenic or polygenic. Monogenic autoinflammatory diseases arise from a single gene mutation and cause diseases including FMF, CAPS, TRAPS, HIDS, Yao syndrome, Blau's, and others. Polygenic autoinflammatory diseases are complex, multifactorial, of unknown etiology, and characterized by dysregulation of the innate immune system. These include Behçet's syndrome, idiopathic recurrent pericarditis (IRP), adult-onset Still's disease, Kawasaki disease, and others.

Diagnosis for both groups of diseases should be based on clinical manifestations, medical suspicion, family history, laboratory findings, and genetic testing. While genetic analysis and identification of pathogenic mutations may be pivotal for diagnosing monogenic diseases, it is important to recognize that in 60% of cases, no gene variant(s) may be found. If so, the patient should be diagnosed as uSAID (undifferentiated systemic autoinflammatory disease) and treated in a timely and appropriate manner.

Another factor impacting diagnosis using genetic screening occurs when patients present in a heterozygous fashion with a single variant of the gene inherited. It is important to know that homozygosity (carrying two of the same inherited mutations) is not necessary for a patient to present with disease symptoms. The same rule applies when patients with compounded heterozygous mutations (multiple single mutations found across monogenic genes) also present with disease-like symptoms.

Further issues for patients arise when genetic results indicate they carry VUS (variants of uncertain significance), benign variants (not known to be disease-causing), unclassified variants (unresearched mutations), or significant mutations that are not considered autoinflammatory-related. Patients may present with a variety of these scenarios listed; however, if symptomatic, treatment for symptoms must be undertaken despite genetic analysis and results.

We have identified other issues, including:

- Genetic access may be variable depending on the country in which the patient resides
- Genetic testing may not provide any answers, leaving the patient with a uSAID diagnosis
- Benign variants, may in fact, be contributing to the patient’s symptoms
- Phenotype-genotype correlation should be considered by physicians
- Family members exhibiting similar symptoms should be genetically tested
- Lack of newborn screening in families with known AID is problematic
- Classification of variant pathogenicity may be different among populations

We advocate for all patients to receive genetic testing and access to their results.

Young Ambassadors Program

The FMF & AID Global Association is searching for young ambassadors between the ages of 15 and 35 who would like to be actively engaged in activities such as sharing lived-disease experiences, growing our online communities across several languages, and ensuring that patients' voices are heard.

There are many autoinflammatory patients on social media, raising awareness. However, if these individual advocates joined our efforts, we would have more strength as a group to make our community stronger.

Ambassadors are individuals who suffer from autoinflammatory diseases themselves and want to have an active role within the community. These representatives should be willing to speak and share their patient journey at events and engage with other stakeholders.

We encourage patients or their caregivers who are interested in taking on this role to contact us.

Please note that if you are a teenager, you will need your parent's written consent to participate in this program. For more information, please contact info@fmfandaid.org.

Mackenzie Cabrera, USA
Mother with FCAS child

Photo credit: Mackenzie Cabrera
advocating for patients

The Power of Good Communication by Marie Jolie Gossing

A healthy and happy life as a rheumatic disease patient depends on various factors, including good communication between the patient and doctor, as it plays an enormously important role. This topic triggers strong emotions in patients, such as anger, despair, fear, and gratitude, which also applies to me and is the reason why I decided to write this article. So, how has the good communication between my doctor and me helped make my life so much better? In order to do justice to this question, I would first like to share my negative experiences.

I spent most of my childhood and adolescence in doctors' offices, ambulances, and hospitals while trying to figure out what was wrong with me. Every time an appointment came up, I was full of hope for answers, help, and understanding. However, these wishes were all unfulfilled as a child and as a teenager. The appointments were always the same: short and disappointing.

Over the years, I have been asked the same questions by different specialists. At some point, I had memorized the answers because I was so familiar with the same questions, as they were all irrelevant: "Are you stressed? Do you like going to school? Is everything okay in your family? Do you have friends? (...) No, I don't have any more stress than usual. Yes, I like going to school, even if I rarely make it."

"I can see my friends there. I often feel too bad to see them in my spare time. Everything is ok in my family." "Well, stress is subjective, and so is pain. If you go to school so little, then you certainly have a lot of fears and little connection. It hits the psyche! Yes, well, now that your mother is there too, what do you want to say as a child?" It did not matter what I said, as the doctors never really listened to me, nor did they take me seriously.

I think the last few lines need no further explanation and speak for themselves. After each of these appointments, I cried a lot, felt insecure, and no longer trusted the doctors. You ask yourself, "Why am I not being helped?" "Why don't they believe me?" "Why do I have to keep living in pain?" Such a conversation cannot lead to any improvement in physical or mental health.

Photo provided by Marie Jolie Gossing

It is very important to me that my doctor and I can communicate openly and on an equal footing. Only if I, as a patient, can clearly express my symptoms, worries, and fears can the doctor make a diagnosis, find a treatment, and try to take my concerns away. As a patient, I need to feel comfortable and taken seriously. So, both parties are essential for good communication. It is give and take. However, the doctor must create a basis of trust in the beginning; otherwise, there is no real cooperation. The openness also helps in terms of clarification, especially if a doctor explains my illness and treatment options to me, then many insecurities are removed.

I can consider myself very fortunate that my relationship with my current rheumatologist is extremely good. My first appointment was different from all other appointments beforehand. I was welcomed with a friendly smile, an open ear, a large portion of understanding, and a lot of time. My doctor didn't ask the same questions as previous doctors. His questions were much deeper. For the first time, they were based on my actual answers. I immediately had the feeling that I was being taken seriously. I could and will continue to talk to him openly about all of my issues, fears, and worries. Going forward, my extremely negative relationship with doctors, which consisted of fear and distrust, has changed to a more positive one.

However, I have had to learn what it is actually like to be listened to by a medical professional and trust their judgment. It is great that I can tell the doctor that a flare was triggered by stress, and he takes that seriously. Due to my enormous confidence in my current rheumatologist and based on our excellent communication, I have been able to obtain suitable treatment.

In my case, having good doctor-patient communication has meant LIFE, and to me, this means walking, dancing, breathing, moving, meeting friends, and being able to EXPERIENCE many everyday things. The great relationship with my rheumatologist means a future for me. The opportunity to realize my dream of studying medicine is now physically possible. Before, I wanted to study medicine because of my bad experiences with doctors and to ensure that I did not become like the others. My goal today is now a positive one thanks to my excellent role model, my current doctor.

So how can this excellent patient-doctor communication become the standard in the future? In my view, several factors need to change to benefit both parties. We need more knowledgeable doctors who have ample time to manage each patient's case. We need more universities and teachers to train clinicians, and they should be taught how to communicate and interact with patients during their studies.

While we have enough students wanting to study medicine, the general working conditions of doctors must fundamentally be improved so that more medical students will enter patient practice rather than go abroad or conduct research.

As a patient, one is very angry if not taken seriously, but it's often due to doctors' ignorance. I do not mean this in a bad way, as the spectrum of rheumatic diseases is enormous and not every doctor can know everything. Therefore, regular specialist training would be great to expand and update the knowledge of autoinflammatory diseases. Finally, it is an advantage if the doctor knows how to communicate and listen. For me, it is a matter close to my heart to say that the social skills of physicians are more important to us as patients, than an outstanding diploma.

Will anything change in the system? Hopefully, yes. I wish all patients health, strength, and perseverance.

September World Autoinflammatory Awareness Month

1 There are **40+** autoinflammatory diseases

2 **14 years** until adult diagnosis

3 years until pediatric diagnosis

3 Individuals of **all ethnicities** and ages can be affected

4 **Common symptoms** include:

fevers, rashes, joint and muscle pain/swelling, abdominal pain, headache, fatigue, etc

Signs of Medical Gaslighting

Service Dogs

In last year's newsletter, one of our members, Brooke Hightower-Foster, wrote about her experience having a Weimaraner dog and training it to become a service dog able to detect fevers and flares in her daughters before they even happen (3–4 days before onset).This ability to train a dog and provide an early opportunity for parents to make necessary medical accommodations before a flare happens is amazing.

After seeing the work and efforts made by our association to support the autoinflammatory patient community, Brooke enabled an opportunity for the Weimaraner breeder, from whom she received her service dog, to donate further. The FMF & AID Global Association was then contacted by Melissa Miller, the breeder from Miller's Kennel (millerskennel.com) in Illinois, informing us that she had decided to donate two puppies to our cause! This was a great opportunity for two children and their families to benefit from having a service dog.

Photo credit: Melissa Miller

After conducting numerous interviews with interested parents and ensuring the families met the needs of the Weimaraner puppies, the first puppy went to Charlotte, a 6-year-old girl diagnosed with CAPS, and the second to Tobi, a 9-year-old boy diagnosed with PFAPA.

Facts about Weimaraners

- They are large-sized dogs weighing from 55 to 90 pounds (25 to 41kg).
- They are very energetic and require a high level of physical activity.
- They have strong scenting abilities.
- They do not do well if left alone for long periods.
- They are fast dogs and run up to 35 miles/56km per hour.
- They are one of the most loyal dog breeds out there.
- They are affectionate, friendly, courageous, and obedient dogs.
- Training and socialization are crucial, and these dogs can be trained for human service.

Webinars organized by FMF & AID and ImmunAID

In an effort to inform and educate the autoinflammatory patient groups as well as providing the latest information to medical professionals, the FMF & AID Global Association has hosted a variety of successful webinars, some in partnership with ImmunAID. Topics presented included PFAPA, FMF, AOSD, and Behçet's disease. These online webinars were offered in various languages to ensure our affected communities had equal access to the information. We are planning for future webinars, and we will inform you of dates and times in due course.

European Reference Network RITA – 2023 Annual Meeting in Utrecht

The European Reference Network RITA (Rare Immunodeficiency, Autoinflammatory and Autoimmune Disease Network) General Assembly takes place every year, and they hosted their 2023 Annual Meeting in Utrecht, Netherlands, on June 15–17. Attendees included the RITA Board (coordinator, network manager, coordination team, stream leads, and four patient representatives), healthcare professionals, scientific societies, heads, and patient association representatives, which also included Malena Vetterli, Executive Director, FMF & AID Global Association, patient lead for the autoinflammatory arm, and RIPAG Chair.

The participants involved in RITA initiatives came together for this yearly program to discuss past work completed and to plan for future activities across the various working groups, including research and molecular testing, patient journeys, clinical guidelines, IT/e-health, transition care, education, and registries.

Apart from these plenary meetings, there were parallel sessions conducted by each of the stream clinician leads: autoimmune diseases, Autoinflammatory diseases, primary immunodeficiency, and pediatric rheumatology.

Patient representative leads for each of the disease arms presented the outcomes of the patient journeys. The autoinflammatory arm represented by Malena Vetterli provided the patient's journey on Familial Mediterranean Fever. The details reported were gathered from a variety of FMF patient focus groups organized in several languages across multiple countries. The next phase of reporting will include input from FMF specialists and healthcare providers. These patient journeys will be available in several languages through the RITA website and will contribute to disease awareness.

Photo credit: ERN RITA

Specialized Center for Periodic Systemic Autoinflammatory Diseases Erlangen (Germany) University Hospital by PD Dr. Jürgen Rech, Senior Physician & Head of the Special Consultation, Autoinflammation Clinic treating AOSD, FMF, Gout, etc.

Establishment

In 2021, due to the constant growth of our adult and pediatric clinics, it was considered sensible and necessary to formally increase the visibility of our special consultation hours.

On March 27th, 2023, the special center for periodic systemic autoinflammatory diseases was established.

Mr. PD Dr. Jürgen Rech, spokesman, University Clinic Erlangen, Rheumatology, together with Dr. Tobias Krickau, Deputy Spokesman, Children's and Youth Clinic, University Clinic Erlangen, Rheumatology, with the active support of clinic management, Prof. Dr. univ. Georg Schett, Medical Clinic 3, Rheumatology & Immunology, University Clinic Erlangen, as well as Prof. Dr. Joachim Wölfle, Children's and Youth Clinic, University Clinic Erlangen, the Center for Rare Diseases (Center A) with Prof. Dr. Beate Winner, the colleagues and respective speakers of the other special centers (B-centres), the Dean Prof. Dr. Markus F. Neurath, the Medical Directorate Prof. Dr. Dr. h.c. H. Iro, Commercial Directorate Dr. Albrecht Bender, was officially decided.

Collaboration

The FMF & AID Global Association (CEO Malena Vetterli) was contacted soon after the opening of the special outpatient clinic. Contact with her confirmed that there was a great need for doctors to focus on autoinflammation and that patients still had to wait a long time for a diagnosis.

Our new patients were surprised and reassured to realize that they would receive appointments with the same diagnostic and treating physician, thus ensuring consistency of patient care throughout the treatment process.

Services

The Erlangen Center offers patients the opportunity to be advised, diagnosed, and receive long-term therapeutic care. We also welcome helping new patients who require a second opinion for their diagnosis or managed care. Since its establishment, the number of patients with autoinflammatory diseases served at our center has increased steadily and significantly. We also have patients from across Germany traveling to Erlangen and even patients from neighboring countries.

Projects

Scientific patient-focused projects with the FMF & AID Global Association and our national and international cooperation partners have already commenced or are currently in the planning process.

Topics of interest include

COVID-19 vaccination response in IL-1-treated AID patients, quality of life factors in AID patients during the pandemic, and other relevant issues will be addressed.

It is critical that the patient and doctor communities are both open to actively collaborating so that all physician and patient voices can join together and implement best practices within our center.

Aims

- Improve the early diagnosis of patients with rare periodic systemic autoinflammatory diseases
- Increase awareness of AID among doctors of all disciplines
- Train and educate students, doctors, patients, and other stakeholders

PD Dr. Jürgen Rech, Spokesman

Dr. Tobias Krickau, Deputy

ZSEER
Zentrum für Seltene Erkrankungen Erlangen

zpsae.uk-erlangen.de

Internal Medicine Center at the University Hospital of Erlangen, Germany
Photo credit: Michael Rabenstein

Book review: "Listen to Wisdom“ by Diana Szaragat

We share with the international community the Argentinean publication "Listen to Wisdom" - Rare Diseases: Strategies for Articulation of the Health System and Socio-sanitary Impact on the Finding of the Diagnosis, authored by César Agustín Crespi and Diana Szarazgat in collaboration with other professionals. The prologue by Dr. Ricardo Sánchez Peña, Director of the Artificial Pancreas Project of Argentina, states: "This is a necessary book given the current times, with greater access to previously unrecognized rights and the fight against discrimination. It addresses the area of rare and difficult-to-diagnose diseases. The conception of the text is focused on the patient, who undergoes the "diagnostic odyssey" with his wheelbarrow loaded with information, as well as the acknowledgement of the lack of specialists, centers, and adequate protocols. It also contemplates the absence of undergraduate courses, especially the need to have well-informed patients and the role of the "expert patient". The role of patient associations is included as part of the solution, and their objectives and support services to the community are highlighted. From this point of view, the patient is an active consultant and participates in the diagnosis, as long as the health care professional knows how to listen. The book goes on to discuss several topics related to rare disease education, diagnosis, and prevalence, among others. Over the last few years, the field of rare or orphan diseases has evolved from what constituted a limited group of unknown entities, academic orphans, research, and institutional interest towards a series of diseases that can be diagnosed and, in certain cases, treatable. However, given the great diversity and low frequency of each of the rare diseases, access to diagnosis is difficult. Rare diseases are those whose prevalence is less than or equal to one case in 2000 people. More than 7,000 entities are included, and when taken as a whole, they are much more frequent than is believed. In many cases, the disease will develop without being identified as such, depending on the information that is available and the ability to recognize it.

Many times, these diseases are misdiagnosed or not diagnosed at all, which leads to significant morbidity and mortality. Some of them have such a low individual prevalence that special and coordinated efforts are needed to detect and treat them.

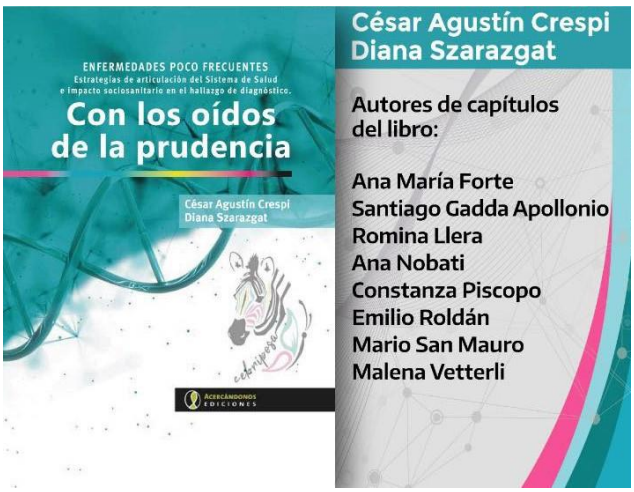


Photo credit: Editorial Acercándonos Cultura

Rare diseases can be overwhelming and sometimes lethal, which implies very important family and social burdens. It also entails a high consumption of health resources due to the demands on the health system.

These diseases are usually severe and put the lives of patients at risk if they are not diagnosed on time or treated properly. Life expectancy is reduced in two thirds of them; 30% of patients die before the age of 10 and 20% between the ages of 10 and 30. Most of the phenotypes compatible with prolonged survival are associated with disabilities, which limit access to schooling, employment, and deteriorate the quality of life.

For some of these rare diseases, there is currently an effective, specific treatment. For the rest, early diagnosis allows adequate family counseling and the establishment of supportive treatment; in both cases, it will be possible to significantly improve the quality of a patient's life, delay the progression of the disease, or sometimes even stop it.

Symptoms can start at any age, whether in childhood or adult life. Often, the first symptoms are common and are confused with those of more well-known and frequent diseases. They rarely have specific signs and are generally suspected when apparently unrelated symptoms are added or when the initial response to common treatments is not as expected.

In general, in the medical community, there is a lack of knowledge about this type of pathology, which is associated with an inevitable delay in diagnosis and, therefore, the establishment of inadequate treatments until an accurate diagnosis is reached. It is important to note that many rare diseases are compatible with a normal life if diagnosis and/or treatment arrive on time.

Many of those affected feel isolated, not knowing where or whom to turn to. This is due to the lack of specialists, protocols, and comprehensive centers, where patients can find a complete and comprehensive answer to their pathology.

This book can be found here. An English version will be available later this year.

Photo provided by Dr. César Crespi

Patient journey: Antonella (Ecuador) with NOMID/CINCA

My daughter Antonella was born with a rare disease called NOMID/CINCA, the most severe form of CAPS, which stands for "Cryopyrin Associated Periodic Syndrome.“

At birth, she had hives all over her body, and the doctors said it was an allergy. Then, at two weeks old, her right arm became paralyzed, and a month later, the same happened to her left leg. Since the doctors in the public health system could not figure out what was wrong with her, we had no choice but to consult various private doctors. The only medication they provided was corticosteroids to reduce the inflammation in her joints. She continues to be on this medication to control the inflammation, although she needs other drugs for long-term control of her disease. These private doctors requested countless tests, including MRIs, lumbar punctures, skin biopsies, and others. She has been hospitalized so many times that I have lost count.

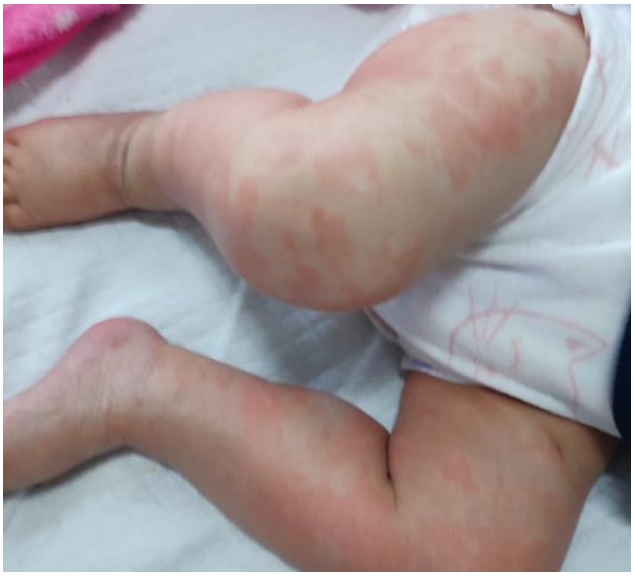


Photo provided by the parents

At 6 months old, one of her doctors suspected she could have a genetic disease, and her blood was sent to the United States for testing. The results were returned, and a diagnosis was made of NOMID/CINCA. Today, at 18 months old, we are still waiting for the local hospital in Ecuador to provide us with the much-needed biological medication used to treat her disease.

Due to the systemic inflammation that affects her entire little body, she barely grows, and at her age, she has only the size of a 7-month-old. In addition, she endures severe daily pain in her joints, head, and throughout her body. She does not talk; she just cries, and even her movements are limited.

Photo provided by the parents

Unfortunately, biologics (IL-1) are infrequently available across many countries in South America due to their high cost, leaving children like my daughter untreated and with a devastating prognosis. We have knocked on many doors and looked everywhere for help. Additionally, we had to take legal action and file a lawsuit to force our health system to purchase, import, and provide biologic medication, as there is no supply chain or reimbursement in place to obtain these specialty drugs. Although we won our lawsuit last month, we continue to wait for the hospital to provide our daughter with the lifesaving treatment that she desperately requires.

From the beginning, the FMF & AID Global Association has provided us with great support and advice throughout this terrible and traumatic situation, and it has been a relief that they know how to navigate the process of receiving care. They have helped us by contacting doctors, advocating for access to biological medications, and providing us with current disease information. I don't know what my wife and I would have done without their help. We thank the FMF & AID Global Association for all their efforts dealing with our family and our daughter.

Photo provided by the parents

Photo provided by the parents

Patient journey: Josiah (USA) with FCAS

Hello, my name is Makenzie, and I am the mother of Josiah, who has Familial Cold Autoinflammatory Syndrome (FCAS). Josiah is 2 years old and has had symptoms since birth, but he was not diagnosed until a few months before his second birthday.

From day one, Josiah was not the typical child. He was born very jaundiced, had fetal tachycardia, and had a weird patchy rash all over his face and body. By 11 days old, we had to return to the P-ICU due to seizures, abnormal blood work, and CSF (cerebrospinal fluid). Josiah presented with three different infections, for which we spent almost a full month in the hospital. As time went by, Josiah did not get any better. He kept developing chronic infections and continued to have abnormal blood work, lymphadenopathy, red eyes, extreme pain caused by cold weather, delayed milestones, and many other symptoms.

When he was a year old, they deemed him a medical mystery. Nobody knew what was wrong with my baby. A few months later, Josiah was diagnosed with generalized "periodic fever syndrome (uSAID)" and "Raynaud's syndrome", which is where feet and hands turn blue, white, or red and is extremely painful. Due to the lack of knowledge, we then saw an abundance of doctors who ran multiple tests—so many that we lost count!

Finally, we saw a rheumatologist and an immunologist together. They suggested maybe he suffered from a genetic disorder and ordered genetic testing for autoinflammatory diseases since other treatments (Tylenol, Ibuprofen, and steroids) did not work. We did genetic testing, and a month later, we received a call informing us that our son had a rare autoinflammatory disease called FCAS.

By the time we received the diagnosis for Josiah, he was in a bad state. He could not be outside for more than 5–8 minutes due to extreme pain in his legs, which were swollen and could not bear weight. He continued to have a whole-body rash with the fevers every 10 days. He was immediately started on an IL-1 biological injection, and to this day, the doctors are still adjusting the dosage and timing.

Photo provided by the parents

Today, Josiah is a fun-loving, high-spirited, and strong two-year-old who went through a great deal and is the reason why I am going for my nursing degree with a specialization in rheumatology with a focus on children and adults with autoinflammatory diseases.

Once the diagnosis was made, we looked for support. While FCAS may be rare, we have found help and support with the FMF & AID Global Association. The association makes patients and the community feel they are never alone in managing their disease. The amount of support this non-profit provides is absolutely wonderful. From the support groups for families, adults, and kids to paying for patients' medications, travel, and other medical expenses as needed. They are truly lifesaving for some families. Before I knew about the association, I felt alone and did not know what to expect when managing my son's disease. This group makes it feel like one big hug is being extended to patients and families suffering from autoinflammatory diseases.

Photo provided by the parents

FCAS

Familial Cold Autoinflammatory Syndrome (FCAS) is the mildest form of Cryopyrin-Associated Periodic Syndromes (CAPS). Cold exposure is the main trigger. Amyloidosis is a rare complication that occurs in less than 2% of patients.

Gene: NLRP3

Onset: Typically begins in infancy or may present later in life.

Flares: Acute flares lasting up to one day

Symptoms: FCAS is characterized by urticaria-like skin rash, fever, chills, nausea, and joint pain. Other symptoms include: muscle pain, headache, drowsiness, extreme thirst, and conjunctivitis.

Treatments: Interleukin 1 (IL-1) inhibitors (i.e. Ilaris and Kineret) are now the recommended standard of care for FCAS. For breakthrough flares, non-steroidal anti-inflammatory drugs and glucocorticosteroids (prednisone) are also prescribed.

Patient journey: Tobi (USA) with PFAPA

Over a year and a half ago, our son Tobi came home from school with a fever of 106.5°F / 41.4 °C. It was frighteningly high. He tested negative for every viral and bacterial test possible. Doctors could not determine the cause. Instead, we were told to "go home, wait, and just treat it like any fever with rest and a fever reducer". After three days of round-the-clock care, Tobi's fever finally broke. He got up from bed and was back to normal, like nothing had happened. Little did we know that this episode was just the beginning. The next month, the same symptoms presented, with the same response from the doctors. The next episodes repeated themselves monthly … you get it.

It took about 10 months to get a diagnosis of PFAPA. Having a diagnosis was calming; however, we knew that the flares were going to continue every 28 to 30 days for several years. PFAPA usually goes away on its own by adolescence, and Tobi was 7 at the time. A tonsillectomy is the only hope for some resolution. It resolves PFAPA in most cases, but not all. If removal is effective, it may reduce the frequency of flares, eliminate the fever, or create fever-less flares. With this PFAPA diagnosis and information, we had to find a new way of functioning.

Like every child affected by PFAPA, there are the obvious physical stressors. With each flare, Tobi would have three days of high fever, which was incredibly taxing to his young body and also caused weight loss from him not eating due to throat sores (a typical symptom of PFAPA). In addition, there are the social and psychological side effects that greatly detract from Tobi's quality of life. The fever and resulting lethargy made it impossible for him to go to school, attend soccer practice and games, play with his brothers, and focus on activities he enjoys.

For the first time ever, we contemplated getting a pet for companionship as we watched Tobi stranded in bed for days every month. Amazingly, as we were considering a support animal for him, FMF & AID announced that they were donating a support service puppy to a child with an autoinflammatory disease. The timing was perfect, and that is how Raya came to us. What a beautiful gift from FMF & AID and Melissa Miller, the breeder from Miller's Kennel (millerskennel.com).

Photo provided by the parents

She is not only Tobi's companion during the PFAPA flares and his hard times, but she is also part of creating a new story for him: a boy and his new best friend, relaxing and building happy memories together. On Tobi's good days, he is dutiful and responsible in caring for Raya. They enjoy playing games together (which Raya always wins). She brings Tobi and his brothers such joy. On the hard days, Raya is always by Tobi's side, and he very much appreciates her company. He even reads out loud to her and, surprisingly, she appears to like it.

While we do love having Raya, it has been a huge adjustment for all of us. We went from a pet-free home, to having a puppy with little preparation. It has been quite a learning curve for both the dog and our family.

Tobi is her primary attachment, and Raya is learning his habits, as well as how to use her senses to identify his different states of wellness. She also has had to learn how to behave properly within the house, and we are learning to teach her the rules most effectively. We hope to have Raya trained as a certified support dog in the future. Meanwhile, we are taking her to the required veterinary visits (for vaccines, spaying, etc.).

Lastly, there's an adjustment to having a Weimaraner as she is energetic, smart, and engaged, and requires daily exercise, connection, and stimulation.

Raya has been a welcome addition to our family. She has become not only Tobi's best friend, but the whole family's, too. In a time that had been clouded by fear and change, Raya has been a ray of sunshine, and we are very grateful for this wonderful gift.

Finally, throughout this learning process working with our new puppy, we have received outstanding support from the breeder Melissa Miller. She and her team know these dogs better and how to train them appropriately. It is fantastic that they are always available to address any of our concerns and have provided support, especially during our initial days of "puppy blues", when our family questioned what we had done adopting this puppy.

Photo provided by the parents

Patient journey: Maria Fernanda (El Salvador) with TRAPS

At the age of 17, after 8 years of fighting without knowing what my daughter suffered from, she was diagnosed with Tumour Necrosis Factor Receptor-Associated Periodic Syndrome, better known as TRAPS. TRAPS is a very rare autoinflammatory disease and has no cure. There are only approximately 1,000 cases known worldwide, and my daughter is the first TRAPS patient diagnosed in El Salvador.

Maria Fernanda suffers from recurring flares every 6 weeks, and each may last at least 3 to 5 days, but typically, they last between 2 and 4 weeks. Symptoms also present between flares. During flares, she presents with fevers, acute pain, abdominal distress (due to inflammation in the digestive tract), muscle pain and reddish disseminated skin eruptions occurring on her extremities. Additionally, she suffers from swelling around her eyes (periorbital edema) and in various areas of her body, including her heart muscle and kidneys. She also deals with joint pain, swollen lymph nodes, and canker sores. During flare-ups, she requires hospitalization and morphine to control the pain, as it is so severe. Luckily, Maria Fernanda has not developed Amyloidosis from her TRAPS, as many are at high risk for the abnormal protein build-up leading to kidney failure.

Unfortunately, the biological treatment that she needs (IL-1) is not available in El Salvador, but thanks to a specialized centre in the USA, they have donated the medication to my daughter. While we are very fortunate, the problem is that the medication only works to control the flares, but it is not dosed (daily) to prevent them. This means we have to wait for the flare to start and then give her 3-4 daily injections until the flare is better controlled. The monthly medication, which is really needed to control her TRAPS, is not provided due to its high cost.

To receive this medication, I am required to take Maria Fernanda to the United States every year to collect the medication and undergo laboratory testing. As a single mother, this means an impossible expense for me to cover, due to the high costs of purchasing plane tickets, paying for accommodations, and covering transportation and meals. Thanks to the associations medical aid program, they have reduced the financial burden and have provided unconditional support, as we are required to take this yearly trip and pick up this life-saving medication. I do not know what we would do without the help of the FMF & AID Global Association’s help, and we are infinitely grateful.

Photo provided by the parents

Patient journey: DeeDee (USA) with FMF

I was recently diagnosed with Familial Mediterranean Fever (FMF) at the late age of 72. I have suffered with symptoms since I was four years old, and not one doctor ever thought to test me for any type of disease.

As a child, the doctors told my Mom, "well, sometimes kids spike fevers and experience growing pains"!! As an adult, I was told the following by many different doctors: "it is your own fault"; "you are fat " (I weighed 138 lbs/62kg); "I think you have Munchausen's"; "you are bipolar and when manic, you are fine, but when you are depressed, you imagine your body is attacking itself". My favourite: "you are lying, there is nothing wrong with you, and see a shrink.“ An emergency room doctor who saw my case believed I should be tested for FMF.

Unfortunately, this was after I had emergency surgery for a double knot in my intestines, caused by two solid weeks of intense violent vomiting. Imagine my relief, and I finally knew what was wrong with me. This disease likely impacted my poor mother, who was hospitalized every year for acute peritonitis! I am so lucky that genetic testing now exists, since all my labs were always excellent! I assume these negative results are why all my doctors believed I was lying.

I haven't had a major flare in 1.5 years now due to the medication (I'm on Colchicine) and I'm delighted to know that no other generation will have to go an entire lifetime with FMF as a mystery illness. Now we have genetic testing, and we have medications to manage FMF. I sincerely hope my generation is the very last one to suffer from FMF (which I have noticed increases in frequency and intensity as we age - if we don't treat it).

The FMF support group from the FMF & AID Global Association is a Godsend. Special and sincere thanks to the association for the information you all share with the patient community, and for making me understand that having this disease is not my fault, and that I am no longer alone having FMF. The group saved my sanity and gave me the support I needed.

Colchicine

- Users: Familial Mediterranean Fever, Behçet’s disease, PFAPA, etc.
- Treats: inflammation, amyloidosis & pain.
- Age range: infants to elders, all ages.
- Pregnancy: safe for pregnant women.
- Brands: NOT all brands are efficacious for all patients.
- Switching: brand change may be more tolerable & effective.
- Inhibits: tubulin, neutrophils, lysosomes.
- Don'ts: do NOT take with grapefruit & certain antibiotics, etc.
- Treatment: daily use and NOT to be stopped in FMF cases

Patient journey: Zaid (Chile) with TRAPS

I am Elsa, a 43-year-old mother of two beautiful children. However, my son Zaid, a 16-year-old young man who, despite how difficult his medical problems have been, smiles and shines like few others. He is the one who is afflicted.

Zaid, at only 7 days old, showed symptoms alerting us that something was wrong and that is when we began this odyssey. Due to my having to spend days and nights at the hospital with him, I lost my job and missed out on much of my daughter's childhood and adolescence. Over the years, I felt my strength faltering, and my despair of not knowing what my son had weighed heavily on my heart. I would see myself in a cold hospital so many times and heard a thousand diagnoses in my head. They would blame me for negligence and looked for a culprit but couldn't find one. I felt so many times that Zaid's life was going to end, and only asked for opportunity. Finally, at the age of 5 and through genetic study, we obtained the diagnosis: TRAPS (TNFRSF1A gene), and autoinflammatory disease, thanks to a test carried out in Barcelona, Spain.

While grateful for a disease name, we began our visit into an unknown world, having no tools to navigate what was being presented. We tried to accept our reality, of which we did not expect. This diagnostic journey was lonely and confusing, and sadly, with time many families, and friends abandoned us. As Zaid grew older, his flares became more frequent and intense, and often with new symptoms. Seeing the deterioration, fatigue, trembling body, and pain of your beloved child is frustrating and overwhelming. TRAPS has stolen Zaid's childhood and adolescent moments and is stealing my life. His condition today is complex, as he is on 2 biological medications and 22 pills a day. Additionally, he must follow a very strict diet since his kidneys and liver are already showing signs of deterioration due to his amyloid build up. Finding out that he has Amyloidosis was shocking and upsetting. I blamed myself and no matter how much we gave, this monster would continue to advance. Despite my broken heart, I could not cry as I had to be strong for my son, who was sitting by my side. I then looked at him, smiled and promised him that everything would be fine, although inside I felt that I was falling apart.

Photo provided by the parents

Fortunately, we have a magnificent team of professionals who are always aware of Zaid's continuing health issues.

Zaid’s doctor fights every month to get the subsidy for his biological treatments, since in Chile, rare or orphan diseases are not covered in the general health system. While we are so lucky to have these excellent health providers, there is much fatigue involved in the endless travel to see these specialists. In many regions, including where we live, we do not have access to the same specialized care as it is only available to those living in the capital of Santiago de Chile.

At times, I feel that if a patient is not bedridden or in a wheelchair, people do not believe in the pain they carry, and how disabling illness can be. We also have anguish with the abandonment of the State, the ignorance and little education of society, and the little empathy of some health professionals in the face of an unknown diagnosis. Begging to survive without having financial means is unfair, and the goodwill of the community, is not enough to afford million-dollar treatments.

There are no laws to protect us, nor are there resources for public health; thus, private health insurance is our only option. Sadly, we do not have the money needed to cover the monthly consultations and I fear the day will come when we will be unable to continue with his care and lose the progress we have made. It is a tremendous problem, there are no autoinflammatory specialists in our public health system.

The price of caring for a rare disease patient is also very high, and not just in financial terms. As my son’s primary caregiver, I suffer from physical and emotional exhaustion, combined with the constant fear of not knowing what awaits us tomorrow. "Surviving is tiring, consuming and slowly kills you". When HEALTH is a privilege and not a right, you often feel that you or your sick loved one will not be able to continue.

In recent months, we have received financial and emotional support from the FMF & AID Global Association, which has allowed us to pay for medical tests and consultations. I never imagined receiving help from so far away, and this has given me huge peace of mind, knowing that a global patient organization can offer assistance in many different countries. Their help has been priceless and admirable, and they cannot imagine how important their role is for assisting sick patients and their families.

Photo provided by the parents

Patient journey: Emmet (USA) with Yao syndrome

Our Yao syndrome journey began in July 2020 when my sixteen-month-old son had his first fever. It took me three months to recognize that he was having recurring fevers. Once I realized this, I got online and began researching. I came across the FMF & AID Global Association website, and learned the power of symptom journaling, but most importantly, that we were not alone. I learned of the struggles that others faced and the importance of documenting everything before presenting this information to my son's doctor.

I then started writing down the dates, times, durations, and temperatures of his fevers, and any other symptoms I noticed. I even dug through my old text messages to get the dates and symptoms I had recorded. I wrote everything down for one year, and after gathering the data, I read through my journal and looked for patterns.

The first thing I noticed was that his fevers lasted 72 hours. Tylenol (acetaminophen and paracetamol) did not work, however, Motrin (Ibuprofen) did, which then led me to believe this was inflammation related. My son was too young to speak, so I had to watch his mannerisms. He often kicked his legs, touched his head, squinted his eyes, and had runny bowels. His breathing would become laboured, and he would develop red and blotchy patches all over his body. His fever would range from 104.5-105°F/40.2-40.5°C degrees.

His hands and feet would be ice-cold during the peak of his fever. After Motrin (Ibuprofen) took effect, he would have 2 ½-3 hours before the fever spike would happen again. Once his temperature dropped to 102 degrees, he usually ran around, laughed, ate, and acted normal. As he got older, he could communicate more about his symptoms. Before a flare, his head, eyes, and feet would hurt. I knew that the fever would hit within a day when he complained of those things. Sometimes there would be visible swelling on his knees and ankles. Other times, they just hurt. His flares were like clockwork and appeared every four to five weeks. After reading his journal, I knew I had enough data to take him in and get help and hopefully be taken seriously.

Photo provided by the parents

When I presented the journal to his doctor, I explained that it proved my son's fevers were not flu or cold-related. My son's doctor read through it, then looked me in the eye, shook the journal, and said, "If you didn't keep this journal, we wouldn't have known anything was wrong." He asked what I thought the problem was and where my son should be referred. I asked for Genetic and Rheumatology specialists. Thankfully, he gave both referrals.

The Rheumatologist was convinced my son had FMF or PFAPA. To our surprise, in November 2021, his genetic fever panel returned with a NOD2 mutation, 2798+158C, better known in medical terms as NOD2 IVS8+158. The Rheumatologist had never heard of this mutation and had no idea what diseases were related to this. After research on her part, she came back and said that my son met all the criteria for Yao Syndrome, formerly known as NOD2-associated autoinflammatory disease. The doctor was unaware of any paediatric research studies, or children diagnosed with Yao syndrome. She reached out to other Paediatric Rheumatologists to find help in treating him. He was started on oral steroids to be administered at the onset of flares until we could find a better option. The steroids worked for the first three months but were unreliable after. Either the medication eliminated the fevers with continued joint pain, or the pain would be alleviated, but the fever would still be present.

The Geneticist referred us to a Gastroenterologist and an eye specialist, but unfortunately, at that time, we moved halfway across the country, and had to start all over again.

Trying to re-establish care took a lot of effort. The children's hospital would only see him if a Paediatrician referred him. Paediatricians wouldn't take him as a patient due to the fact that Yao is not well known. Therefore, it took months of calling and going through the interview process with every local paediatrician. The story was always the same, "No, we won't take him as a patient." Finally, after three months and calling over twenty doctor's offices, we found one who would see my son! The referrals were made.

Photo provided by the parents

His new Rheumatologist set appointments with Gastroenterology and Eye specialists. Everything looked fine in these tests, but we still had to monitor him every six months. His Rheumatologist was attentive and upped the steroids, which helped relieve his symptoms. At every meeting, we would discuss if any research had been published regarding children with Yao's, unfortunately without any findings.

My son is now four years old and about to start preschool. After moving, his flares became sporadic, and eventually, they stopped altogether. He has not had a flare since October 2022.

Even though he has not had a fever since then, he still suffers. The joints in his legs hurt, and he requests leg stretches after being active for a few hours. His eyes are still sensitive to light. There are days when he will cover his head for hours to escape the light and times when his whole-body aches, but that hasn't slowed him down one bit. His fine motor skills, balancing on one leg or touching his finger to his thumb, are difficult for him. We have yet to determine if that is from Yao or something else. His doctor has continued to treat him with steroids and will change treatment once flares become consistent again. Through all this, he's still a typical four-year-old, who is obsessed with dinosaurs and making sure everyone laughs with his comedic skills, even on his rough days.

In the last three and a half years since starting our journey, the most important advice I can give is journaling. Write everything down, even if it is not seemingly related, it may be important later. There are still many unknowns for the future, but we have hope! Like my son's Geneticist says, "Give it ten more years, and every day, we are learning and studying. Ten years from now, you will start to get more answers, so have hope for the future."

Yao Syndrome

Yao Syndrome is an autoinflammatory disease causing episodes of fever and abnormal inflammation affecting body parts including the skin, joints, and gastrointestinal system.

Gene: NOD2

Onset: Typically, in early adulthood, but may start in childhood.

Flares: Lasting several days to weeks

Symptoms: Rash presents with reddened /inflamed areas usually on the face, chest, and back, joint pain, swelling of the ankles, legs and feet, abdominal pain, bloating, cramping, diarrhoea, headaches, dry eyes & mouth, canker sores, chest pain, swelling of lymph nodes.

Patient journey: Zsombor (Hungary) with HIDS

My son was born in 2019. His first year of life was normal except that he had a terrible reaction to vaccines. We did not know the reason back then, but right after his first birthday, he had the first flare which lasted for 14 days. He presented with a high fever, stomach pain, and a skin rash. Also, his iron and hemoglobin levels had dropped significantly so that he had to be hospitalized.

Initially, the doctors thought it was a virus, but after we went home, his symptoms kept recurring every 2 weeks. Every time, he had the same symptoms: severe abdominal pain, fever unresponsive to antipyretics, enlarged lymph nodes, and mouth ulcers.

Months later, after getting a vaccine, a flare was triggered, and my son had his first febrile seizure. He collapsed suddenly and stopped breathing! That night still haunts me. As scary as it was, little did I know that this would be the first step towards his diagnosis.

Zsombor had to be hospitalized yet again, but this time his doctors started to test him for a whole range of diseases. They told me that they were considering everything from gluten intolerance to leukaemia. The doctors even examined a sample of his bone marrow. One afternoon, a rheumatologist came in for an examination, and suggested, for the first time, that my son possibly had a periodic fever syndrome.

The doctor mentioned that genetic testing was an opportunity we should take, and we agreed to the blood test in June 2020. I was then told that the result could be expected by September.

Luckily, during the summer, my son felt better, and he started having feverless flares only. However, by September, his regular flares came back with a vengeance! More tests were done and elevated levels of mevalonic acid were found in his urine. My son was in so much pain, and I was furious, so I called the laboratory and requested the genetic results. I was told that they could not process my son's sample that year. It was 2020, we were in the middle of the world pandemic and laboratory resources were limited and reallocated.

Photo provided by the parents

I knew I had to figure out something and started looking online. I found the FMF & AID Global Association and one of their volunteers got back to me immediately and offered assistance. I was informed about genetic testing opportunities outside Europe and with the organization's help, we could accelerate the process. Within a few weeks, the results were returned, showing a single mutation in the MVK gene. At first, our doctor was doubtful if it was HIDS, but we searched for studies and publications, and finally, we got the green light for biological therapy.

Our life has changed since then. My son feels better, even though stomach pain became a daily issue. He still flares quite a lot; however, they are milder and most of them are triggered by viruses or vaccines. We treat his flares with steroids because insurance coverage for biologics is limited.

My son, at 4 years old, has already experienced so much pain, injections, and blood tests, more than I ever had in my entire life. As parents, we also tested ourselves and found out that I have the exact same mutation that my son has. I never had any symptoms, but every time he flares, I wish it were me instead of him.

We have our struggles explaining his disease to friends, the kindergarten teacher, workplace and even to family. I worry a lot regarding his future, education, and adult life.

My husband and I discussed having another baby but knowing of my mutation and the possibility of passing it on again, it was definitely not an easy decision, but we welcomed our second child three months ago. Our little daughter was tested immediately and luckily, she does not have the MVK mutation. I believe that she was born into our life with the purpose of taking care of her brother.

I am incredibly thankful to the FMF and AID Global Association, because with their help, we managed to get my son diagnosed within a year and they also treated my soul with kind words during the most difficult times.

Photo provided by the parents

Photo provided by the parents

Patient journey: Cláudia and Sofia (Brazil) with FCAS2

My name is Cláudia (51 years old), and I have FCAS 2 with a NLRP12 mutation. My childhood was very difficult because I lived in a freezing region of Brazil. My symptoms presented with swollen lymph nodes, lesions on my skin with topical burning sensations and redness. I had joint pain, fatigue, spasms, and spots on the skin that appeared for a few days or even hours.

My labs only showed elevated C-reactive protein, which the doctor said it could be to laboratory error. Other exams showed no findings, so they treated me as a rheumatic patient. I also suffered from depression (doctors believed it was psychosomatic), sleeping issues, and ADHD.

My diagnosis was made in 2015, after my daughter Sofia, who is now 14 years old, was diagnosed with the same FCAS2/NLRP12 mutation.

She had been misdiagnosed with Mastocytosis when she was only 4 months old. However, I knew that some of her symptoms were inconsistent with that disease.

As a newborn, she would have recurring fevers of 40 to 41°C/104-105.8°F degrees every month. As she grew, I noticed that she was in pain every time she tried to crawl or walk. Her joints were red and hot, and her eyes looked as if she had conjunctivitis.

She also had many canker sores in her mouth and on her private regions. She also suffered from fatigue and was a ticking time bomb in terms of her mood swings. Just like me, she also had sleeping issues and recently was diagnosed with ADHD.

Knowing that we had a name for our diseases brought such relief. Our days of questioning and wondering if these symptoms were in our head are over. I am grateful to the FMF & AID Global Association for allowing me to share our story with my suffering as a patient, as well as being a mother of a child who also has autoinflammatory disease.

As an adult, it is very sad and depressing that it took so long to get a diagnosis. It is also very frustrating to have self-doubt, thinking these symptoms were just psychological. Every time I saw a new doctor, it was the same remarks; "there is nothing wrong with you. It’s all in your head". This caused many emotional problems for me, and I am still learning to deal with the trauma.

Photo provided by the parents

In addition to the diagnosis, a turning point occurred after finding the association. With their support, I no longer feel alone, as I see stories and communicate with people who have the same difficulties as myself. This has given me hope and strength to face the disease, for both, myself, and my daughter. It is a shame that more doctors have not learned about all the symptoms patients with autoinflammatory diseases are burdened with.

It is wonderful to ask questions and confirm symptoms that the doctors dismiss and confirm, with other group members, that they have the same symptoms presenting in their disease.

Thanks to the FMF & AID Global Association for providing a great support system, and I appreciate the hard work to assist patients everywhere.

Photo provided by the parents

FCAS2

Familial cold autoinflammatory syndrome-2 (FCAS2), also called NLRP12-associated systemic autoinflammatory disease (NLRP12-AID), is a rare autoinflammatory disease. In some patients, cold exposure, physical exertion, fatigue and stress can be triggers.

Gene: NLRP12

Onset: variable, ranging from the first year of life to middle age.

Flares: Lasting from a few hours to several days.

Symptoms: FCAS2 is characterized by periodic fever, localized or generalized urticaria-like rash, musculoskeletal pain, headache. Other symptoms may include: joint pain, abdominal pain, sensorineural hearing loss, lymphadenopathy, and fatigue.

Patient journey: Anonymous (UK) with Idiopathic Recurrent Pericarditis

At some point in my 20s, while I was working out at the gym, I suddenly had muscle weakness, and felt unwell. This also occurred while I was walking upstairs, as I felt breathless and thought to myself, I must have caught a virus. Shortly afterwards, the chest pains and abdominal pain started, and I felt fatigued and ill. I had a laparoscopy done, but nothing was found. I saw my GP and was referred to a pulmonologist, who believed I had onset asthma and prescribed me an inhaler. He also ordered a chest x-ray to rule out blood clots in my lungs. Other tests were ordered, of which all were returned as clear and normal.

Without answers and a lot of concern, I used my private healthcare to see a cardiologist, who immediately booked me in for an echocardiogram the next day. An effusion was seen, and the doctor put me on a high dose of Ibuprofen. Unfortunately, it did not help, so he then prescribed me diclofenac and then secondary Naproxen.

I decided to do my own medication search and came across colchicine and its use for chest pain. I discussed it with my specialist at my next appointment, and while he was uncertain about its use, as he knew it as a treatment for gout, he fulfilled my request. I came away with a private prescription for colchicine. Sadly, I was not improving, and my fatigue became so incapacitating, I had to leave my job and applied for benefits. Luckily, I received my Blue disability badge.

I continued to have various tests, including MRI's and blood tests, to try and determine the cause of my symptoms. I saw another cardiologist, who was surprised I had not been on colchicine since my symptoms began. I was then diagnosed with chronic fatigue syndrome (CFS), in addition to the Pericarditis. Over the next few years, I sought help from two rheumatologists, an endocrinologist, a pulmonary specialist, and numerous alternative therapists, as well as trying all types of alternative remedies and therapies. My stomach was a mess from high doses of NSAIDs.

I had had two endoscopies with suspected ulcers and gastritis, despite being on PPIs (proton-pump inhibitor drugs).

My breakthrough came when I requested to see a physician specializing in autoinflammatory diseases. Since I was unable to travel, the doctor referred me to a specialist in my own city, who had experience treating these rare diseases. The specialist reviewed my files and then took a full medical history from birth. She reminded me that I had experienced chest pains in my twenties, of which they thought it was a blood clot in my lung, and that I had been put on warfarin and told to remain on my sofa and not move for several weeks. During that period, I had seen a chest specialist, who noted that one of my ribs was raised where it met the sternum and there was concern that I had Bornholm disease.

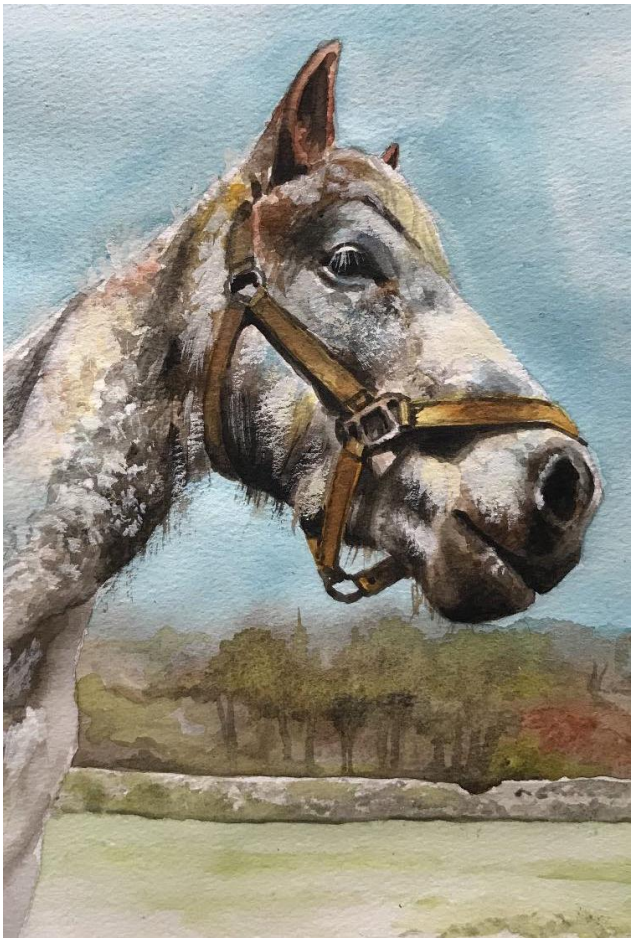
This incident passed fairly quickly and was forgotten. She also noted in my records that three years before my pericarditis diagnosis, I had suffered from lung pains, as I was under stress rehearsing for an amateur musical. My general practitioner, at the time, believed it was down to chest infections. In retrospect, the symptoms were most likely from pleurisy and possibly an early warning sign of onset pericarditis. This has continued as I get lung flares after exertion as well as heart/chest pain.

The new doctor tested me for RA, LUPUS, FMF and CAPS and surmised that I had autoinflammatory recurring pericarditis, which I probably had for an extended length of time, flaring especially during stressful activities. She increased my dose of colchicine to appropriate levels used in autoinflammatory diseases. I then stopped NSAIDs and started the long journey of weaning off PPIs. We discussed the use of biological medications, but I did not qualify as my CRP was normal, and NHS (UK), to this day, does not approve them for Pericarditis. The only treatment I am currently able to obtain is colchicine.

I am a lot better than I was, but still flare if I overexert myself and have to carefully manage my life to lessen the fatigue. My physical and social activities are less than they once were. I have lost many hobbies that I loved, including mountain biking, singing, dancing, and travelling.

I have adapted to less stressful activities and have started painting watercolours. I have lost a few friends throughout my medical journey, as they and my family do not really understand why I cannot do the same things that they can.

Life throws these curveballs, and as a patient, you have to make the most of what you are able to do. I hope my experience can help others with Pericarditis. I have learned a great deal about this disease and its impact on the patient's quality of life. I am hopeful that my case will help the doctors I have worked with, to better understand our challenges: medically, personally, and mentally, so that they will serve the pericarditis community more effectively in the future.



Watercolor painting by this patient

Patient journey: Charlotte (USA) with CAPS

Our daughter, Charlotte, has been showing discomfort and illness since birth. Being a relatively complex infant, soothing her was sometimes impossible, and doctors said, "It's just a virus," or "She's colicky and will grow out of it." Little did we know that she would inevitably get worse as she progressed through the years, leading us down an unimaginable medical path. She has blessed us by opening our eyes to many new perspectives and understanding the true meaning of resilience, bravery, compassion, and love, especially in the face of adversity.

Our saving grace was a nurse at our paediatrician, who had heard of different fever syndromes and referred us to infectious diseases and immunology, when Charlotte was 1.5 years old. Based on blood work and physical symptoms, she was initially diagnosed with PFAPA. However, her condition did not improve while taking colchicine for flares.

She became so ill that she would begin to hallucinate and have manic episodes, so bad that we would have to lie in bed with her, arms and legs wrapped around her, until she finally fell asleep from exhaustion and fever. She would also lose her ability to walk when sick.

One day during winter, we took our kids snowmobiling. Once inside and warmed up by the fire, Charlotte developed a face rash and spiked a fever of 103°F/39.4°C degrees within 30 minutes of being outdoors.

We knew then that this could not be PFAPA and requested genetic testing. Unfortunately, our doctor refused and was adamant that he was correct in his diagnosis. Desperate, we returned to the paediatrician and requested a rheumatology referral, which they obliged. We finally received the genetic results and found a mutation in the NLRP3 gene, and Charlotte was subsequently diagnosed with CAPS, which stands for Cryopyrin-associated periodic syndrome, a rare autoinflammatory disease.

Charlotte's disease has brought on many challenges for her and our family in her six years. As a family, we faced the struggles of daily injections of Kineret, which was mentally distressing for all involved, including her siblings. Not having the choice of what happens to your body was a heartbreaking scenario for us to witness and be a part of, as well as seeing a loved one who was so ill.

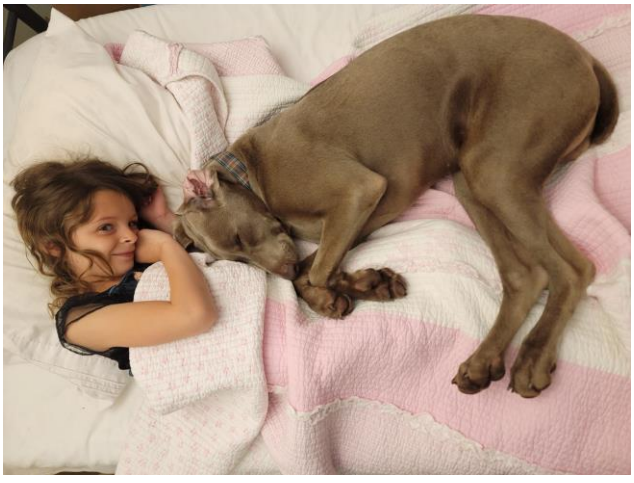
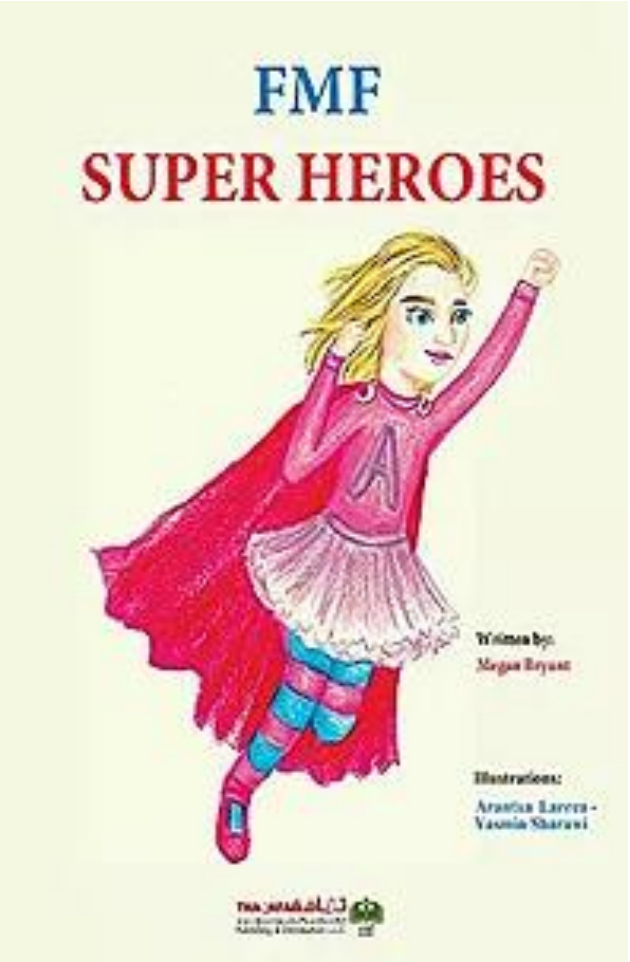


Photo provided by the parents

Two years into CAPS treatment, Charlotte now takes Ilaris and Kineret together as she still experiences breakthrough flares where she cannot walk properly. Other therapies include PT for her joint problems and for her pelvic floor after losing control of those muscles until 5.5 years old, soft ankle braces, counselling for mental health, as well as receiving extra time and help with school as she gets exhausted quickly. This past winter, she had many mobility problems and asked us if she would need a wheelchair. We have a wagon for her worst days, and we carry her a lot and help her dress.

Through all these struggles, we have become a part of the beautiful community of the FMF & AID Organization; they have given us a wealth of information, tools, and guidance through a community of people with rare diseases. They even have helped us receive a Weimaraner puppy from a line of service, guide, and therapy dogs through Miller's Kennel (millerskennel.com).

His name is Ghost, and he is working on becoming Charlotte's service dog. So far, he has been excelling at basic training and helping her cope with sensory overload and discomfort. It is incredible how well they already respond to one another. We are so thankful to no longer feel alone on our medical journey.



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: Ellen (Germany) with FMF

"Are you miserably warm today, too?" That was a question my relatives constantly asked me for as long as I can remember. As a kid, that question, often asked, was an absolute yes, even if "warm" was an understatement. As I later realized, it is actually not normal to feel like you are a heating system that has come to life, to which people could warm themselves in winter. It is not normal that I often felt miserable, as the feeling of warmth was a constant issue. It was surprising to realize that the reason for this warmth was due to a fever the whole time.

It is concerning that my symptoms may have been passed down through the generations, while being considered normal to have terrible stomach-aches, fevers, chest pains, and other symptoms. It is typical in my family to have recurring, flu-like symptoms for a few days, and then everything magically disappears.

My family comes from Italy, and has the gift of not recognizing fever, that impacts us all. A temperature above 38°C/100.4°F degrees was simply labelled "I am so hot today". This problem affected many in my family, but nobody ever felt the need to get to the bottom of what was medically wrong.

I have had these symptoms since childhood and cannot remember the last time I looked in the mirror and did not see crater-sized bags under my eyes.

Stomach pain and nausea were my constant companions, but as everyone used to say, "The child needs more fresh air" or "the child needs to eat more". These statements were ridiculous, and I did everything to cover up how bad I felt.

Some days I could hardly get out of bed because of the nausea. Often, I could not pay attention in class due to the exhaustion and I experienced the worst stomach cramps every four days.

I gradually began to doubt my self-perception, since there were always days when I felt fine. So of course, as soon as I complained of being unwell, I was dragged, at lightning speed, to the child psychologist and after a breakdown that found me in the hospital yet again, the comments continued; "We did various blood tests, everything is fine. Does your daughter have a lot of school stress at the moment?" No! For God's sake, the only stress I had on a daily basis was not being able to enjoy life in relation to my ever-stagnating health and the fact that I could not live normally.

Photo provided by J. Tschan

The good days became fewer and the bad days longer. I was passed from psychiatrist to psychiatrist, I was prescribed more and more psychotropic drugs, and was taken less and less seriously. Until it went so far that I was now considered a faker. How can someone be so sick and have such a good blood count? One of the most harmless quotes from a doctor was "clearly there must be a psychological problem behind your illness, as you must be depressed because I can see from here, you are perfectly healthy!“

I was tired of being exposed to medical gaslighting and could not take it for another 15 years. At the age of 25, I completed my technical qualification and training, despite a high rate of absenteeism. I had been to the family doctor more often than to any educational institution. I was very tired of sitting in the waiting room with a bad conscience, also with the absolute clarity that I was sick, and not lying or pretending. I suffered every day and young people do get sick, but nobody wants to hear, see, or believe that!

After years of disappointment, investigation, and discussion, I started from scratch to write down all the background medical facts and did so with my family and especially my father. He had collected all kinds of diagnoses himself throughout his life, as had many other relatives. I had assumed that we all have different diseases.

However, after many detailed conversations, it became clear that a pattern was forming of our similar and surprising symptoms. How the hell had no one noticed this before?

Why did the doctors not ask about other relatives’ illness, symptoms, and origins? After so long, it finally clicked that clearly our family had a hereditary disease. The blood results now made complete sense, as well as the flare-ups, including the fevers (feeling warm), severe abdominal cramps, severe chest pain, were NOT appendicitis or heart attacks!! Sadly, our blood labs were only good because nobody could get to the doctor during a flare. This eternally long search for an answer, finally came to an end with a redeeming diagnosis.

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Photo provided by patient

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No sooner than this was done, I researched and received information from the FMF & AID Global Association, and went to their recommended rheumatologist, who for the first time in my life listened, took me seriously, and asked the proper questions. The diagnosis of Familial Mediterranean Fever (FMF) was now crystal clear and matched my medical history, as well as that of my family members.

With this diagnosis, I was finally able to sleep peacefully again. Sadly, years ago, there were premature and meaningless deaths in my family, a mystery now solved. FMF, when correctly recognized, can be treated, and I am incredibly grateful to the association, and their online social media support groups because without them, I would have never been able to get a diagnosis and to understand that many others suffered the same way my family had. At this point, a heartfelt thank you again to the FMF & AID Global Association, especially to Malena Vetterli, without you, I wouldn't be where I am today.

Photo by Howie R on Unsplash

Familial Mediterranean Fever

FMF is the most common genetic autoinflammatory disease, causing recurrent episodes of painful inflammation. While prevalent to people of Mediterranean origin, FMF can affect individuals anywhere. Patients may present with symptoms whether they are heterozygous or homozygous.

Onset: usually occurs in childhood or the teenage years, but can occur much later.

Flares: usually last 1 – 3 days, but can last up to a week and can vary in severity.

Symptoms: recurrent fevers, severe abdominal pain, skin rash, joint pain/swelling, headaches, chest pain, muscle aches, constipation and/or diarrhea, bloating, nausea, fatigue, anemia, etc. Amyloidosis is the most serious complication of FMF if left untreated, causing kidney failure.

Patient journey: Marija (Switzerland) with Still’s disease

I am a 44-year-old woman with 2 children and currently work 40-60%. I had my first symptoms when I was five years old, which started with inexplicable bouts of fever, lasting 1–3 days and then just disappeared. My lymph nodes in my groin would swell on both sides, it was very painful and eventually, had to be operated on.

During puberty, I always had a fever flare after school hikes, sports days, or simply when I overdid it. I also suffered from migraines since age 11. I was also diagnosed with asthma and a congenital heart valve defect from childhood. My symptoms calmed down after puberty and I had no further issues apart from migraines and asthma. I was susceptible to the flu, colds, pneumonia, and mouth ulcers.

Photo provided by patient

Unfortunately, my heart defect kept getting worse as I aged, and I was diagnosed with severe cardiac insufficiency, requiring a major heart operation in 2018. After the surgery, I had increased ferritin levels, and in October 2019, I had my first flare requiring hospitalization, where I was put on painkillers and IV antibiotics. I had severe pain in my neck due to lymph nodes swelling, which also affected my chest area, as I could not breathe properly, increased heart rate, fever up to 41.7°C/107°F, a pink rash covering my neck, chest, and thighs, severe joint pain, as well as upper abdominal discomfort.

My leukocytes were slightly elevated; however, my CRP value was very high. In 2020, I had two flares, for which I was prescribed cortisone. It worked well and eliminated all the pain and fever. I was given cortisone as a long-term therapy for two years. However, I started having breakthrough fever attacks where my CRP was elevated. This was triggered when I overexerted myself or worked too much. In addition, I had chronic generalized pain and extreme migraine attacks. My doctor suspected that I could have polymyalgia rheumatica.

I sought help from a rheumatologist who did not take me seriously at all. His assessment was that I suffered from a complex set of symptoms related to chronic pain syndrome and related to psychological factors. He referred me to a psychiatrist, who was the one who referred me to immunology.

In immunology, I was immediately given various tests and diagnosed with an autoinflammatory disease. I started therapy with colchicine and Ilaris (a biological drug). Since using Ilaris, I have not had a fever and my CRP has remained normal, but all my other symptoms have continued to be quite debilitating. I receive 300mg of Ilaris every 4 weeks, but the effect of the medication does not last for a full month.

I joined the FMF & AID group on Facebook and contacted them. They provided many helpful tips and still support my case. I recommend them to everyone. Ironically, I sent them a copy of my medical report and to my surprise, they asked me if I had been evaluated for Still's disease due to my symptoms. They organized an appointment in Erlangen, Germany, where I was diagnosed by a specialist as having Still’s disease. It was a great experience, and luckily, my immunologist in Switzerland fully understood why I got a second opinion. Happily, my doctors collaborate and work well together, and they take my symptoms very seriously.

Often these diseases make it impossible for those affected to live a normal life. Pain, physical impairments, doctor's appointments, therapies, medications and authorities, renunciation, and isolation, all must be taken seriously as a part of our everyday life when living with a chronic, autoinflammatory, and invisible disease. My faith, my children, and my husband gave me strength.

I am grateful to the FMF & AID Global Association for providing me comfort and support in managing my Still’s disease.

Photo provided by patient

Still’s Disease

Still Disease is a systemic inflammatory condition, primarily presenting at two age intervals (16 to 25 and 36 to 46). It is characterized by daily fevers, arthritis, and a salmon-pink rash.

Symptoms: high daily fevers (typically spiking during the evening), rash, splenomegaly, lymphadenopathy, myalgia, arthritis, hepatomegaly, pericarditis, pleuritis, severe anaemia, abdominal pain, renal dysfunction, fatigue, headache, sore throat, and weight loss (may be rapid).

Patient journey: Michaela (Austria) with Still's disease

In October 2020, I first had the feeling that a nasty flu was looming - and of course the main suspicion at the time was Covid. I had severe pressure in my chest with massive pain and at some point, I could only sleep sitting up. At the hospital, they noted I had mild pericarditis, fatty liver, and elevated inflammatory markers. I had to be hospitalized for a week, but since nothing tangible was found, I was sent home.

Two weeks later, I started to get worse day by day. One morning, I woke up with what, I thought, were hives. Then my body became weaker, and I could not even get up. Suddenly, my face began to swell, then my whole body, as I was struggling to breathe. I called an ambulance, and in the hospital, I was given an oxygen mask and large amounts of IV cortisone due to my inflammation markers being extremely elevated.

My body repeated the same scenario daily when I woke up each morning. The rash would begin on my arms and then spread to the rest of my body, disappearing again towards the evening. In the afternoon, my fever started to spike, and the next morning, I would be fever-free again. The doctors were at a loss, as I was examined intensively with MRI's, CT's and ultrasound. My rash was dismissed as urticaria and not taken seriously, and I was treated badly and with disrespect every time.

They were all stressed because my pericardium was so badly inflamed, and I had fluid in my lungs, but at the same time, the doctors were overwhelmed with the situation. If I had been physically able, I would have gone straight home! At some point, a young resident told the doctor that she had heard of this combination of pericarditis, fever episodes and skin rashes before and that it was something rheumatic. Unfortunately, she was ignored.

My days in the hospital were all the same - sad and empty, in pain, alone, tired and without emotion. I had neither fear nor hope. I asked my mother to schedule an appointment for me with a cardiologist, who believed it was something rheumatic. The rheumatologist looked through my thick file with the medical reports and my photos. After only 10 minutes, he said: "You have Still's disease... an autoinflammatory disease that affects your organs, nerves, bones, joints, and muscles."

Photo provided by patient

The rheumatologist said he would prescribe me a biological drug to be injected every 4 weeks. The medication stopped my fever and rash, but not my pain. According to the rheumatologist, my inflammation is well under control. However, recently, the interval of my medication got extended from 4 to 6 weeks (I believe my doctor is under a lot of pressure to stretch it due to its high cost...), but ever since, I'm doing a lot worse than before.

A few weeks after my diagnosis and knowing that my disease was incurable, it began to weigh heavily on me. In the summer of 2021, I had reached the point where I knew I could no longer manage it alone. Even though I was always good at helping other people, I suddenly felt utterly helpless. The pain in my legs and back worsened to the point where I was no longer able to drive.

In April 2022, I had the most massive flare, which put me in the hospital for 6 weeks. The worst thing about this disease is that the flares always come without any warning!!! I had just had breakfast, everything was great, and 10 minutes later I could not breathe, my face was blue, and my body was swollen. This flare put me in a wheelchair as I was suddenly unable to walk.

This disease has taken a mental as well as physical toll on me, to such an extent, that I need regular psychiatric therapy.

Every 4 months, for 3 to 5 weeks at a time, I check in for this therapy. I was not aware that there was treatment for people like me, who suffer from depression, but are able to participate in a normal life.

As a Still's patient, I am investigating additional doctors who might help with my case under the direction of the FMF & AID Global Association. This organization has been very helpful providing guidance and support to aid in my case, and I am most appreciative of their help.

Photo provided by patient

Webinar organized by the Chilean Auto-inflammatory Association by Josée Abourbih

The Chilean Autoinflammatory Association (Asociación Autoinflamatorias Chile) is organizing a symposium, via webinar, for patients, their families, health professionals and the public, for September 8, from 3:00 p.m. to 7:00 p.m. (Chilean time). For patients or general interested people in Europe who wish to participate, the webinar will start at 9pm CET.

Topics of interest for patients and medical professionals will be presented, including:

- Genetics in Autoinflammatory Syndromes: What happens when a genetic cause is not identified? Dr. Juan Ignacio Arostegui
- The importance of Genetic Diagnosis in rare diseases: Dr. Gabriela Repetto
- Autoinflammatory syndromes in Adults: Dr. Marcela Ferrada
- Cutaneous manifestations in autoinflammatory syndromes: Dr. Camila Downey
- Therapies and precision medicine in autoinflammatory diseases: Dr. Cecilia Poli
- Well-being and mental health in patients with rare diseases: Psychologist, Josée Abourbih

After the presentations, there will be a round table exchange where everyone can ask questions. An invitation will be sent to all Spanish-speaking patients and affiliate associations through the social networks of the FMF & AID Global Association for everyone who wishes to attend.

Autoinflammatory Association of Georgia by Davit Tatoshvili

Our Georgia organization has created a process for supporting direct care to patients with suspected autoinflammatory diseases (AID). We believed there was a gap that required patient-centered solutions. Our two ongoing projects fulfil this need.

The first project includes offering affordable genetic testing to all the patients who meet the Tel Hashomer criteria for FMF. These patients are offered PCR testing for the MEFV gene for only $30. This test checks for 12 common mutations in MEFV gene, as well as SAA1 isotypes. Currently, more than 200 patients have been tested and treated under this program.

The second project focuses on offering affordable blood testing to patients with both the clinical or genetic diagnosis of autoinflammatory conditions. These patients are able to undergo routine lab testing for only $20, and the test includes Complete Blood Count with differential, ESR, C-Reactive Protein, and urine analysis.

This project has been a huge financial help to our patients with AID as there are no special health care programs in Georgia. Thus, with the reduced cost, the genetic testing is priced at $300-400 and routine lab testing is priced at $80-100. We are grateful to able to offer our community these important tests at an affordable price to ensure the patients get the care they require.

Comments on the Summit by the Turkish Association by Fatih Metin, Chairman of the Board

Our Turkish association BEFEMDER (Patient Association for Behçet and Familial Mediterranean Fever) attended the European Summit for Autoinflammatory Patient Associations', to which we were invited by FMF & AID and attended on behalf of our Turkish patient community. I was pleased to meet the representatives of other NGOs affiliates who share our same challenges, struggles and problems.

At the summit, we had the opportunity to compare NGOs' institutional status, successes, and challenges. We evaluated the associations' strengths, weaknesses, capacities, and opportunities. In the focus sessions, we were able to evaluate all the affiliates' health systems. Additionally, we discussed the matter of facilitating access to autoinflammatory disease diagnostics and reducing the bureaucracy for obtaining treatments and medications. Again, it was emphasized the limited number of physicians available to see patients with autoinflammatory diseases, and the need to increase the number of specialized centers.

Find Turkish group at: behader.org

Although there are some difficulties in accessing diagnosis and treatment within the Turkish health system, free genetic testing is available to patients, which is advantageous compared to other countries. It was also discussed that travel to specialty centers may put some patients at a disadvantage, due to travel distance and cost, which could be overcome by consultations with legislators.

The group discussions regarding different aspects of health policies in various countries emphasized the necessity of NGOs to cooperate closely with the legislators/government and researchers in regard to the issues emerging from this meeting.

I would like to take this opportunity to thank Malena Vetterli from the FMF & AID Global Association for inviting us to this meeting and hope to meet soon at future events.

Photo provided by BEFEMBER

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

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Italy | Brazil | Thailand | Morocco | Italy | Spain | Middle East | France