FMF & AID Newsletter

2022 Edition

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

IN THIS ISSUE

Editorial

By Malena Vetterli

Dear Readers,

As summer holidays are concluding, we are excited to embrace the upcoming September WORLD AUTOINFLAMMATORY AWARENESS MONTH, as we continue to support thousands of patients internationally, who are living with the burden of autoinflammatory conditions. FMF&AID's efforts enable our affiliated autoinflammatory associations to unify and raise awareness about these diseases.

As a patient organization, we continue offering direct support to patients via a variety of channels including the Zoom support groups, therapy sessions for children and our medical assistance program. These have been some of our top priorities for the year 2022. All this help wouldn't have been possible without the financial support received from generous foundations and private donors.

Our volunteers have been working on a series of informative brochures that have been reviewed by medical professionals and will be a strong tool for patients' self-advocacy use.

FMF&AID continues its ongoing work with ImmunAID, a European research project, as well as officially representing patients in the European Reference Network for autoinflammatory diseases (ERN RITA).

cont. Editorial

This edition of the newsletter also includes our continued sharing of patient and family journeys. We encourage our readers to share this newsletter with friends and acquaintances to raise awareness of these rare innate immune diseases.

We would like to announce our paper, The FMF&AID Survey - A Patient Organization Driven Survey for Autoinflammatory Diseases, which was accepted by EULAR as a poster abstract and published in the Annals of the Rheumatic Diseases in May 2022. FMF&AID is proud of our hard work developing this paper in collaboration with esteemed autoinflammatory physicians. The data utilized was based upon our 2021 survey and we are thankful for everyone's contribution.

Finally, an update on the situation in Lebanon from our previous newsletter. With the help of our local representative in Beirut, FMF&AID purchased over 1,000 boxes of colchicine and had them delivered to local pharmacies throughout Lebanon, where many patients were able to collect a 6-month supply at no cost.

We hope you find this newsletter informative, helpful, and thank you for your continued support.

Warm regards,

Malena Vetterli, Executive Director and the FMF & AID Team

September is World Autoinflammatory Awareness Month

As September approaches, FMF & AID again wishes to highlight the lived experiences and speak out on behalf of the autoinflammatory patient community with regards to the impact on the quality of life of these diseases. These debilitating conditions continue to profoundly affect patients on a daily basis and remain invisible, and often result in delayed appropriate care. This is especially pervasive in the adult patient population.

We continue to highlight the challenges patients and their families endure along their autoinflammatory journeys. There is an increasing global health emergency due to the lack of autoinflammatory specialists to treat our ever-growing pediatric and adult patient population. Despite the tremendous gains in genetic sequencing, funding, research, and the countless published articles on these innate immune issues, few practicing medical specialists and general practitioners are aware of how autoinflammatory diseases manifest in all age patients.

Additionally, since status of gene pathogenicity takes years to be documented and may be different (benign in some and disease-causing in others) in various populations, many patients continue to fight for treatment despite their negative genetic results. Patients should encourage their treating physicians, to request and review genetic benign variant reports, where available, for clues that may be indicative of their disease. Patients' symptoms should not be dismissed nor based upon a piece of paper, be it a negative genetic report or inconclusive lab results.

Endorsed by the European Reference Network RITA

cont.

While we applaud the ever-growing effort to identify novel autoinflammatory diseases, FMF caused by the MEFV mutation has been sequenced and now studied since the 1990's with literature dating back to the 1950's. Most concerningly, we are still witnessing MEFV mutated patients struggling to receive medical help, despite having the most widely known of all autoinflammatory diseases.

There has been little advocacy to improve medical awareness outside of university and government-funded research initiatives, which have failed to serve the majority of patients. FMF&AID continues to address these issues on many fronts, attempting to close the gaps between advances in genomic technology, patient diagnosis and medication access.

FMF&AID continues to receive urgent requests for care and treatment referrals. It is a growing concern as more patients are identified with autoinflammatory diseases in countries where there is no access to appropriate biological medications.

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

FMF & AID Milestones 2022

As FMF&AID reflects back on this past year, we believe our work has positively impacted our global patient community, despite the COVID-19 pandemic, as we move forward to accomplish our outreach goals. Our social media presence remains strong as we maintain online groups in 12 languages, the largest of these is our Arab-speaking group with over 34,000 members. We welcome new members daily to our many groups. It is also our third year offering the Medical Needs Assistance Program, addressing patients' individual medical challenges in an expedited manner.

Currently, we are developing a series of comprehensive brochures to enhance patient communication while educating their treating physicians, employers, school authorities and family members with regards to their unique autoinflammatory disease needs.

Additionally, our abstract "The FMF&AID Survey - A Patient Organization Driven Survey for Autoinflammatory Diseases" has been published by EULAR in May 2022, in collaboration with several key doctors. The abstract based on our 2021 survey, provided fantastic information and we are appreciative of our patient community's participation and feedback. We thank everyone who contributed their time.

Many of our other additional activities are presented in this newsletter to provide our audience with timely and updated information.

Interview with Prof. Bruno Fautrel, Clinical Coordinator of the ImmunAID consortium

by Fredéric Peyrane

Bruno Fautrel is Professor of Rheumatology. He is the head of the Rheumatology Department at the Pitié Salpêtrière University Hospital. He is also the co-director of the IMIDIATE clinical research network, a nationwide network dedicated to clinical investigations in immune-mediated inflammatory diseases, as well as the clinical coordinator of the ImmunAID consortium.

FMF&AID: What is ImmunAID and what is its aim?

Bruno Fautrel: ImmunAID is a research project designed to improve the diagnosis of systemic autoinflammatory diseases (SAID) and the medical care of patients. We are building the world's largest SAID cohort, thanks to the involvement of more than 30 centres in 11 countries across Europe. Our first goal is to identify a common signature to SAID, enabling rapid differential diagnosis. We also would like to characterise, in depth, the specific pathogenic pathway of each studied disease and identify the corresponding biomarkers. Additionally, we want to build a robust algorithm enabling reliable triage among the whole SAID spectrum.

FMF&AID: Could you tell us a bit more about how ImmunAID is different from other research initiatives tackling autoinflammatory disorders?

BF: Firstly, because of the main target: we want to shed light on these sets of SAID where no genetic mutations are known, which does not preclude us from improving the understanding of monogenic diseases. Secondly, because of the methodology: we will analyse the samples using state-of-the-art technologies to generate an unprecedented amount of 'omic' data (genome, transcriptome, proteome and microbiome) without any a priori hypothesis. In parallel, we will take a deep dive into already known or suspected molecular mechanisms, such as the inflammasome, lipid mediators and other agents of the immune system (i.e. cytokines, etc.). All the data generated will constitute the first ever "SAID immunome", which will be subsequently subjected to the latest artificial intelligence and modelling techniques to reach our goals.

FMF&AID: How far along are you with the project and have you been impacted by the COVID-19 pandemic?

BF: The project started 4 years ago with 34 clinical centres actively recruiting. We have been able to enrol about 300 SAID patients and another 120 parents to improve the relevance of the genomic analyses. Most samples have already been analysed biologically and the first computational results have started to emerge for the sub-categories of diseases with a significant number of patients. However, it is true that the COVID pandemic has had a tremendous impact upon us.

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Hospitals have been overwhelmed, both in medical care and administrative departments, patients have rightly postponed their visits, and even the research activities in the labs have been disrupted by the multiple lockdowns and illness of the researchers themselves. With the situation improving on the health front, we are returning at full speed to achieve ImmunAID's goals.

FMF&AID: How do you see the role of patient associations in such project?

BF: Patient associations are essential. FMF&AID has been a member of the consortium since the project started and it has helped us a lot to reach out to other patient associations across Europe, to raise awareness on the project, on the recruitment process, and to keep a focus on patients' concerns, both in terms of recruitment constraints and the project's objectives. While clinicians can sometimes be locked into their jargon and concepts, it is good to have someone who brings us back to the everyday concerns of the patients.

ImmunAID Participation Opportunity

ImmunAID is still actively recruiting participants at their many sites throughout Europe. The ImmunAID research project is trying to identify biomarkers that could help diagnose autoinflammatory diseases faster. If you are interested in contributing to this research, please visit www.immunaid.eu. If you wish to participate in this study, please send an email to info@fmfandaid.org.

CHILDREN and ADULTS diagnosed with any of the following diseases, and who meet the study criteria, can apply to participate and help with research on:

- ✓ FMF
- ✓ TRAPS
- ✓ CAPS
- ✓ HIDS
- ✓ CRMO
- ✓ Pericarditis
- ✓ Still's disease
- ✓ Schnitzler
- ✓ Vasculitis (Kawasaki, Behçet, Takayasu)
- ✓ Inflammation of unknown origin

There are 37 participating hospitals from the following 11 countries:

- Belgium
- France
- Germany
- Greece
- Italy
- Netherlands
- Slovenia
- Spain
- Switzerland
- Turkey
- United Kingdom

ImmunAID in a nutshell

We build
a patient cohort

Patients with active SAID are screened by physicians

Samples and clinical data are collected and anonymized

Samples from all over Europe are stored in a biobank and further dispatched to biolabs

We do
biological analyses

Exploratory unbiased approach

Exploring at large scale new causes of innate immune dysregulation

DNA
Identify mutated genes...

RNA
...and the impact on RNA levels

miRNA
Map RNAs interfering with protein expression

Proteins
Identify changes in protein expression and distribution

Microbiota
Identify microbiome profiles activating inflammation

Hypothesis-driven approach

Exploring already suspected pathways and effectors underlying inflammation

Inflammasome
Inflammation activation: Unravel the dysregulation of inflammasome function

Lipid mediators
Inflammation resolution: Explore possible defects of lipid mediators

Cytokine, immune cells, etc.
Inflammation regulation Decipher the role of key immune system effectors

We do
computational analyses

Combine hypothesis-driven and free-data clustering, integration, modeling and validation

Therapy groups for English & German speaking children

Since 2020, the FMF&AID offers interactive therapy groups for children between 4 and 14 with autoinflammatory diseases on a monthly basis. These groups are moderated by professional therapists/psychologists. Currently, 60+ children have been registered by their parents, and on average, 18 to 20 kids participate in the English-speaking group every month. Additionally, we also offer a German-speaking group.

Living with an autoinflammatory disease is burdensome. Many children have difficulties coping with the disease, especially the recurring symptoms such as high fevers, pain, mouth ulcers, etc and often miss out on childhood activities. Being sick on a regular basis means being away from school, missing out on social engagements such as birthday parties, sports, as well as having limited friendships. Furthermore, topics including trauma caused by doctor visits, medical procedures, blood draws, injections, etc, are all captured in the group discussions. More importantly, kids learn how to advocate for themselves. In the July group, the kids learned about them being the "experts" of their own bodies, and how to approach the hospital/doctor's office. They also learned to concentrate on good emotions such as love, acceptance, positivity, and strength.

The therapy group brings these often-isolated children together. It demonstrates that they are not alone, and they belong to a group of very special children, our little superheroes!

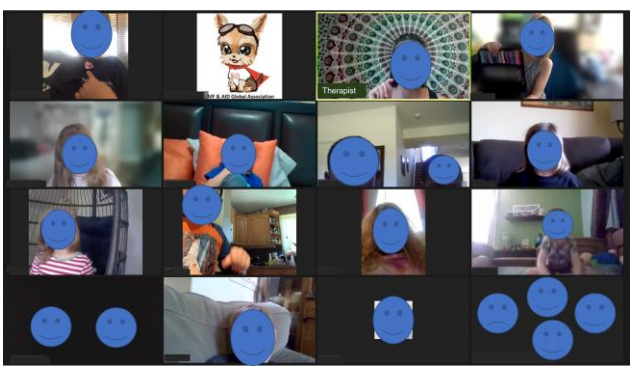
It is important to us that these children make great connections with others like themselves. The therapy group is fully funded by FMF&AID, thanks to our generous donors, thus removing the financial stress from the parents, and allowing any interested child to join the group.

Parent feedback:

"I wanted to let you know that he's had a blood test since our last meeting, and he did such a great job advocating for his body. He asked for the arm he wanted to be pricked and asked to have the nurses count with him to 10. He did so well, and I know it's because of the last group session."

"Our daughter truly loves joining this group. I love that she feels so accepted and open, as it's something that can cause herself defeat and low self-confidence a good amount of time. I am extremely happy (and maybe a little emotional) to see that she had in fact written lists during group going along with what you were discussing. I really am so appreciative of this group and all it brings into her world. "

"Words cannot express how wonderful it was to hear our daughter interacting with all of you. She was late getting on because she had a meltdown. She's really struggling lately, but we heard smiles and a lift in her voice. Thank you for what you do!"



Patient Tools – Self-Advocacy Brochures

by FMF&AID

FMF&AID encourages our patient community to advocate for themselves and their children by empowering them to communicate effectively the issues they encounter from having an autoinflammatory disease. FMF&AID is developing a series of informative brochures, reviewed and endorsed by autoinflammatory specialists, to educate regarding relevant topics that impact patients medical and daily lives. These tools will help raise awareness, educate, and promote discussions with patients' physicians and healthcare practitioners, teachers, administrators, and family/friends.

To date, five brochures have been completed and will shortly be ready for release to the patient community in several languages. The goal of using these comprehensive and easy-to-read brochures will help autoinflammatory patients to provide easily accessible, yet comprehensive, and physician verified information to various audiences encountered frequently. FMF&AID hopes to reduce the communication challenges and stress of the patient, while helping to educate those lacking understanding on any given topic relevant to autoinflammatory patients' care.

Brochure topics that have been completed include Colchicine, School, Pregnancy, Pain, and Familial Mediterranean Fever. Additional brochures for other diseases and topics are being developed.

If there is a topic you think would be brochure worthy, please do not hesitate to make suggestions by contacting FMF&AID.

RESEARCH Review

by Ellen Cohen

FMF&AID closely monitors global research and data on all topics related to innate immune issues and autoinflammatory diseases. We share articles in our various groups to help patients have a more comprehensive understanding of important topics and to facilitate exchange should patient wish to share research with their treating physicians. For those who are interested, several recent publications have been included for your information and review.

We strongly believe that patient education is a critical key to successful self-advocacy.

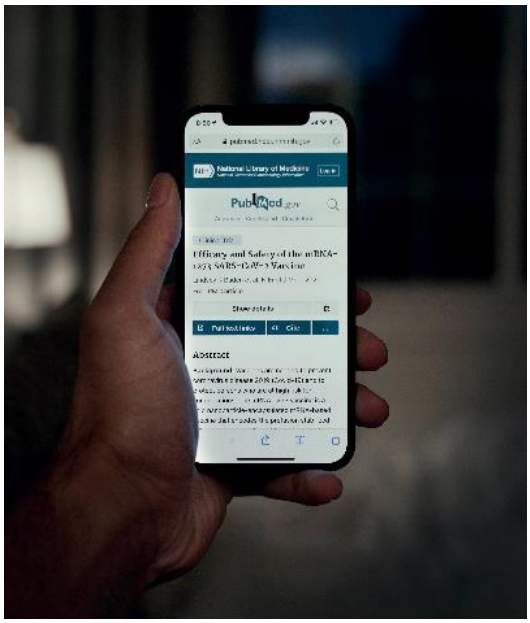


Photo by Karthik Sridasyam on Unsplash.

Papadopoulos Vasileios P., Antaniadou Christina, Ritis Konstantinos, Skendros Panagiotis. MEFV Mutations in IBD Patients: A Systematic Review and Meta-analysis. J Gastrointestin Liver Dis, March 2022 Vol. 31 No 1: 85-97. https://www.jgld.ro/jgld/index.php/jgld/article/view/4070/1792

Tian Yi, Wu Bingxuan, Peng Linyi, Wang Jian, Shen Min. Three Chinese pedigrees of A20 haplo-insufficiency: clinical, cytokine and molecular characterization. Front. Immunol., 26 July 2022 https://doi.org/10.3389/fimmu.2022.955079

Lee J, Bizzocchi L, Jain R, Tagoe CE. An autoinflammatory syndrome with compound heterozygous MEFV and NOD2/CARD15 gene mutations successfully treated with tocilizumab. Rheumatol Adv Pract. 2022 May 9;6(2). https://www.ncbi.nlm.nih.gov/pmc/articles/PMC9113507/

Serarslan G, Cura SE, Kimyon G, Üçgül G, Karadağ M. Association of MEFV mutations and vascular involvement in Behcet's disease: a study from Hatay, Turkey. Reumatologia/Rheumatology. 2022; 60(3):220-223. doi:10.5114/reum.2022.117843. https://www.termedia.pl/Association-of-MEFV-mutations-and-vascular-involvement-in-Behcet-s-disease-a-study-from-Hatay-Turkey,18,47408,1,1.html

Blank N, Schuetz C, Henes J, et al POS0221 Long-Term Efficacy and Safety of Canakinumab in Patients with TRAPS (Tumor Necrosis Factor Receptor-Associated Periodic Syndrome) – Interim Analysis of the Reliance Registry. Annals of the Rheumatic Diseases 2022;81:347. https://ard.bmj.com/content/81/Suppl_1/347.2.info

François Rodrigues, Laurence Cuisset, Bérangère Cador-Rousseau, et al. AA amyloidosis complicating cryopyrin-associated periodic syndrome: a study of 86 cases including 23 French patients and systematic review, Rheumatology, 2022; keac145, https://doi.org/10.1093/rheumatology/keac145

Gaslighting

by Ellen Cohen

Gaslighting is a form of psychological abuse in which a person causes another person, over an extended period of time, to question their own sanity, thoughts, or perception of reality. People who endure gaslighting may feel confused, anxious, or as though they have lost their confidence and self-esteem.

Medical gaslighting

Medical gaslighting is when a medical professional dismisses a person's health concerns as being the product of their imagination. They may tell the person their symptoms are "in their head" or label them as a hypochondriac. A worst-case scenario is when a medical professional accuses a parent of Munchausen's syndrome and/or tries to have the child removed from the parents' care.

We have many parents in our community that have obstacles in getting their children diagnosed due to medical gaslighting and physician lack of knowledge. FMF&AID is very concerned about this issue and feels that it deserves further discussion and attention to help our community tackle this problem.

An important question to ask is: why are the medical communities for both pediatrics and adults failing to recognize periodic fever and innate immune dysregulation syndromes?

The answer to this question is multifaceted and includes lacking medical awareness, knowledge, education, and low prevalence of patient numbers. While there may be few patients due to the rare classification of these diseases, there are thousands of papers, based upon millions of dollars' worth of research, dating back to the 1960's, which provide substantial evidence and information on these conditions.

While physicians may not have the time to substantially read about autoinflammation, there is no reason to deny or dismiss patients who present with symptoms, historical data, and photo diaries, demonstrating these life-altering problems. Doctors treating any patients, should be able to recognize, when they lack expertise, the need to provide referrals to a more appropriate specialist or to take the time to educate themselves and offer proper care to the autoinflammatory patient.

cont.

Several parents in our patient community have been labelled as likely having Munchausen Syndrome or a related condition, where the parent or caregiver to an autoinflammatory child has allegedly fabricated physical symptoms in their child.

As a result of medical gaslighting, parents will often pursue repeated contact with various specialists and medical professionals in an attempt to get their child appropriate diagnosis and treatment. FMF&AID is concerned with the outcome of these cases.

School gaslighting

School gaslighting occurs when teachers, administrators or school health professionals ignore or dismiss a child's health concern as being unimportant. This may happen despite parent request for school accommodations to help with autoinflammatory symptoms presenting during the school day. The child may be labelled as attention seeking, lazy, unwilling to participate in school activities.

Photo by Kelly Sikkema on Unsplash

Parents too may be labelled by school officials as overprotective. These types of interactions between the child and school staff may lead to emotional trauma for the child and family.

Friends and family gaslighting

Relationship gaslighting occurs when friends and family refuse to acknowledge that a symptomatic child actually has a medical condition. This type of gaslighting is more personal than the medical variety, as parents have to deal with relatives who often impede medical care, belittle the caregiver, ignore illness clues, and bully family members who are trying to help.

Collectively, adding stress to the caregiver of the child who has autoinflammatory disease. Consequences of this may impact the family unit, resulting in divorce, custody issues, family abuse and violence.

Recommendations:

- If the treating physician is not helpful over a reasonable period of time, see another physician.
- Have treating physician write a protocol letter for emergency dept visit, dentist, school and travel.
- Document all illness manifestations: diary of symptoms, photos of skin rashes, mouth ulcers, etc.
- Periodic review of patient's electronic medical record for accuracy.

cont.

Quotes from our community:

I had an internal medicine MD tell me "it was all in my head". Also told by my Pediatrician that "because I'm anxious, they would do genetic testing", child had been to ER 6 times in last 7 months.

My ex-husband got custody of my kids because he convinced a judge I had Munchausen by proxy even when a medical doctor said it was truly a sick child.

I can't tell you how many doctors and nurses downplayed our symptoms, blamed the thermometers (I had 3 different kinds), and told us it's just a virus (during quarantine). Many family members thought I was crazy (maybe they still do). I'm so thankful that we have an excellent doctor that is compassionate and is providing a treatment plan.

I have also been told I'm an overprotective mother and he's fighting for my attention. My son was also forced to admit he was faking it. I was questioned by school health and treated like a mad woman who was keeping him at home against his will.

Photo by Vitolda Klein on Unsplash

Barriers for Autoinflammatory patients in the UK

by Rachel Rimmer

Rare Autoinflammatory Conditions Community UK (RACC-UK) continues to be concerned about the UK's "crisis of diagnosis" for its autoinflammatory patient community. This is despite the COVID pandemic coverage introducing common terms, which are also used in innate immune monogenic diseases, such as: cytokine storms, inflammatory conditions, pericarditis, and fever issues.

Despite having the above terms now more commonly known and used in media describing COVID, the doctors still don't apply them properly to autoinflammatory diseases, thus doctor continues to lack basic knowledge, gaslight patients, and roadblock patients' referral requests for secondary specialist care (rheumatology, immunology, etc). Patients now face additional risk factors as E-Consults (telehealth) are declining and being phased out of practice, which drastically impacts all rare disease patients.

Acknowledgement for autoinflammatory disease investigation in many paediatric cases prior to the pandemic was highly problematic, and continues to be.

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Countless parents are still mistakenly told "it's just a viral infection and children get sick" despite these patients having recurrent fever patterns, rashes, abdominal pain/GI issues (even prior to COVID-19) and while isolating at home without attending daycare or school.

Our organization viewed the circumstance of these lockdowns as "proof of concept" that our community of children with repeated symptom flares, were not caused by shared environmental infection transmission, rather presenting from innate disease inheritance. Thus, we began a support campaign to write and disseminate letters on behalf of our patients and carers to educate their General Practitioners. We took this unusual opportunity to inform multiple doctors as to the symptoms of autoinflammatory conditions and to highlight that lockdown actually lowered the burden of viruses and bacterial infections, thus providing supporting proof that patients were actually disease burdened from genetic mutations.

In countless cases RACC-UK, requested that patients be referred to Rheumatology and/or Immunology services for extensive blood and genetic testing to aid a correct diagnosis of Autoinflammatory conditions, despite the extended back-up of COVID-related lab issues. Despite our efforts, the communication response has been sub-standard and there has been little feedback from contacted physicians.

Collectively, causing long-term damage (both mental and physical) to our patients who remain undiagnosed and without timely/effective treatment. We continue to advocate for our patients and ask everyone impacted to please document symptoms via written and photographic diaries.

The NHS has funded a few new specialized treatments and RACC has been privileged to participate in consultations and technology appraisals to enhance their availability, despite cost and stringent diagnosis criteria. While this has been a positive step to further treatment options, there have been many worldwide shortages of these medications due to continued supply chain issues as a result of the pandemic. This is not specific to the UK, rather a global problem and has impacted our patients being properly and timely treated.

Finally, while one of our UK autoinflammatory fever centres is fully booked until December 2022, we continue to advocate for all UK paediatric and adult autoinflammatory patients to get the qualified care they deserve.

Photo by Martha Dominguez de Gouveia on Unsplash.

School Robot

by FMF&AID

The FMF & AID Global Association came across a smart product that could make things easier for children of school age, who often miss school due to them being absent, as a result of flaring or not feeling well. Since we are not advertising this product, we are not mentioning its name nor the company selling it. We simply believe it's worth mentioning that such a product is available, because many children with an autoinflammatory disease would certainly benefit from it, if it was available at their schools.

Many children suffering from an autoinflammatory disease have debilitating symptoms, preventing them from attending school on some occasions. Sometimes it will be due to high fevers, and other times it will be abdominal pain, joint/leg pain, headaches, fatigue, etc.

While some children will only miss school a few days a month, there are others who will be away from class for several weeks at a time, and on repeated occasions. This situation is not easy for both, the child and the school. The longer a child has to stay home, the more isolated they will feel due to a lack of social contact and interaction with other children. We understand that depending upon the severity of the symptoms the child may present, this robot may or may not be of any help, because the child may be incapacitated until the flare is over.

However, it can be quite helpful in other cases where the child is simply exhausted or unable to walk but is able to listen to the class.

This small robot basically enables children to take part in class, as if they were there among their friends, and can also participate actively. It basically takes the place of the child in the classroom, allowing the child to follow the class from their home or hospital.

It's encouraging to see that some companies are developing products with the minorities in mind, by addressing the social and educational needs of sick children and giving them a sense of belonging and normality.

We know that this robot is being used successfully in several schools across Europe. It's not exactly cheap, and for this reason, it makes more sense for schools to buy it so that it can be used by several children, providing they are not all sick at the same time.

Medical Alert Service Dog

by Brooke Hightower-Foster and Nick Foster

Due to our children often being sick with fever caused by their autoinflammatory diseases, we decided to see if there were other options that could give us early warning, since traditional medicine was not enough. This led us to see service dogs, in particular, to see if we could find a medical alert service dog trained in fever detection, to assist one of our daughters who needs help the most.

After much research, we narrowed the breed down to the Weimaraner, for being a versatile hunting dog known for its ability to detect all types of scents. It's also known for having an outgoing personality, being family friendly, and a very intelligent working dog. It took a few years to find a breeder that raised Weimaraners to become service dogs. The breeder is known for working internationally and sending service dogs all over the world.

Our daughter's Weimaraner uses its sense of smell, which is more sensitive than a human's, to detect the unique chemical pyrogen, which is known for producing fevers. According to a medical study on the accuracy of dogs in scent detection, it stated that they are even more accurate/better than sensors. We worked with a trainer and then trained our Weimaraner to do fever detection as a medical alert service dog. The dog now alerts us, to whenever our daughter has an imminent fever.

This enables us to give her medication early enough to prevent or lower the symptoms, as well as make the necessary accommodations.

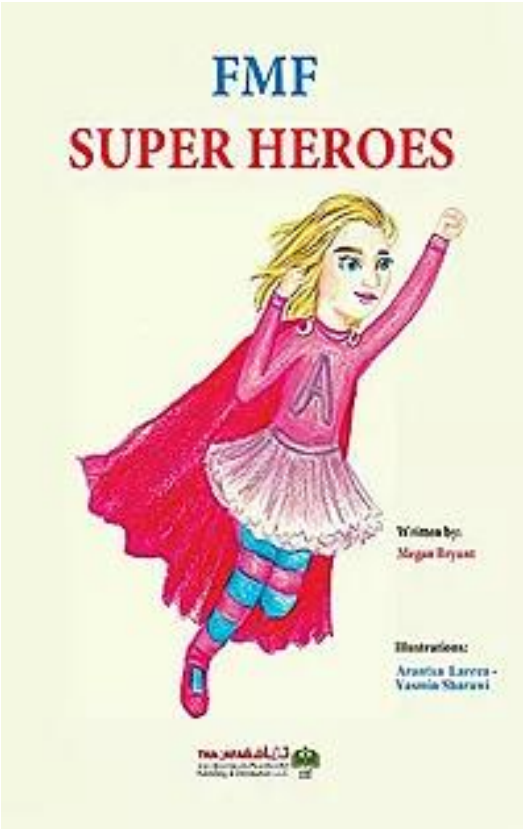
Shadow, our daughter's service dog, is a very special dog. When we brought her home, she instinctively alerted us to one of our daughter's fevers immediately without having been trained. Initially, we didn't understand what she was doing when she went to our daughter's side and cried for a long time. Finally, we checked our daughter's temperature, and she had a high fever. We thought this was a coincidence, but Shadow kept repeating the same behavior with each of our daughters' fevers. It was amazing to watch! Shadow has an innate ability to detect fevers. Side note, Shadow is only trained to alert one of our daughters, but she is so intelligent that she checks each of our daughters and alerts us when they have a fever. Her sense of smell is so accurate that she can alert us to an oncoming fever, even up to a few days in advance.

Shadow is also trained to do gluten and allergen detection, which is a different type of training from the fever detection. Training is an ongoing and daily process that has to be maintained to improve the dog's skills and behaviors to build her cue recognition, obedience, manners, and more.

cont.

If Shadow does not get enough down time from working, then she could miss cues to alert us of a coming fever. Play, relaxation, and sleep are so important to Shadow, that she might not be able to work effectively and use her obedience training, or even worse, lose her ability of impulse control, if she doesn't have sufficient down time.

Lastly, it is important to educate people on how to approach a service dog when they are "working". We try to educate people whenever they see a service dog, especially when they are wearing its special vest or leash on, to never distract the service dog by touching or talking to it. This is because the service dog may miss an alert or even worse not be able to independently think to perform tasks that are needed in emergency situations.



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's work. Links to our books are found on Amazon:

https://www.fmfandaid.org/books

The Benefits of Mindfulness

by Cindy Myers

For your stress and relaxation, I would like to introduce myself as a physical therapist with 20 years' experience as a Mindfulness teaching expert and autoinflammatory patient. I am offering our autoinflammatory community some useful tips on how to adapt Mindfulness strategies into your daily routines to help alleviate tension, pain, anxiety, and nervousness. These calming strategies may help patients and caregivers be more productive, while remaining focused and centered in tackling life's work.

Definition of Mindfulness

Mindfulness is the human ability to be aware of the present moment, of where we are, and what we are doing, without thinking about the past or worrying about the future. If we concentrate our thoughts, senses, and energy on our PRESENT life, in a positive way, it will improve our relationships, mood, health and brain function.

Introduction to Mindfulness

The nervous system is the major controlling, regulatory, and communicating system in the body. When a patient goes through a stressful situation, the panic/alert state of the nervous system is activated. This alert state is only meant to be temporary to help the individual overcome whatever life event they may be experiencing, but after a short time, it should automatically shift back into the calm/normal state.

In some cases, the individual remains in this permanent alert-like state, causing negative emotions such as panic, stress, pain, etc.

Mindfulness methods/activities

There are many methods of calming including: rocking, drawing or coloring, listening to music, warming drinks, deep breathing exercises, light walking, Yoga, Tai Chi, massage, and meditation.

Practicing Mindfulness is very individualized and what works for one person may be different for another. It is productive to trial various methods to see which one is most impactful. It may be beneficial to practice these techniques and teach children or other family members these adaptable skills. Mindfulness activities should be practiced on a daily basis for at least 15 minutes to be beneficial. For more detailed information, please contact me at cmyers@in-motionpt.com.

Photo by Sixteen Miles Out on Unsplash.

PATIENT JOURNEYS

(written by patients or caregivers)

FMF in Austria

I want to share my story and also give you some hope, which I got back thanks to the FMF&AID Association. After 29 years of searching for answers, and 29 years of suffering, I received my diagnosis of Familial Mediterranean Fever (FMF) last year. Actually, it was only "thanks" to my 4-year-old son, who is, unfortunately, also affected like himself. I was happy when I received my diagnosis because I finally knew what I had. However, I never expected the problems I was going to face afterwards, here in Austria.

My childhood and youth were characterized by me being constantly ill. Due to my school absences, I almost got kicked out of school. I often suffered from high fevers, tonsillitis, abdominal pain, and a therapy-resistant iron deficiency anemia (including all the problems that this entails). I struggled to cope with my everyday life and spent a lot of time with doctors and being sick. As a teenager, I had a doctor who took me seriously and was surprised by my blood results and symptoms. Despite his intensive efforts, he never found the cause of my illness. Once, I also had a referral for an appendectomy, but then the hospital concluded that all I had was constipation. I was also sent to neurologists and internists, but nobody ever found anything.

Since the doctors didn't know what I had, it was easier to blame everything on my period.

In 2014, I was again hospitalized due to severe abdominal pain and cardiovascular issues. The diagnosis was iron deficiency anemia and histamine intolerance, which allegedly were responsible for all my symptoms. Then in 2016, when I became pregnant, I also had the Epstein Barr virus. Then my symptom presentation changed, and the frequency of my flares increased. Ever since, I have painful lymph nodes, massive pain in my limbs, neck and back, as well as severe pain in my joints, arms and legs. I have had to be hospitalized several times in my life, and twice, I was suspected of having meningitis, which required lumbar punctures.

cont.

Looking back, my inflammation levels have always been massively elevated during my flares. However, due to my blond hair and blue eyes, nobody ever thought of me possibly having FMF.

In 2017, my son was born. He used to cry daily for more than 12 hours straight. During the first 8 months of life, he hardly slept. He always had a bloated stomach and was often sick. As soon as he was able to communicate, he started pointing at his stomach. He had recurrent urinary tract infections, which are atypical for a boy, as well as chronic bronchitis, for which he had to be hospitalized several times. He also suffered from therapy-resistant iron deficiency anemia, like me. His symptom presentation is not the typical one with flares. Instead, he presents with chronic pain, which is always accompanied by respiratory issues. He has abdominal pain every day, as well as inflamed lymph nodes. Unfortunately, he only managed to attend kindergarten for half a year because he was sick the rest of the time. In 2020, my son had his first clear flare with a very high fever and his (daily) abdominal pain. I took him to the hospital and his appendix was removed. Parallel to my son's unknown disease, the doctors kept looking for the cause of my symptoms. We both show elevated inflammatory markers and the same symptoms (at least until my pregnancy).

Following a capsule endoscopy and celiac disease testing which did not reveal any information, the endocrinologist decided to order a genetic test of the MEFV gene. Three months later the result came back as positive for Familial Mediterranean Fever. I requested the children's hospital to run the same test on my son. The result was the same as mine. Unfortunately, this is when our odyssey really started. My son was sent from the children's hospital to an "alleged" FMF specialist. We were told that, due to us being Austrians and without having typical flares (fever with abdominal pain), and symptom-free intervals between flares, it is very unlikely that we have FMF, and we were both immediately dismissed and sent away without any treatment.

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According to some doctors, heterozygous patients are not symptomatic. Nevertheless, I was prescribed colchicine. I was very happy but at the same time very concerned because so many "FMF experts" said to be careful with colchicine and its side effects. Unfortunately, despite taking it for several months, it didn't work for either of us.

I have never been able to work full-time in my life. I had a lot of part-time jobs, all of which I lost due to this disease. I also tried to study, but I was very often ill, tired, and weak. My circle of friends became smaller and smaller because I am always ill. I was also diagnosed as having a psychosomatic disorder. There are so many social and financial problems that this disease brings with it. Unfortunately, despite the diagnosis, nothing has changed, because we are still not taken seriously by the people who matter when it comes to sickness/ rehab/care allowance.

The comprehensive accusations affect all areas, but nobody inform themselves about the disease or at least look at our medical reports. This is absolutely incomprehensible to me, since the FMF&AID, for example, does such great educational work and gives out such informative brochures.

Unfortunately, it has not been possible for me to work for a long time due to the increasing intensity of my son's and my disease, sometimes triggered by various factors such as stress, physical activity, infections, cold, heat, etc.

I came across the Facebook page of the FMF&AID association while looking for other affected people. I was so grateful to finally be listened to, taken seriously, and understood. I was provided with scientific articles, but at the same time I was encouraged not to give up when it came to treatment and the authorities. I was networked with specialists in Germany, and I was even offered financial support from the association. Honestly, I first thought all this was too good to be true. To my surprise, the doctors contacted me immediately, I sent them our medical reports and they looked at everything and got back to me shortly afterwards. It all happened so quickly, in a friendly manner, very understanding and unbureaucratic, unlike all my experiences here in Austria. I thought there must be a catch.

cont.

When I finally arrived at the University Hospital in Erlangen - Germany, I could hardly believe it. Both doctors, the adult rheumatologist, Dr. Jürgen Rech, and the pediatrician, Dr. Tobias Krickau, took a lot of time to look through all our medical records, let me explain our symptoms in detail, and also explained clearly and in detail their recommendations. They also wrote an action plan with us and decided the next treatment steps. I also got information material for the kindergarten (and later) the school, which saves me long explanations.

I am used to the fact that there is hardly any time for patients in hospitals, my symptoms being dismissed and a lack of understanding. This visit has given me renewed hope for our situation, and also for the future of my son, whom I want to spare from the long ordeal I have had up until now.

I am especially glad that we are now in competent hands and that our illness, and everything that goes with it, has now been confirmed by specialists, which hopefully will end or at least shorten the long discussions with authorities and experts.

I would like to express my deepest thanks to Dr. Rech and Dr. Krickau, and of course to the FMF&AID association, which supports us on so many levels, be it the online help group for children and those for adults (both very healing and recommended!), specialist literature, networking with doctors or help with other concerns.

I can't say it often enough: THANK YOU! We are finally on the right track. If you want to shorten your patient journey, do it with the FMF&AID association!

uSAID in Chile

I am the mother of a 9-year-old boy, and we are from Chile. In 2020, my son was diagnosed with an autoinflammatory disease, after a genetic test with a mutation of uncertain significance. At first, I was told it might be PFAPA, and when this was mentioned to me, my world fell apart. I didn't know what PFAPA meant, much less autoinflammatory diseases. I searched for information on the internet and managed to find a group where the FMF&AID Association was mentioned. From the first moment I contacted the association, my son and I were warmly welcomed. The association's volunteer took all the time necessary to give me information about these diseases, why they occur, and the consequences if left untreated.

Unfortunately, in Chile, autoinflammatory diseases are not covered by the social healthcare system, therefore, parents have to bear all the costs themselves. Luckily, there is the FMF&AID, an association that offers its help to those who need it. Thanks to the financial support I received from this association, I was able to buy my son the medications that were prescribed to him by his doctor and that were not covered; even the special food my son needed due to being underweight. However, since my son's condition did not improve, the treating physician prescribed a biological medication which is not covered in Chile either.

When the FMF&AID found out about it, and after receiving a copy of the prescription, they immediately organized the purchase of the high-cost biological medication and had it delivered to the hospital.

Unfortunately, due to medical issues, nowadays my son cannot continue with the treatment and false accusations have been made against me by one of the treating physicians. As a result, my son and I were referred to a psychiatrist. Once again, I contacted the FMF&AID Association for help and this time it was suggested to make an appointment with a specialist abroad, so that my son could be seen objectively and by doctors who have seen many more cases and consequently have much more experience.

cont.

However, due to the accusations that have been made against me, the long-awaited appointment had to be cancelled for the time being, since I am not allowed to leave the country with my son.

Despite the sufficient evidence (photos, recordings, medical reports) I have collected of my son's recurrent symptoms (rash all over his body, ulcers in the mouth, abdominal pain, absence seizures, eye inflammation, pain and joint swelling, etc) over the years, today the autoinflammatory disease has been ruled out. In addition, I am being accused of fabricating my son's disease and making him sick so that he can be treated. Luckily, we have a lot of evidence in our favor, as well as testimonies from many other patients with stories similar to mine, stories which my lawyer will use to defend us in Court.

I am grateful for all the support that the FMF&AID has given us over the years and continues to give us, as well as for not giving up on us. We are deeply indebted, so to speak, not only for the help we have received, but also for the unconditional care and support. It is comforting to know that my son and I are not alone, that someone takes us seriously, and that we can count on their support.

FMF in Egypt

I am a 25-year-old Egyptian woman who suffers from Familial Mediterranean Fever (FMF), an autoinflammatory disease, which was passed down to me by my mom. It is well known that, if FMF is left untreated, on the long run it can affect the organs. I live my life knowing that I will get a flare on a monthly or quarterly basis despite treatment. These flares or attacks happen completely unannounced, and when they are over, luckily, there is no damage to my body.

My mom used to tell me that if I take my colchicine every night, I'm going to be okay. Unfortunately, despite having many people in my country suffering from this disease, not many people know the impact it has on daily life. After suffering a lot from FMF physically, mentally and emotionally, I decided to do a search of my own and find someone that can help me. That's when I found the Facebook groups of the FMF & AID Global Association and joined them. I was surprised to learn that the disease is way more than a flare or an attack and that it has so many dimensions to it. The group was an amazing help for me. I immediately reached out to Malena Vetterli on Messenger, and she offered to meet with me virtually in a one-to-one call. I talked to her about everything, and her support, guidance, and knowledge were amazing and truly appreciated. Now that I understand my disease, I can take better care of myself thanks to the Facebook group.

SEPTEMBER 2022
World Autoinflammatory Awareness Month

1. There are **+40 autoinflammatory diseases**
   Some of these are: PFAPA, FMF, CAPS, FCAS, MWS, NOMID, TRAPS, HIDS, DIRA, Behcet's disease, Yao Syndrome, AOSD, sJIA, Schnitzler's Syndrome, PAPA, etc.
2. **14 years -** until adult diagnosis
   3 years - until pediatric diagnosis
3. Individuals of **all ethnicities** and ages can be affected.
4. **Common symptoms**: fevers, rashes, joint and muscle pain/swelling, abdominal pain, malaise, chills, headache, fatigue, enlarged lymph nodes, red eyes, mouth ulcers, diarrhea, constipation, vomiting, etc.

FMF in France

We are a Sephardic Jewish family with four children and three of them have been diagnosed with Familial Mediterranean Fever (FMF). According to the physicians, our disease presentation is totally atypical, probably because according to the medical literature, only one child out of the four should be affected, but in our case, three of the four kids have FMF. They even have a severe presentation of the disease, despite having negative genetics. It took 3 years for my children to get properly diagnosed, but before we got that far, we had to deal with derogatory and demeaning remarks from healthcare professionals. My children experienced frequent visits to the pediatrician, many visits to the emergency department, many hospitalizations until the day we finally came across a doctor who knew this disease. Our eldest son had his first fever episode when he was only 3 weeks old, and these episodes kept recurring every 3 weeks. His flares were so predictable, that we knew in advance when these would happen.

After years of suffering, lack of understanding, multiple visits to physicians (i.e. orthopedist, radiologist, physiotherapist, pediatric psychologist, pain clinic), we finally had a diagnosis and treatment. It was by chance that we found on the internet about the CEREMAIA pediatric reference center in Paris, where our children are treated and still go for their follow-up visits every year.

The team is exceptional! When our son was 14 years old, he ended up in the emergency department with only partial active kidney function, pleural effusion, 10 kg weight loss, a very bad situation. We couldn't understand how in the course of 2 years, our son had FMF and all of a sudden, it was so severe that he had to be hospitalized for one week. However, while our son was recovering, our 2 other kids were sick too. It became clear to us that our fight is far from over. Adolescence is a very difficult period for this disease, especially with the crazy hormones.

During the hospitalization of our son, we were lucky enough to meet yet another great doctor, the head of the rare kidney disease department. He gave us information that nobody else had in the past 10 years we had been there. We are glad to see that everything changes, and things improve. We finally have the impression of moving forward.

cont.

Actually, we have a young man who no longer wants to hear about FMF or take any medications, a pre-teen, who is fed up with her joint pain that limits her daily (writing, walking, all aspects of her life), and a child who flares periodically without typical symptoms but with many canker sores.

Our daughter has been on a biological medication for 1 year, after many discussions with the nephrologist. Great school accommodations have been implemented thanks to the hospital teacher; but frankly with no improvement. We requested a dosage increase, but the doctor thinks that she may have yet another disease(s) in addition to FMF, so she will have to be hospitalized for additional examinations to be done. We might be wrong, but we think it's just the same disease but uncontrolled.

As for our youngest child, despite being on colchicine, he is still flaring. There is no question in our minds that he needs to be switched to a biological medication. However, who are we to say anything? We are simply the parents of 3 sick children with a totally atypical presentation. As parents, we are glad to have a team of amazing doctors, who examine them from every angle, who listen to them, who talk to them, who understand what we are experiencing. Despite this, they are not in our lives, facing our daily problems and dealing with our sick children. Every week one child is sick, as if they would take turns.

Due to the school absences, some teachers feel they have the right to judge them, to imply that they are faking it, and pretending to be tired and exhausted.

The only real support we have is from the FMF & AID Global Association, which has been great organizing and covering the costs of an additional genetic test. Unfortunately, despite the large number of mutations tested, it did not reveal any findings. Today, after 21 years of illness, our children are diagnosed with FMF (without any genetic evidence) but are still sick. All this is very complicated. We are tired, but despite everything, we thank God for our children.

uSAID in Peru

I am the mother of a 3-year-old boy with an undefined autoinflammatory disease. My son is my adoration, the most awaited because I was able to have him only after 8 years of trying. I also present with some medical issues such as endometriosis, fibromyalgia and an autoimmune disease, which is yet to be diagnosed. After seeing my son's case, I realised that I may also suffer from an autoinflammatory disease.

My son started to have fevers when he was 6 months old, and we went through various viral and infectious misdiagnoses. At first, my son suffered from diarrhea, blood in the stool, increasingly recurrent fevers every 3 to 4 months, mouth ulcers, etc., but later on, he was diagnosed with blood disorders such as anemia and neutropenia.

After many tests, my son was hospitalized thinking he had leukemia or lymphoma, due to his enlarged spleen and liver. In addition, he had many adenopathies (swollen lymph nodes). Several genetic and bone marrow studies were carried out, in which several inflammatory anomalies were found, but nothing significant that would serve to diagnose him.

Then one day, he had a severe intestinal bleeding and had to be hospitalized. For the first time he was seen by an immunologist, who said that my son possibly suffered from an autoinflammatory disease.

This was the first time we heard about these diseases.

I started looking for information on the internet, and that's how I found the FMF&AID association. We contacted them and we were welcomed by one of their volunteers, who immediately offered the help of the association. They helped us at no cost, and on top of that, they covered the expenses of two genetic tests (complete panels). Unfortunately, the results did not yield any information, but neither the volunteer nor us as parents gave up. In Peru, doctors told us that they couldn't do more for my little boy because it was the first case in Peru, so we were only given the option of palliatives but no treatment.

This is when the FMF&AID offered to put us in contact with an overseas rheumatologist expert in autoinflammatory diseases and they also covered the cost of the consultation. This foreign doctor suggested we try prednisone at the onset of the fevers, and it worked like a miracle.

cont.

He also recommended us to contact a local immunologist and gave us the contact details. In case the local immunologist couldn't help us and if all other options had been exhausted in Peru, the association would help us take our son to specialists in Argentina.

Thanks to the FMF&AID, we will be able to get my son treated abroad so that his case can be further investigated, especially since his symptoms worsen day to day. So far, he has only been diagnosed with Raynaud's syndrome. Other symptoms that my son presents are facial redness, bruises, inflammation of the joints, possible vasculitis, and Lupus.

At the moment, we are waiting to see if we have to take our son abroad. FMF&AID is committed to continuing to support us with the treatment and whatever else is necessary. We are very grateful as parents to FMF&AID for always responding so quickly every time we contact them and for helping us without expecting anything in return.

May God watch over and protect all of us who go through these diseases that limit the quality of life, not only for the patient, but also for the entire family. As long as we have strength, we will continue to fight for our son!!!!

KIDS CORNER

BLOOMING BUDDIES NEWSLETTER

MARINE LIFE SENSORY BIN

MATERIALS:
. PLASTIC CONTAINER
. 2 CUPS OF WATER MIXED WITH 1 DROP OF BLUE FOOD COLORING
. SHAVING CREAM
. SMALL MARINE ANIMALS
. SMALL BOATS
. SYRINGE/ DROPPERS

INCORPORATING SENSORY PLAY AT HOME

BY ASHLEY VENTRICE

Sensory play has so many benefits and one of them is it's calming ability. When having a child who is home sick sensory play can help by providing a calm activity for your child. Sensory play is also known to boost problem solving and creativity. It builds nerve connections in the brain's pathways, which helps a child cope and complete more challenging tasks. Sensory play also fosters language development , cognition, social interaction, fine motor skills, and gross motor skills.

What is sensory play?

Sensory play is any play activity that stimulates a child's senses through touch, taste, smell, sight, and hearing.

What is a sensory bin?

A sensory bin is a container filled with materials and objects that stimulate a child's senses. The picture above shows a sensory bin filled with shredded colorful paper and wooden marine animals.

How to make a marine life sensory bin

This sensory bin is great to make with your child and is suggested for children ages three and up. Find a sturdy plastic container. Add two cups of water to the container and one drop of blue food coloring. Then mix the water and food coloring. Children love to mix the water and see the color change! Next, spray a few blobs of shaving cream in different parts of the water (do not mix in). Add your favorite marine creatures, spoons, cups, and droppers. Let your child explore and enjoy!

How to make your own sensory bin

You can make your own sensory bin by adding any of the below fillers to a bin. Then you can add toys that interest your child. Last, you can add fine motor tools like spoons, tongs, and cups.

SENSORY FILLER IDEAS

. Sand
. Water
. Soapy water
. Ice
. Snow
. Cooked noodles (cooled)
. Uncooked pasta
. Dry oats
. Dry beans
. Cereal
. Rocks
. Sticks

. Bird seed
. Dry rice
. Corn kernels
. Cut up paper
. Shaving cream
. Cut up straws
. Pom poms
. Beads
. Cut up ribbon
. Shredded paper
. Tissue paper
. Legos
. Cap tops
. Water beads

"Play gives children a chance to practice what they are learning."

MR. ROGERS

Tag us with your sensory bins!

@bloomingbuddiesco

Authors

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The patient stories included in this newsletter, have been provided to the FMF & AID Global Association voluntarily and 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 are royalty free photos from Unsplash. Proper credit has been given to the owners of the pictures underneath each photograph.

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

List of disease abbreviations used in this Newsletter

FMF	Familial Mediterranean Fever
uSAID	Undifferentiated autoinflammatory disease