FMF & AID Newsletter

September 2021

FMF & AID Global Association and affiliated associations - www.fmfandaid.org

INSIDE THIS ISSUE

Editorial

By Malena Vetterli

Dear Readers,

We are excited to share this issue of the FMF & AID newsletter for our autoinflammatory patient community, while also recognizing **September** as being **World Autoinflammatory Awareness Month**.

Issuing this newsletter, we believe is a great opportunity to highlight how our organization is assisting patients all over the world. We have included patient stories from several countries that have been shared with the FMF & AID and demonstrate how we have been able to successfully serve autoinflammatory patients. We are a unique and growing umbrella of autoinflammatory organizations collaborating together to improve patients' lives.

The FMF & AID Global Association and its affiliates inform, educate, assist, and advocate for patients with autoinflammatory conditions, as well as seeking to educate physicians about these diseases. One of our aims is to increase physicians' ability to recognize key symptoms associated with these rare diseases. Additionally, we will help refer patients for further evaluation, to appropriate rheumatologist, immunologist, or infectious disease doctors who are specialized in treating autoinflammatory diseases.

We appreciate all reader feedback with regards to future newsletter ideas and topics of interest. Please share your comments with info@fmfandaid.org.

We would like to wish everyone a healthy September.

The FMF & AID Team

September is World Autoinflammatory Awareness Month

Every year, during the month of September, thousands of patients worldwide, who are living with one or more autoinflammatory diseases, come together to make their voices heard and raise awareness.

For the majority of these patients, the toll of having a debilitating condition reaches far beyond their physical health, affecting every aspect of daily life. These often-invisible diseases, frequently result in patients being dismissed, ignored, and neglected.

Every September, FMF & AID launches a month-long social media campaign to share important facts, discuss lived experiences, and speak out about the impact of autoinflammatory diseases impacting patients' quality of life.

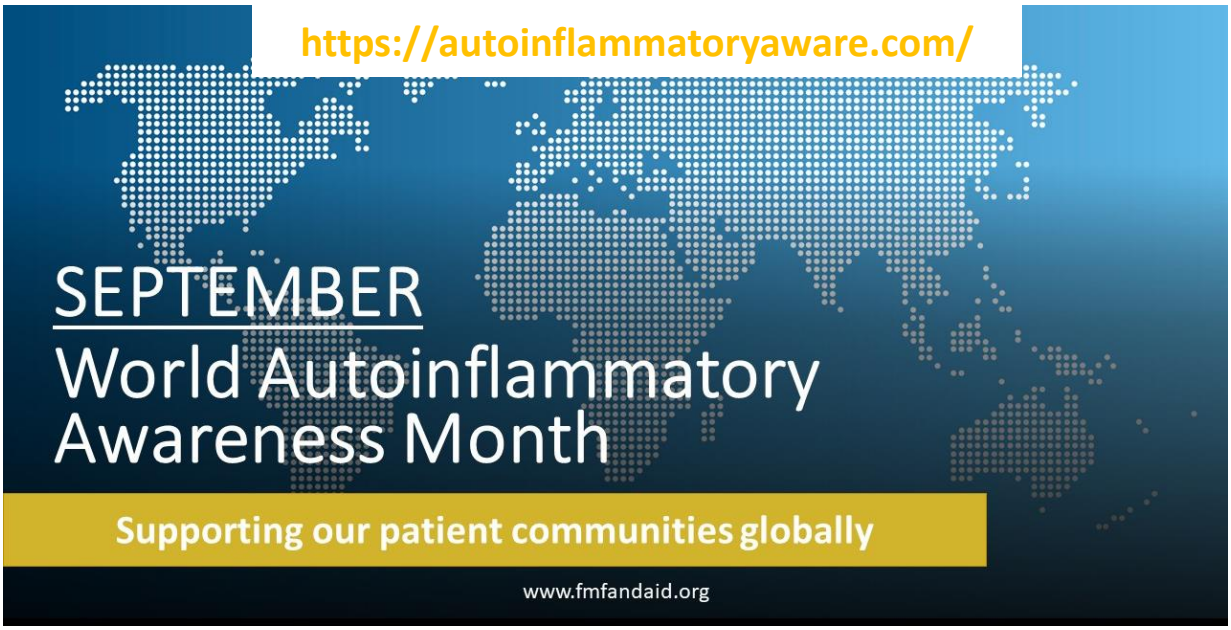
Once again, we are highlighting the challenges that patients and families experience in their autoinflammatory journeys. We may be living with a pandemic, but for our community, there is an ever-growing global health emergency in the world of Autoinflammatory diseases, because patients continue to be undiagnosed and unable to receive the necessary treatments.

Children in Ukraine, as well as patients in South and Central America, still have no access to biological therapies.

In other countries, such as Lebanon, patients are unable to access vital medications, due to the financial crisis there and medical hoarding due to COVID.

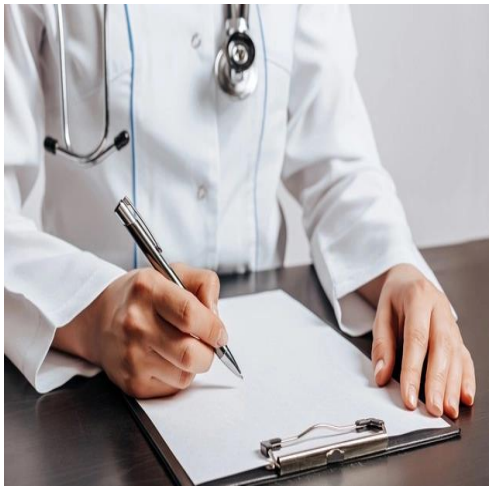
The consequences of being left untreated are potentially fatal. Chronic inflammation is harmful, and can cause long-term damage to the joints, heart, brain, and other organs. Some patients may develop Amyloidosis, a protein that builds up in organs, resulting in organ failure.

We call upon Government Officials, Health Ministers, Pharmaceutical Companies and Healthcare providers to ensure that no autoinflammatory patient goes undiagnosed and untreated.



Medical Needs Assistance Program

Thanks to generous donations received, FMF & AID has been able to continue helping patients through our Medical Assistance Program for a second consecutive year! To-date, we have already provided financial medical assistance to the value of ca. €20'000/US$23'500. This assistance has helped patients in France, Chile, United Kingdom, Switzerland, Canada, Ukraine, El Salvador, Greece, United States, Spain, Uruguay, Germany, Australia and Mexico.



The funds have been used to pay for genetic testing, prescription medications and private consultations not covered by patients' health insurance, travel & accommodation costs for consultations with a specialist in other cities or countries, as well as psychological therapy for children.

With your support, the FMF & AID will continue offering Medical Assistance Program for patients in need. For many of these patients, we are their last hope. Please consider making a donation to support this program. To donate, please visit www.fmfandaid.org.

Some of the cases we have assisted with are detailed in this newsletter.

GoFundMe campaign for Lebanon and Chile

As you may know, Lebanon is going through a serious financial crisis. The pharmacies are out of medications and patients/parents of children with Familial Mediterranean Fever are desperate because they have run out of colchicine. The representative of the FMF & AID Global Association in Beirut got in touch with the local distributor, and they still have colchicine. They are just not delivering it to the pharmacies due to financial constraints. We are purchasing colchicine to be delivered and dispensed to pharmacies to aid the supply chain issues. Patients will be able to collect it with a prescription, and if their name is on the list, they will receive the medication at no cost.

Another current problem, children in Chile diagnosed with autoinflammatory diseases (HIDS, TRAPS, CAPS, FMF, uSAID) and requiring biological medications. Most of them are between the ages of 1 and 5 years. Unfortunately, there is no coverage in Chile for autoinflammatory diseases and children are dying needlessly. More details can be found in the GoFundMe campaign. We are running against time. Please help us to raise enough funds and donate. Your donation can save lives. Thank you! https://gofund.me/3dc6005e

Zoom patient support groups

"Helping people to help themselves." True to this motto, we were able to confirm that the Zoom support groups that the FMF & AID is offering have been well received and patients are grateful for this opportunity.

The desire of patients getting together had already been acknowledged. However, when COVID hit, it was clear that something had to be done and fast. Since meeting in person was no longer a possibility, the FMF & AID came up with the idea to offer professional support groups on Zoom to the autoinflammatory community.

Psychologists were approached and while some offered their services for free, others required a fee. Several patient groups were organized: a group for young adults, adult groups, support groups for parents whose children are affected, and even a therapy group for children where they receive tailored-professional psychological support. Most of these support groups are offered in English, German, French and Spanish.

A group in Italian was also offered, but unfortunately, there were insufficient numbers for it to be viable.

The FMF & AID was also able to organize two Q&A sessions so far: one with an adult rheumatologist, Dr. med. Jürgen Rech, and a paediatric rheumatologist, Dr. med. Tobias Krickau, from the Autoinflammatory Centre at the UniKlinikum Erlangen, and a second one with a paediatric rheumatologist, from the same centre. Both Q&A sessions went very well, and parents very much appreciated the opportunity they were given to ask questions and moreover, they were very grateful to have their questions answered by experts in the field.

Special thanks to our dedicated psychologists:

- Alev Kayan
- Josée Abourbih
- Karin Purugganan
- Kirstin Grös

Kids' therapy group funded by the FMF & AID

by Mary Frank

In one of the FMF & AID Facebook support groups, a parent wrote, "Would anyone be interested in forming an autoinflammatory support/social group for our kids? My 11-year-old has never met anyone with the same condition." The response was overwhelming. 28 parents chimed in that their kids were in the same situation. FMF & AID Global Association was there to listen and support.

The group developed into a once per month zoom meeting let by Karin Purugannan, MA, MEd, NCC, LPC and Mary Frank, MEd. Karin is a children's counsellor, who additionally comes along with years of working as a nurse in a pediatric hospital, and Mary is a parent of a child with FMF and an early childhood educator. The group is funded by the FMF and AID Global Association and meets once a month for two hours. The ages of the kids range from 4.5 to 13 years and is a mix of boys and girls from all over the world connecting.

The Zoom group gives kids a chance to meet others with a rare condition, share their experiences and struggles, and cheer each other on during challenging times. The kids are given a chance to discuss topics related to living with autoinflammatory diseases such as: how to deal with frequent absences from school, what it feels like to tell friends about their condition, strategies for getting through a flare, and have even discussed tips on how to get through uncomfortable medical procedures such as drawing blood and getting injections.

The group is also given "tools" to add to their social emotional toolbox of dealing with a rare disease and life in general such as belly breathing, using I statements, exploring a feelings wheel and circle of control, and grounding exercises. Lastly, kids are given a chance to just be kids with each other, and to have fun and talk. Kids have become pen pals and gaming partners and formed lasting friendships with others with their condition around the world.

A few quotes from parents:

"I just wanted to write and say thank you for holding these sessions and thanks for including us. My son really enjoyed the session and can't believe he met another kid with PFAPA! He's looking forward to the next session and so are we."

"I cannot thank you enough for this opportunity to have the kids connect with others experiencing the same issues. It's so refreshing, and my heart is just so full of appreciation to you and all that you do to put this together."

"I'm so grateful that my son was able to participate today. Thank you for doing this for and with our kiddos!"

"As a parent, I am grateful to know that there is an organization that cares for our kids and will give them an amazing opportunity to connect."

www.wonderologie.com

Suggestions for recording symptoms by Ellen Cohen

Communicating with health professionals as a rare autoinflammatory patient can be filled with anxiety, misunderstandings, and potential pitfalls when conveying symptoms and relevant details with your healthcare provider. Several strategies can facilitate this process to ensure a thorough and relevant exchange.

A symptom diary with detailed notes on all organs/body parts impacted before, during, and post flare can be very helpful to track and present how disease manifests in a particular case. Recording fevers (AM/PM), GI distress, bone pain, fatigue status/activity, rashes, pain rating, all per specific body part can help the clinician visualize what is going on during a flare. Many symptoms will also present in breakthrough, mild form, or in new ways outside flare time, which also should be recorded to evaluate efficacy of treatments, be it prescribed or over-the-counter medications. Keeping this diary over time will also allow patients to spot trends and ask additional pointed questions.

Another way to help develop a disease diary is by taking pictures of rashes, flushings, sores, hair loss, skin discoloration, swelling, etc. Pictures can be taken daily/weekly/monthly depending on how often these issues occur.

The pictures can be noted in a diary for reference and shared with the doctors to determine what type of treatment or additional consulting might be needed.

Other tips that may be helpful:

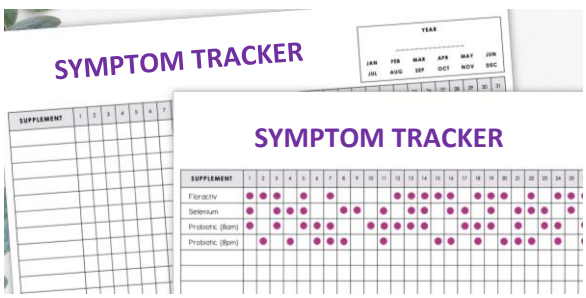
-Make copies or screenshots of all labs and prescriptions ordered if doctor provides written scripts, allowing recall of test name at result time.

-If on medications used are biologic shots or in IV form, take a screenshot of box or bottle with batch number every time medicated monthly so should a manufacture recall occur, patient already has detailed information. If doing daily shots, screenshot batch number at time of monthly delivery.

-Keep a written list of doctor contacts and medications where a family member can access, should the patient require emergency help.

-If getting scans, x-rays, or MRI's always ask for a copy be sent to you after the doctor has been provided with a copy, if allowed.

Please let us know what tips have worked in your case so they can be shared with our patient community.



PFAPA Support Group

by Lisa Scott

I am honored to be helping FMF & AID by moderating their PFAPA Support Group, which has almost 6k members. When my oldest daughter was born, we started to experience symptoms that many doctors would state were viral due to my daughter being in day care. We persisted and with the assistance of our pediatrician who also slowly started to realize our daughter was having fevers way too often and triggered by vacation and major events in our life. I would be amiss to not add the tenacity I myself had to put forward to get my child diagnosed and genetic testing. It seemed like it was a diagnosis of PFAPA they were going to give until I pushed and said I wanted to have testing done to make sure no other condition could be causing this. The support I received from FMF & AID assisted me to feel confident enough to request this.

From what I have experienced as a moderator is an overwhelming number of families being told its PFAPA merely off of "a steroid worked" or "your child does not have all the symptoms" and the "oh you are not of that ethnic group". Genetics show that individuals of many different ethnicities, single mutations and even no mutation (that we know of) ARE suffering. Genetic testing is costly, and getting it covered in a timely fashion is a battle many families fight as a reality today. When you do find a family, who is going to advocate for this step you see the frustration they have when a doctor says, "It is not a necessary step." FMF & AID has been advocating for our members to get in touch with caring and knowledgeable doctors around the world to provide one-on-one guidance and support. In our groups we collaborate to make sure our families are heard, supported and getting the much-needed treatment they are searching for.

PFAPA Support Group

by Lisa Scott

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Sadly, we have many members in PFAPA Support Group that are misdiagnosed. When we explain to them the importance of genetic testing and the high misdiagnosed rate of statistics of those diagnosed PFAPA that are not it is jarring. Many have gone on for genetics, still many are fighting for the opportunity to have it completed. The consequences of flaring un-controlled, the daily toll it takes on the child and family as well as the expense and financial discord that occurs can be daunting.

Lastly, I leave you with this photo of my daughter. She went on vacation and within hours of arriving at our destination flaring began reaching 105.7F (40.9°C). This was the time when our doctor told us to go straight to a Children's hospital 2 1/2 hours away. This is when I advocated to stay in the emergency room as the Advil (Ibuprofen) had brought her down to 100.6 (38.1°C) and its probably viral, they said. We pushed and within an hour of other testing (for inflammation) I insisted on and the Advil wearing off and fever returning she was finally admitted. We finally had a treatment team and on a road of discovery, sadness and then eventual treatment and remission from PFAPA and now secondary diagnosis of FMF. We now have what we all want for our children, a little bit of normalcy. We just pray all children have the opportunity for such care and parents who can survive the early days of trying to get a diagnosis.

School

Several parents in Switzerland report continuing issues with schools due to lack of understanding autoinflammatory children's cases. Affected students are thus brought into uncomfortable situations and often not taken seriously.

For example, if a child is unable to take part in physical education classes due to pain or rash, they often forced to participate because they are not believed, resulting in unpleasant consequences for the child and parents.

Parents who have to justify their absences from school or are pressured / threatened by the authorities. This has often been reported in Switzerland, but this is certainly not an isolated issue. In many schools there is an urgent need for more information, awareness and acceptance of affected children.

FMF & AID recommend schools should develop a plan (called 504 plan/IEP in the USA) to give kids with autoinflammatory diseases the support they need and to ensure that accommodations are made to ensure their academic success and access to appropriate learning environment. For example, they might be given extended time on tests or the ability to leave the classroom when needed without having to ask for permission.

FMF & AID Patient Support Groups on Facebook

FMF & AID developed a training program tailored for autoinflammatory patient associations, Facebook administrators and moderators in collaboration with Sobi.

Sobi is a specialized international biopharmaceutical company focused on rare diseases. Sobi provided its support for organising and funding this project for the benefit of the patients. Our program offers this professional support to benefit FMF & AID volunteers and affiliated member associations.

For the FMF& AID, offering training to volunteers will help sustain the high level of advocacy and support, but also to more effectively identify and address patients' needs.

FMF & AID online Arab community represents the largest group of patients worldwide with 24.1k members and growing. The group has 50 admins and is led by two fabulous volunteers: Rami and Bushra.

Our Turkish partner, the BEFEMDER association has a large group with 14.1k members.

PFAPA support group has over 5.7k members, followed by two growing FMF groups, totalling 6.3k members respectively.

FMF&AID also has an international group for autoinflammatory diseases with 2.4k members.

FMF&AID is proud to be an international organization, serving patients in multiple languages across the globe.

Newly established association in Chile: "Autoinflamatorias Chile"

Since its inception, three months ago, Autoinflamatorias Chile's Board has been dealing with the first and probably their greatest hurdle, which is trying to have autoinflammatory diseases incorporated under the law "Ley Ricarte Soto".

Unfortunately, autoinflammatory diseases are not officially recognized in Chile (uncoded) and do not have any coverage in the health systems, neither public nor private. To obtain a diagnosis, patients require expensive genetic tests that must be sent abroad, since Chile does not have the infrastructure to do so. The same goes for hospitalizations, medications or any additional tests that may be required.

Some patients (children and adults) require high-cost biological medications. The monthly price of these drugs ranges between €2'250 / US$2'770 (2 million pesos) and €14'700 / US$17'350 (13 million pesos), per patient. Currently, this cost, as well as all other costs – such as frequent hospitalizations - must be borne by the families, through various charitable campaigns, which are stressful and unsustainable over time.

PATIENT JOURNEYS

(written by patients themselves or their parents)

FMF in Chile

More than eight years ago, my daughters began to present health problems, fevers over 40°, every 7 days like clockwork, idiopathic urticaria for no apparent reason, hematuria (blood in the urine), cervical lymphadenopathy (swollen cervical nodes), tachyarrhythmias, frequent pneumonia, all of the above frequently and without an infectious focus. Due to the aforementioned symptoms, there were a series of misdiagnoses that caused both girls to develop antibiotic resistance.

It was a long and unknown road, but thanks to a pediatric rheumatologist, who came into our lives, due to a medical emergency, they came up with a diagnosis that no doctor previously was able to give. No immunologist for three years could reach an accurate diagnosis regarding my daughters' disease, not even doctors from renowned clinics in Santiago de Chile. They were seen by nephrologists, immunologists, hematologists, etc.

At three years of age, one of the two girls was diagnosed with refractory epilepsy, this in the same medical emergency mentioned above. There were no more tests to be carried out, it only remained to know which autoinflammatory disease my daughters suffered from.

For this, a specific genetic test was carried out for autoinflammatory syndromes in which a pathogenic mutation responsible for Familial Mediterranean Fever was found, along with other variants. In addition, it was concluded that her epilepsy was due to it and not refractory as originally thought.

Currently, my two daughters must take daily many medications to control the epilepsy, thyroid, esophagitis, among others. We spent a year with a biological treatment which was unsuccessful and now we depend on another expensive biological treatment. Their disease has no cure, but there is a lifelong treatment which will give them a better quality of life.

Currently, neither of the girls has been able to attend school regularly, nor can they be with children of their age, or with people other than those who live with us at home.

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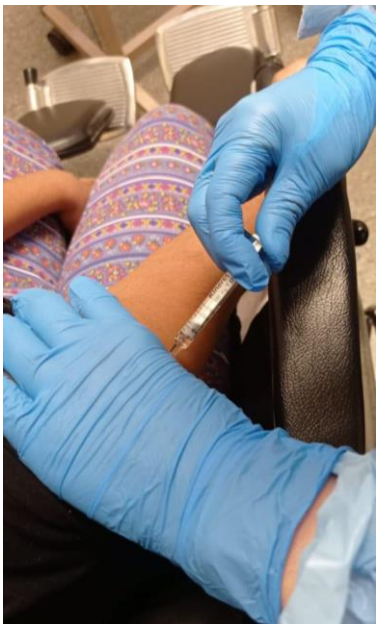
They attend the classroom of the hospital where they are treated. Even before the pandemic, they had classes at home since going out into the streets is exposing them to a great risk to their health.

Comment by the FMF & AID

Families who have a child affected with an autoinflammatory disease, know how hard it can be, not only emotionally, but also the heavy financial burden the family has to face. These issues quickly multiply when two or more members of the family are affected, which is the case with these sisters in Chile.

In South America, it is often the case that the best medical care is usually only available in the capital. For this reason, affected families have no option but to travel, often long distances, to obtain the much-needed specialized treatment. Due to the uncontrolled inflammation, one of the girls had lost the strength in her legs and was not able to walk anymore. Shortly afterwards, the other sister started showing the same disease manifestations. There was no time to lose if the girls wanted to get their lives back. The only solution was for the family to fly to Santiago so that they could undergo a series of tests. To make the stay possible, the FMF & AID found a suitable apartment located very close to the hospital and covered the accommodation costs in full for a whole month.

As a result, both girls are now receiving biological treatment and it has had a major positive impact on their quality of life and that of their family. The FMF & AID also covered the costs of some prescription medications for both sisters.



FMF in France

I am a teenager who has suffered from high fevers, abdominal pain, headaches, joint pain & swelling, back pain, dizziness, vomiting, nausea, bloating, diarrhea, ever since I can remember. The first few years, it was only fever, but then other symptoms started to appear. I was never taken seriously, not by my family or by any of the many doctors I saw. It got to the point where I couldn't stay any longer with my family, and doctors blamed everything on it being psychological. The FMF & AID not once doubted me and did everything they could and beyond to help me,

They arranged for me a consultation with a specialist in autoinflammatory diseases. However, this doctor did not want to help me and even told me that I had nothing and that I should stop taking colchicine, despite the fact that I told him that once I stopped taking the medicine and I had a very bad flare. At this point, I had almost lost all hope. Then another appointment was arranged for me somewhere else. This time, my inflammation markers were not high enough during my hospitalization, so I was back to square one. I contacted a few French associations, but no help was offered. Once again, I contacted the FMF & AID, and it felt great to have someone who cared, someone who was always there to answer my calls regardless of the time.

My health was deteriorating rapidly, the flares were coming almost weekly, and the pain was unbearable! I asked them to help me and that I was prepared to go anywhere as soon as I turned 18. Thanks to the kind heart of the volunteers of the FMF & AID, as well as their representative in France, right after turning 18, they organized a trip for me to Switzerland, because all doors closed up for me in France. They knew I was on my own, so the FMF & AID paid for the flight to Switzerland, accommodation, consultations, medications, medical insurance in Switzerland, and everything I needed during my extended stay. Upon my arrival, I was immediately taken to a great immunologist at a University Hospital who the FMF & AID works closely with. He listened and took me seriously. This doctor reviewed my medical history with other eyes, got in contact with my French doctor, confirmed the diagnosis and put me on medications I had been denied in my own country. Not only I had my life back, but it also had a meaning again. After over half a year of healing and recovery time, I went back to France. I will never be able to put in words how grateful I am. Thank you from the bottom of my heart.

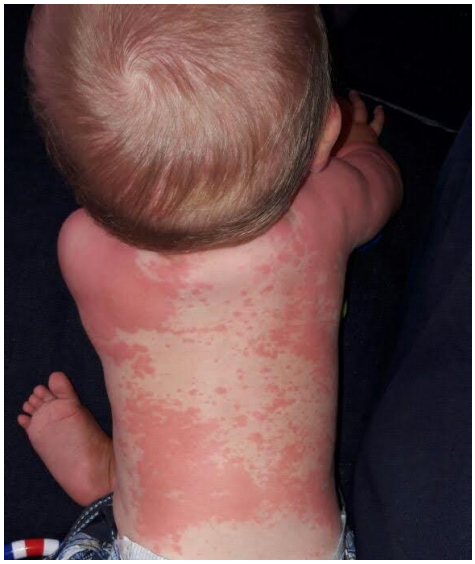
FMF and PAPA in Germany

My husband and I have 5 children. We always dreamed of having a large family. As a child and adolescent, I was very sick, and it took a long time until I was diagnosed. I was diagnosed when I was 16, and I was said to have discoid lupus and fibromyalgia. My life was very difficult, and I had to endure many operations (spleen, liver, appendix, tonsils), which now looking back, did not improve my symptoms and were probably all made on the basis of incorrect diagnoses and were therefore superfluous. With the first pregnancy, I finally felt better. Our 3 older children were born in quick succession. Our second son was often sick from a young age, but everything had been dismissed as infections, childhood migraines, abdominal migraines and later on, as depression. My daughter (3rd child) started with the same symptoms when she was about 5 years old. Nobody knew what was going on, many visits to the doctor followed, and in the end, everything was attributed to childhood depression again. For us parents, it was hardly bearable, and we couldn't explain why our children suffered from depression. What were we doing wrong? It was hard to see the pain they had to live with. When our fourth child was born, we initially thought he was fine.

Until the same symptoms started shortly before he started school. His first diagnosis was again: depression. What were we doing wrong in driving all of our children into depression?

Many of the symptoms of our four children were familiar to me. A struggle with life began, a struggle for our children, who, due to their unrecognized illness, experienced severe suffering such as bullying, exclusion and a lack of self-confidence. It hurt the soul to see how the children suffered and how none of them really got their feet on the ground.

It wasn't until our 5th child was born that we finally received the correct diagnosis. He had an extremely high fever with severe pain and screaming fits for the first time when he was 4 months old. At first, it was thought to be infections again. So once more antibiotics, like the older children used to get, were prescribed. At that time, we already knew it was severe, but we didn't know what that meant. Five weeks later, the next fever episode came. Fever up to 41°C for 5 days. Nothing helped. Once again the diagnosis was an infection.



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From then on, I had the feeling that something was wrong. This time, with his fever and pain, he got a violent rash all over his body. Ears, hands, head and face, everything was swollen. The body was fiery red, and it burnt and hurt. The flares repeated until he was 1 ½ years old. Then finally the diagnosis came: FMF. We were all examined, and the result was the saddest day of my life: All of our children have FMF and so do I, which means that I have passed it on to them all. I and the 3 older ones also have PAPA syndrome. My husband is healthy. Since then, everyone has been treated and everything is going a little better, but not yet well. Unfortunately, our second and third children took too long to get the correct diagnosis. As a result, they struggle with secondary diseases such as vasculitis, arthritic psoriasis and osteoarthritis.

In the meantime, everyone has been affected mentally by this ordeal. As they were never believed, and they were labelled as liars and faking it, they were mistreated for years.

We all suffer a lot, and our lives are very depressed and difficult. We try to give our children a good life, but it is not easy. Our oldest child is the most severely affected. He still has a few flares but is now able to work. Our second child is also severely affected and is being treated with biologicals.

He requires strong pain medication. He cannot work and has extreme sleep problems. Our third child is also severely affected. She is also treated with biologics. In winter, she can hardly do anything because her feet swell and harden due to vasculitis.

Our fourth child is moderately affected, is given biologics and can usually cope with everyday life. Our fifth child is extremely affected. Since the drug no longer works in high doses, he now receives a new drug every 14 days in the hospital. Unfortunately, it's not that effective.

We all hope that there will be a breakthrough in medicine at some point. Our children suffer and have such a difficult fate to bear. As parents, we are very sad and in constant worry.

I would particularly like to thank Malena Vetterli from the FMF&AID. She helped us so much to locate an appropriate doctor. She is always there to provide guidance and support. Thank you for everything.

"I can only recommend all parents of affected children, keep trying, believe your child's symptoms, see as many doctors as necessary until you get a diagnosis. Stay strong and never give up hope."

uSAID in Italy

I am the mother of a boy whom I will call Mario. Before autumn 2012, Mario had no particular symptoms except migraines, excessive sweating in the limbs. He had a normal physical and mental growth. His school performance was excellent and he participated in many sport activities.

In the summer of 2012, puberty began, and in the autumn, the following symptoms started to appear: burning eyes, diarrhea, vomiting, nausea, recurrent fevers of up to 40C° (104°F), sometimes with a small break of one month in between. Other symptoms independently from the ones previously mentioned: joint pain, muscle pain, fatigue, mouth ulcers, skin rash, myalgia, arthralgia, chest pain, abdominal pain under the diaphragm that can only be controlled with opiates, etc. Two PET scans detected an active arterial vasculitis of the great vessels in first degree in 2016, and the second PET scan in 2019 was assessed as second degree. Generally, the symptoms are more evident in winter, and in summer they tend to decrease. The duration and the intensity of the flares has been increasing over the years, with short periods of well-being between episodes.

While searching for a diagnosis, my son had several hospital admissions, plus 7 hospitalizations. These were carried out in 6 different pediatric and non-pediatric hospitals.

Several specialists were involved during each visit in search of a diagnosis.

Genetic testing revealed the presence of pathogenic variants in the genes responsible for CAPS, FCAS2, as well as several aggressive variants for FMF. The diagnoses were: Munchausen and expulsive parent, psychodiagnosis, Bechet, FMF, Takayasu, FCAS2 or NLRP12. The diagnosis is currently still uncertain.

Mario, after years of being without treatment because he is intolerant to Colchicine, has been treated with several biological drugs for auto-inflammatory diseases with a partial positive response. He is currently receiving an infusion every 6 weeks which seems to be effective, but it's too soon to judge if it's really working. We must wait until autumn before any conclusions can be drawn.

This painful journey has led to a significant loss of quality of life for Mario. Mario was a child who had many friends and a very active social life. In the past few years, he was only able to attend school for one month per year, he had to stop all sports, and all the activities he enjoyed in life.

If any specialists reading this story can help, please contact the FMF & AID (info@fmfandaid.org).

uSAID in Mexico

My daughter was born in Mexico, the country where we live. During the first 3 years, she grew and developed normally, although with reflux problems and allergic rhinitis. When she was 3.5 years old, she developed fevers of up to 42°C (107.6°F), accompanied by severe stomach pain that was unresponsive to paracetamol or ibuprofen. We turned to different pediatricians but had no answers. Each time, we were told that it was probably a stomach infection, which they treated with antibiotics, but my daughter's health was not improving.

We even went to see a renowned gastroenterologist, who told us that it was food allergies (supposedly to wheat, corn and eggs), so we were told to eliminate these foods from her diet. Despite this, there was no improvement. The fevers continued month after month and my daughter became increasingly exhausted and malnourished. By then, she was in the 2nd year of Preschool and was unable to attend classes regularly due to her being ill several days a month.

When my daughter was 5 years old, she had a very strong flare, but we kept hearing that it was a bacterial or viral infection. Nevertheless, the fevers continued. In the summer of 2014, an infectious disease specialist and an immunologist became interested in the case and finally gave us the diagnosis that it was an Autoinflammatory Syndrome.

They proceeded to treat her, but now the fevers that she presented every month were accompanied by inflammation of the tonsils, which seemed to indicate that it was a bacterial infection, and once again, she was treated with antibiotics. At 6 years of age, and thanks to these specialists, my daughter was admitted to a renowned Pediatric Hospital and the problem was treated with corticosteroids. However, her health issues continued to be present every month, but now in a less intense way.

At 8 years of age, the immunologist decided to remove her tonsils, which helped to improve the fevers and monthly flares, although the intestinal issues appeared. To this day, my daughter continues with intestinal issues and has also been diagnosed with a Vascular Syndrome.

Cont.

As a mother, I can say that it was and is very difficult to get to a clear diagnosis. These types of illnesses generate a lot of impotence, having to see innumerable doctors without any improvement, seeing our daughter flaring month after month, not to mention the tremendous physical and emotional wear, stress and very heavy financial burden that such a rare disease causes.

I feel very proud to belong to the FMF & AID Association since it supports us and allows us to share our doubts and experiences with people who suffer from this type of Autoinflammatory Syndromes diseases in different countries.

Moreover, they put me in contact with medical personnel here in my country, to clarify doubts and for my daughter to receive specialized care. My daughter is currently being treated with high-cost medications and we also receive financial support from the FMF & AID Association with this. Thanks a lot.

uSAID in Spain

When my daughter was born, it was a difficult delivery. A few hours after birth, a blood test was performed, and her CRP was high. Then a little later the same day, her temperature was taken and surprisingly, she had a fever. During the first week, my baby was put on antibiotics. We still did not know, but an ordeal was beginning that has lasted for 10 years. At 40 days old the fever returned, and she had to be admitted to the hospital. The pattern of fever showed no infection, but her leukocytes and CRP were high.

Since she was born, that has been our history, admissions, blood tests, as well as genetic testing but with negative results. After years, my daughter was finally put on a biological medicine, which didn't help. Currently, some symptoms have changed, such as the wounds on her skin now appear with or without fever. Fatigue is a common daily symptom, and she also gets headaches, earaches, and abdominal pain. There is always something. For this reason, we cannot go anywhere without taking corticosteroids and anti-inflammatories with us, since at any moment, we know that the pain and discomfort can start. She underwent a tonsillectomy when he was 7 years old.

If any specialists reading this story can help, please contact the FMF & AID (info@fmfandaid.org).

uSAID in Switzerland

Our journey with an autoinflammatory disease began the day after our son was born. He got a fever for the first time. From then on, our lives were ruled by bouts of fever and so our odyssey began.

For no reason, he started to have fevers every 2 weeks. He screamed and screamed; we couldn't lower the fever with medications. Again, and again, we went to the children's hospital and also to various doctors. Nobody seemed to want to find out what was going on. No real examinations were done, and all we kept hearing is that it was just a viral infection.

Once a pediatrician told me to buy vitamins at the supermarket, that it helps just as well as the expensive ones. We received no understanding from the doctors and certainly no help.

One night, when I was lying next to my son, who was crying and couldn't fall asleep because once again he had: a fever of over 40 degrees, the whole throat full of painful canker sores, a headache and joint pains. So I started searching the internet. I kept telling myself, there has to be an explanation. I have to help my son.

At this point, the PFAPA SYNDROME was already known to me, but it was always clear that it had to be something else. So, I searched and searched until I came across a Facebook group. Luckily, I got to know Malena Vetterli from the FMF & AID. At last, there was someone who understood exactly how I felt.

Cont.

From then on, things finally moved forward. I was put in contact with doctors who took us seriously and who confirmed that we were absolutely right. It took 7 long years and over 8 years until we could start therapy. However, to this day, we still do not have an exact diagnosis. Finding a suitable therapy is still very difficult as the disease is still largely unexplored, and many doctors have never even heard of it.

What does it mean for us as a family to have children with a cold-induced autoinflammatory disease? We live very isolated. Our daily life is determined by the disease. We have to constantly check temperatures. Wind, rain, outside temperature etc. cold air, cold water, rain or snow can trigger a flare-up.

It is not possible for us to simply go to an outdoor pool spontaneously in summer unless it has been warm enough the previous days. In winter, simply going sledging or skiing spontaneously is not an option either. Even building an igloo has consequences. Everything has to be weighed up, what are the consequences? In addition, emotions can also have a major influence. If we are planning a trip and our son is happy, it is not uncommon that the excitement will trigger a flare and therefore everything will have to be cancelled. We cannot plan 24 hours in advance. We live a very spontaneous life, where only doctor's appointments are fix.

Cont.

Bad nights being in constant pain, be it joint pain or abdominal pain is a very common issue for us. There is never a really pain-free day. This leads to fatigue. A lot of rest breaks have to be planned. Stress also triggers our child to flare.

When our son takes part in everyday things such as a sports day, school trip, etc., this is usually too much for the immune system. It overreacts and triggers an inflammatory reaction. Hand, ankle or even hip joints inflame, so everything has to be cancelled again.

It happens often, that the society has no understanding. It is also difficult for schools to understand because the disease is largely invisible. Our son often suffers from various skin symptoms, even then there is a lack of compassion and understanding when he is tired because the skin symptoms itch and it was almost impossible to get some sleep the previous night. It is ridiculous that our son is made to participate in physical education class, despite having informed the school/teacher that his joints and muscles are currently affected. The problem is that not everyone accepts that they are dealing with a special child.

Our everyday life can only be managed because one parent is at home. External support is impossible. A parent would also have to call in sick all the time. Not reasonable for an employer either. Our social contacts have become enormously limited.

Our circle of friends has been massively diminished. For this reason, I am extremely grateful for the Zoom support groups offered by the FMF & AID. Being in these groups gives me the opportunity to exchange experiences with people in a similar situation and it helps me not to feel so lonely anymore. I feel understood and the exchange is good. Even our son benefits from the free therapy sessions and enjoys communicating with other children who are also affected with an autoinflammatory disease. We are very grateful.

FMF in the UK

My son had his first flare of FMF at the age of 15 and was misdiagnosed until the age of 20. His flares affected his lungs and ribs. Once we had a diagnosis, he was prescribed an oral medication as a preventative measure. Although it didn't stop the flares totally, it did seem to help but the side effects were stomach pain and loose stools. At the end of 2019, aged almost 24 he began to get more frequent episodes which would cause him to hyperventilate, and I would have to get him emergency rushed to hospital, where he would end up in Resus for hours and having all sorts of pain relief to stop him hyperventilating. This began to become a regular thing and went on at least monthly for over a year.

Cont.

His life was terrible as he was anxious all the time waiting for another flare to come, and it was so debilitating for him.

He would be screaming in pain with every breath and couldn't even speak to the ambulance crew. His inflammatory levels were sometimes very elevated, and he would be drenched in sweat. It was horrendous for me to see him in so much pain and not be able to do anything to help. It was heart-breaking!!

In March 2021, he had a full-blown episode which had him off his feet and in bed for 18 days with two four-day stays in hospital. I was constantly trying to get answers from doctors and specialists but was told that the oral medication he had been given was the only thing which could help!

My son said that he couldn't live like this anymore… I had to do something, so I joined the Facebook FMF groups and found out so much more from other sufferers across the world. I was contacted by Malena Vetterli from the FMF & AID Global Association, who was super helpful. She called me and explained many things and gave me so much information and signposts in the right direction. I got straight on to relevant specialists and was given an urgent appointment for my son within a few days. He then started on a biological medication after being quickly approved by NICE (the National Institute for Health and Care Excellence) so it's all covered by the NHS (National Health Service), which is amazing.

It has changed my sons and my life as he hasn't had a flare since the treatment started in April 2021. Heartfelt thank you to Malena for helping us so much!!

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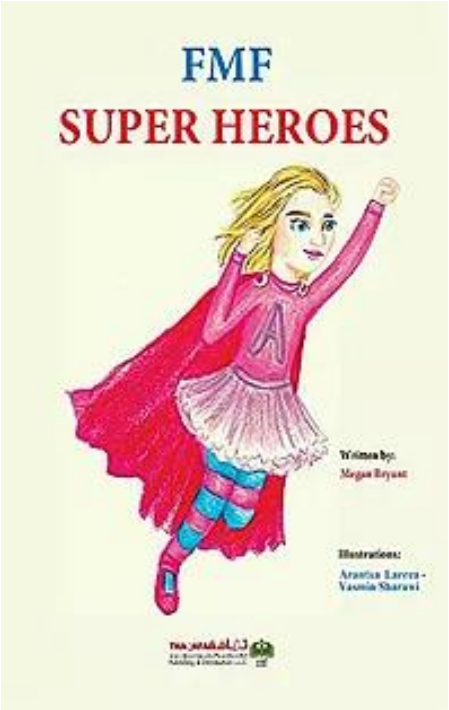
The educational children's e-book 'FMF Superheroes' is available in English, German and Arabic.
This book is highly recommended for all children. You can purchase a copy and in doing so, you will also be supporting the FMF&AID's work. Links to our books are found on Amazon: https://www.fmfandaid.org/books



NOMID/CINCA in Ukraine

For years my daughter had to endure fevers of 40°C (104°F) to 42°C (107.6°F) day and night, pain, a persistent rash, as well as severe joint pain and swelling, so bad that she couldn't even move. She was only able to sleep when the pain and fever were gone. All these symptoms had slowly debilitated her, and she was very weak. She had been prescribed a hormone therapy, which almost damaged her eyesight! She had no quality of life.

When my daughter was 4 years old, she was finally diagnosed with NOMID/CINCA. Her condition requires biological medication, but the medication was completely out of our reach. Unfortunately, it's not available in our country and there is no state assistance because the medication is not registered in Ukraine.

Therefore, before my daughter could start the medication, I had to raise the necessary funds. It took me a year before I was able to buy it. After receiving the first injection, I can say that our life completely changed. It was like having one life before and a completely different one afterwards.

From that moment onwards, our life changed. She started to sleep all night without having fevers, and she would wake up with no rash. For the first time, she was able to walk without any help, and also the first time she smiled! It was the beginning of a new life for us. However, while my daughter was learning to walk, jump, sit and dance, I had to spend my time looking for money to keep buying the medication.

Luckily, I came across the Italian "Associazione Iris ODV". I contacted them and asked for help, and they immediately offered their help and started to raise funds so that my daughter could have her medication.

Then COVID started, and we were unable to travel to Italy anymore to receive the medication, so we had to look for other options. Then the Italian Federation for Rare Diseases "UNIAMO" and "Associazione Iris ODV" contacted FMF & AID Global Association. Through their efforts, my daughter was granted compassionate use of the medication and now she gets a regular supply of the medication she needs. This has given me and my daughter our lives back. I no longer have to ask people for money, but instead, I can concentrate on my daughter. The help provided by FMF & AID was priceless!

HIDS in Ukraine

I am the mother to an adult patient who was repeatedly misdiagnosed for years. Now that my son is a grown up, I can sadly see that his disease has taken away his strength, his present and future.

It all started with my 3-month-old baby presenting recurrent high fevers. This was the first time he was hospitalized, and unfortunately, it would be one of many hospitalizations. Like every time, the fever was attributed to a bacterial infection, so it was treated with a course of antibiotics, and he was discharged from hospital. However, the fever wasn't the only symptom. My son also suffered from many other symptoms such as abdominal pain, acute respiratory viral infections, allergies, chills, pharyngitis, runny nose, mouth ulcers, to name but a few. My son used to wake up at night screaming in pain. His liver and spleen were found to be enlarged.

When my son was 10 months old, he allegedly had appendicitis and his appendix was removed. Two months later, he was diagnosed with sepsis. He was treated for septic carditis, pneumonia, pleurisy and several blood transfusions were necessary. This time, he was hospitalized for 3 months.

Over the years, my son received the following diagnoses: Fanconi Syndrome and Juvenile Rheumatoid Arthritis. Despite treatment for these conditions, he still suffered from the symptoms mentioned above plus sore and painful joints, headaches, laryngitis, and even anaphylactic shock.

As a concerned mother, I followed all doctors' instructions exactly, but my son's condition did not improve. He was also put on hormones, in fact, for 10 years but the frequency of the flares remained the same. Many doctors did not take me seriously, but luckily, there were a few others who believed me, even though they didn't know what to do to help us.

The years passed by, and my son wanted to enjoy life like other children in school. He missed a lot of school due to his illness and it affected his relationship with his friends. He felt different from his peers.

When he was a teenager, my son refused to keep taking hormones, claiming that the treatment wasn't helping. Somehow, he managed to graduate from school, trained as a goldsmith and began to work. However, due to his frequent sick absences, he saw no option but to quit his job. He tried to hold a job again and again, but due to his severe symptoms, he was forced to quit. Depression became his permanent state.

When he was in his late 20's, my son suffered an ischemic stroke (when a blood clot blocks the flow of blood and oxygen to the brain). Almost a year later, he suffered an acute intestinal obstruction requiring surgery. Then 4 months later, he had a second ischemic stroke. This time it affected his memory, the ability to speak, as well as paresis of the limbs (restricted or partial muscle weakness).

Cont.

Only after the strokes, an immunologist suggested that it could be an autoinflammatory disease and a genetic test was performed abroad. The results revealed that indeed, it was HIDS (Hyper-IgD Syndrome), an autoinflammatory disease. After reading about his newly diagnosed disease, I realized that his symptoms matched the disease, except for the strokes. After all these years, we still don't have an answer for the strokes.

Apart from all the struggles we had to endure, after his HIDS diagnosis, I also had to deal with the additional stress of trying to raise every month the necessary amount to buy the biological medications prescribed to him. According to the law in Ukraine, orphan medicines must be provided free of charge by the government, but this is not the case. The price of these biological medications (injections) is exorbitant. For people to know how insanely expensive these medications are, our family's monthly income is enough for three doses, that is, for three days. In July 2021, the Ukrainian government finally provided adult patients with a year's supply.

For 1.5 years, we had the stress of having to buy and import this medication ourselves. We raised funds thanks to charitable foundations such as the FMF & AID Global Association, relatives, friends, and classmates. My son and I are fortunately that we were able to receive help from the FMF & AID, who we know helped several patients in Ukraine.

Although my family should feel relieved that the financial slavery has ended, there is no guarantee that this help will be forever. At least we now have a name for one of his conditions, but the cause of the strokes is still unknown. Unfortunately, there is no doctor in our country who has experience in treating adult patients with HIDS. Our rheumatologists feel that it should be immunologists and geneticists dealing with autoinflammatory diseases.

If any specialists reading this story can help, please contact the FMF & AID (info@fmfandaid.org).

CAPS in the USA

I've spent a lot of time thinking back over all the details of my now 13-year old's life. Although he had quite a few infections when he was small, he really seemed to be healthy until the age of 8. Around that time, he started getting stomach aches and would vomit almost cyclically. It wasn't severe but became bothersome and slowly he started missing school due to the discomfort. His pediatrician ordered some tests and we were repeatedly told everything was normal. When the condition didn't resolve, it was determined that he had chronic constipation and his pediatrician made a remark about how my son was being dramatic. I began feeling like I was going crazy.

Cont.

He was missing so much school and although the medical professionals kept reassuring me there wasn't anything "scary" going on, I knew something wasn't right. The unknown is very scary.

I finally changed pediatricians around the same time his fevers began when he was 10. At first, it was considered recurring strep, but he would have episodes of a sore throat and fever without testing positive for strep, then scarlet fever, then it progressed to PFAPA. By that point, he was unable to walk unassisted during episodes (which were occurring every three weeks), had horrible mouth sores, 105°F (40.5°C) fevers and was severely nauseated refusing to eat.

A few months later, after a tonsillectomy, at the age of almost 12, we were referred to rheumatology.

They informed us that his CRP was high in a test, and they were surprised it had taken us so long to be referred. They ordered a small genetic panel, started him on medication and although his symptoms improved, he was still missing almost half of school, unable to participate in sports, missing out on birthday parties and activities. He had occasionally mentioned pain in his ankle, and although I brought it up often, it wasn't severe so was dismissed. We really like my son's rheumatologist. She is kind, knowledgeable, and good admitting when she doesn't know something.

In March, he started a flare that lasted 7 weeks.

He was so sick so after a few weeks I took him into the ER where he was dismissed. They said it was "sad" when kids have conditions like his because they can't help them. He started to lose hope, felt so isolated and the happy kid I knew was feeling broken. They finally did imaging of his foot, and it showed multiple stress fractures which had probably been there at least a year and they think has to do with his condition. The doctors are stumped and although he has seen more than 7 different specialists, he is still a mystery. We are overwhelmingly grateful for the FMF & AID Global organization.

My son looks forward to the zoom meetings with other kids with similar conditions. It has given him hope, community, connection and he doesn't feel alone. He has had to stop playing sports, find new activities (which are limited), there has been much medical trauma that has occurred, and he has suffered more than any child should ever have to. Being rare can be so hard but we hold onto the glimmers of happiness and hope.

NEWS FROM OUR AFFILIATES

Italy – Association Iris ODV

My name is Massimiliano Lucchetti and I represent the association "Iris ODV", established in March 2015 with headquarters in Casalnoceto, Italy.

It all started when the parents of a child with CAPS in Ukraine, contacted the Giannina Institute Gaslini in Genoa-Italy, asking for a consultation with Dr. Gattorno. Due to the language barrier, Dr. Gattorno asked my wife, if she could help as a translator. The outcome of the consultation confirmed the pathology which had already been diagnosed by a specialist in Russia.

After this experience and seeing the desperation and impotence of this mother, my wife and I decided to set up a non-profit organization to help this child. Through various initiatives and activities, we raised enough funds to cover the costs of the consultations at Gaslini, as well as the cost of the biological medication required.

Recently, the Iris Association changed its legal form from ONLUS has ODV. This change will enable us to help not only this one child, but also many other children with CAPS.

When the Covid Pandemic started, it basically blocked people's social life.

Recently, on July 15th 2021, we were finally able to resume our activities and organized a solidary dinner to start raising funds again.

September is "World Autoinflammatory Awareness Month" and soon we will be deciding how to best raise awareness, but of course always compatible with the new anti-covid regulations.

If you would like more information on our association, please visit www.irisonlus.org.

Italy – Association AMRI

Support group for young people with rheumatic diseases

In recent months, the first meetings of the group of young people suffering from autoimmune diseases organized by the Association for Childhood Rheumatic Diseases (AMRI) were held. Connected together, from various parts of Italy, a nice mixed group has formed, consisting of adolescents and young adults with childhood rheumatic and auto-inflammatory diseases: Juvenile idiopathic arthritis (JIA), connective tissue disease, fibromyalgia and dermatomyositis.

Although some pathologies have their own characteristics, such as the absence of a cure and chronic pain (in addition to being still little known and recognized), the group's reflection focused on some emotional experiences that transversally affect all rheumatic diseases.

Among these, for example, the impact of the disease on the quality of life, the limitations, the sharing (or not sharing) of the diagnosis with the group of friends and peers, the efforts related to the management of the disease (exams, hospitalizations, day hospital, therapies), the impact on school, social and self-image life.

The majority of children with intermittent symptoms report that they have often underestimated (and challenged) their disease, that they have had difficulty sharing the diagnosis with their peers (especially in the case of diagnoses made in adolescence) for fear of being treated "differently "or not being understood. The totality of the children with chronic symptoms, on the other hand, reported that they felt great anger over the disease that changed their life.

In this case, precisely because of the impact on everyday life, it was not possible to "hide" the disease, some report having found empathetic and understanding friends and teachers, while others felt little believed and welcomed.

The balance of this first meeting is positive because they have the desire to get to know each other and share unique experiences.

Among peers prevails that lead to forge ties with those who can understand them because they have lived similar experiences.

The initiative will go ahead in the ways that the participants themselves will choose in the coming months, it is always possible to join or receive more information by writing to psychologhe@amri.it.

Turkey – BEFEMDER

Behçet and Familial Mediterranean Fever Patients Association.

Our aim is to promote Behçet and FMF (Familial Mediterranean Fever) disease, to raise awareness in the health sector with social sensitivity and public support, and to carry out social and cultural studies that will make patients and their relatives feel better and happier.

Our current support projects are:

1- During the Covid-19 process, we organized informative online patient meetings for adult patients via Zoom and with the participation of a rheumatologist/urologist.

2- The "RaDiChal" project competition was organized for the second time this year by Üsküdar University Transgenic Cell Technologies and Epigenetics Application and Research Center (TRGENMER), 13 teams from 13 universities will compete for a FMF treatment. The aim of the project is to find gene therapies for rare diseases with genetic backgrounds. Dozens of biologists and geneticists are participating in the competition. As an NGO, we give all our support to these projects.

PARTNER GROUP

Morbus Behçet in the USA by Beth Hope

My journey began from childhood. I was sick often and couldn't quite figure out what was wrong with me. My main symptoms were joint pain, mouth ulcers, eye inflammation, stomach ulcers & issues, migraines & fevers with heavy fatigue. It took about 20 years of doctor appointments, labs & hit or miss guessing to finally be diagnosed with Behçet's disease. It's been a rocky road & I have become an advocate for my community to help others be diagnosed quicker & help provide education, empowerment & support. I run the largest support forum for Behçets in the world, called Behçet's disease: You are not alone!

Authors

The articles in this Newsletter have been written by:

Malena Vetterli, Paul Morgan, Janine Tschan, Rachel Rimmer, Ellen Cohen, Lisa Scott, Mary Frank and various anonymous patients.

The patient stories included in this newsletter, have been provided to the FMF & AID Global Association voluntarily. All patient stories are being published with the written consent and permission of the patients/parents. The patients/parents who shared their stories, were requested to do so anonymously, and without identifying names of hospitals, doctors or medications. The FMF & AID Global Association has also received written parental consent for the images displayed in this Newsletter. All other images found in this Newsletter have been bought from stockfresh.

The translations have been done internally by volunteers of the FMF & AID.

List of disease abbreviations used in this Newsletter

CAPS	Cryopyrin-associated periodic syndromes
FMF	Familial Mediterranean Fever
HIDS/MVK	Hyper IgD syndrome / Mevalonate kinase deficiency
NOMID/CINCA	Neonatal Onset Multisystem Inflammatory Disease
	Chronic Inflammatory Neurological Cutaneous Articular Syndrome
PAPA	Pyogenic arthritis, pyoderma gangrenosum, and acne
uSAID	Undifferentiated autoinflammatory disease