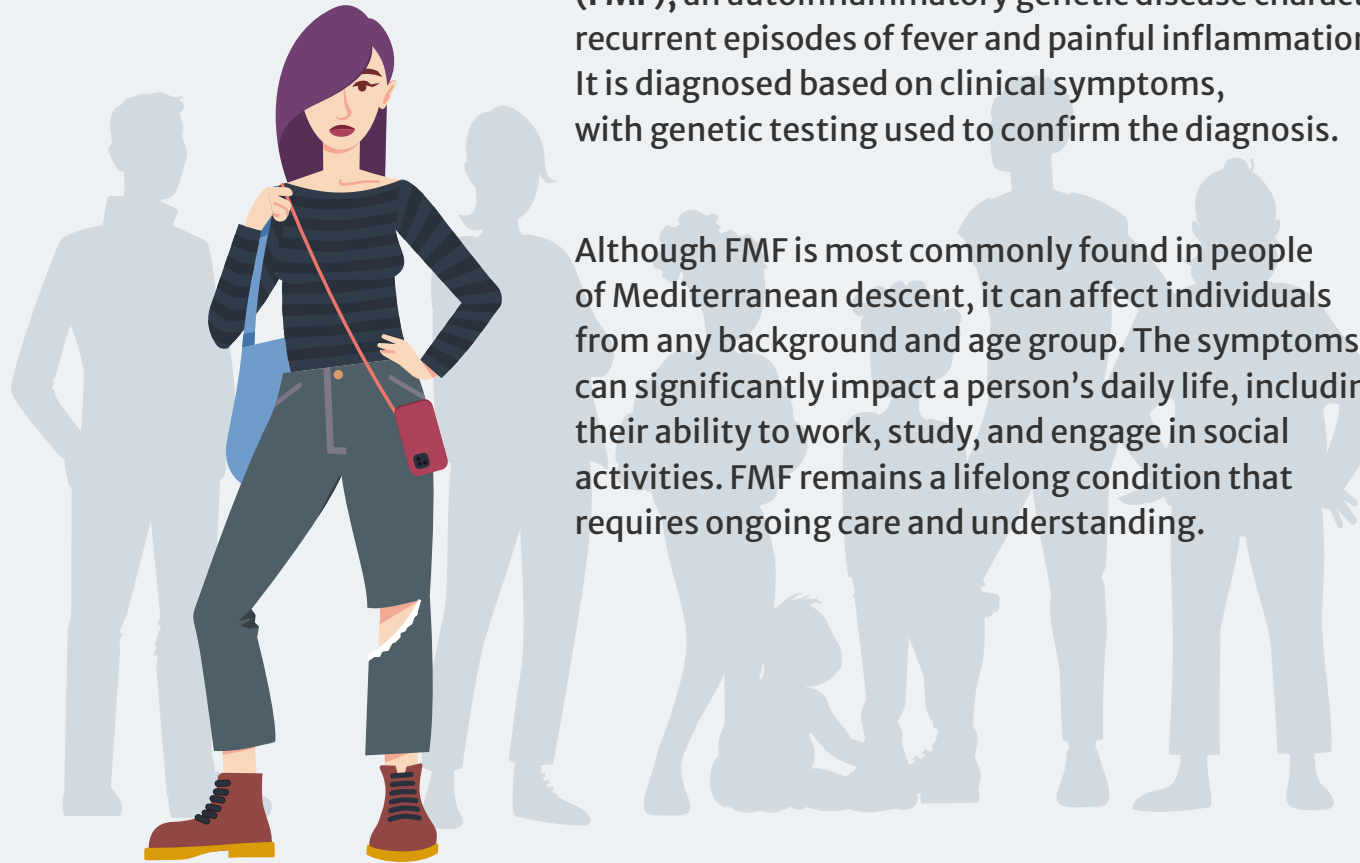


The journey of a patient

Familial Mediterranean Fever

Meet Amira, she is a patient with **Familial Mediterranean Fever (FMF)**, an autoinflammatory genetic disease characterized by recurrent episodes of fever and painful inflammation. It is diagnosed based on clinical symptoms, with genetic testing used to confirm the diagnosis.

Although FMF is most commonly found in people of Mediterranean descent, it can affect individuals from any background and age group. The symptoms can significantly impact a person's daily life, including their ability to work, study, and engage in social activities. FMF remains a lifelong condition that requires ongoing care and understanding.



Symptoms



Diagnostic



Treatment



Follow-up
& aging



Resources



Symptoms description



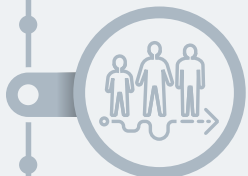
Symptoms



DIAGNOSTIC



TREATMENT



FOLLOW-UP & AGING

GENERAL PRACTITIONER

Low-grade temperatures
High fevers (not always!)
Swollen jaw/jaw pain
Fatigue
Night sweats
Hot/cold body parts
Insomnia
Weight issues
Nose bleeds
Occasional canker sores

CARDIOLOGY

High blood pressure
Tachycardia
Pericarditis

HEMATOLOGY

Anemia

GASTRO-ENTEROLOGY

Chronic abdominal pain
Bloating
Severe abdominal pain
Intestinal blockages
Diarrhea
Nausea/vomiting
Liver and spleen swelling

NEPHROLOGY

Kidney pain
Blood in the urine
Frequent urination
Painful urination

UROLOGY

Fertility issues
Scrotal swelling

ORTHOPEDIC

Carpel tunnel (wrists and hands)
Plantar fasciitis

OPHTHALMOLOGY

Eye issues
Dry eye
Episcleritis

NEUROLOGY

Headaches
Muscle weakness (tingling, numbness)
Light and sound sensitive
Seizures
Mood swings
Anxiety
Depression

PULMONOLOGY

Respiratory issues
Shortness of breath
Dry cough
Asthma
Pleuritis (chest pain)

DERMATOLOGY

Rashes
Bruising
Skin problems/eczema
Erysipela-like
Henoch-Schönlein purpura

IMMUNOLOGY

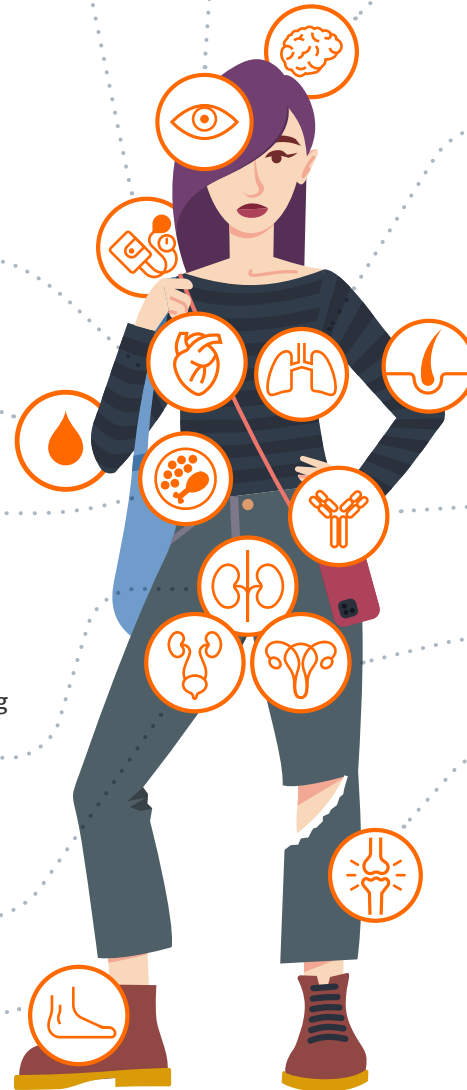
Drug intolerances
Food intolerances
Infections

GYNECOLOGY

Painful periods
Fertility issues

RHEUMATOLOGY

Joint pain/swelling
Lower back pain
Shoulder pain
Leg pain/blocked hip
Fluid in the organs or joints (oedema)
Chronic pain
Tendinitis
Synovitis
Swollen lymph nodes
Muscle pain



REQUIRED PRACTITIONERS

General practitioner

Pediatrician

A&E/Emergency

Symptoms Challenges



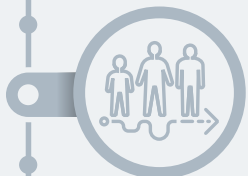
Symptoms



DIAGNOSTIC



TREATMENT



FOLLOW-UP & AGING



ACCESS TO MEDICAL CARE

Finding knowledgeable doctors who recognise autoinflammatory diseases

Doctors don't recognise the pattern of disease symptoms easily

Lack of collaboration between specialists and doctor/clinics



ONSET AND SYMPTOMS

Onset ranges from baby to elder

Symptoms vary and change overtime

Overlaps with other disease categories



PAIN

The pain is severely underestimated

Lack of access to pain meds

Ability to access appropriate testing and treatment



SOCIAL-FINANCIAL BURDEN

Doctor shopping to find care

Travel expenses to see specialist

Unable to remain employed due to debilitating symptoms

Difficulty getting disability



DIAGNOSIS ODYSSEY

Risk of permanent organ damage with elevated SAA

Symptoms are treated instead of the disease



MENTAL HEALTH

Not taken seriously with life-impacting symptoms

Blamed as being psychosomatic

Labeled as a drug-seeker

Anxiety

Depression

Fatigue

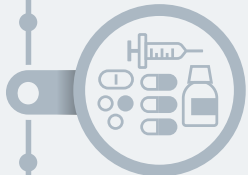
Symptoms Needs



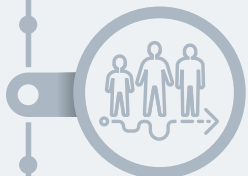
Symptoms



DIAGNOSTIC



TREATMENT



FOLLOW-UP
& AGING



GENETIC TESTING

Easier access to genetic testing

Full access to genetic results incl. benign variants report



OUTREACH & ADVOCACY

More research with patient involvement (i.e. surveys, medication efficacy, inflammatory markers, pain control)

Patient disease needs acknowledgement by medical teams, schools, family, friends, work colleagues, etc.



DOCTOR TRAINING & COLLABORATION

AID training needs to be provided to clinicians

General practitioners should be willing to work in close collaboration with a variety of specialists required for comprehensive care

Increased awareness by the medical communities

Referral of patients to specialists must be done in a timely manner

Telehealth consultations should be offered



EDUCATION/WORK

Homeschooling for kids who can't attend school

Remote working accommodations for adults

Diagnostic description



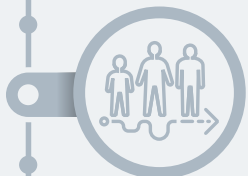
SYMPTOMS



Diagnostic



TREATMENT



FOLLOW-UP & AGING

BLOOD

Elevated CRP, ESR, SAA (not always!)
 Elevated liver enzymes
 Low ferritin
 Vitamin D deficiency
 Vitamin B12 deficiency
 Immunoglobulins too low or too high
 Elevated cytokines
 High cholesterol
 Proteinuria
 High white & red blood cells

OTHER TESTS

Endoscopy
 Colonoscopy

SCANS/ULTRASOUND

Sinovitis/fluid build-up
 Joint inflammation
 Organ inflammation

COMORBIDITIES

Infections & immunodeficiency
 Epilepsy/seizures
 Sicca syndrome
 Hashimoto's
 Behcet's
 Psoriasis
 IBD
 Hypermobility/EDS
 Restless legs syndrome
 Sleep apnea
 Raynaud's disease
 Von Willebrand disease
 Factor V Leiden
 Factor VII deficiency
 Factor X deficiency
 Sinusitis/post-nasal drip
 Postural orthostatic tachycardia syndrome (POTS)
 UTI (urinary infections)
 Genital yeast infection
 ADHD
 Idiopathic intracranial hypertension
 Fibromyalgia
 Psoriatic arthritis
 Ankylosing spondylitis
 Splenomegaly
 Hepatomegaly
 Non-alcoholic fatty liver



REQUIRED PRACTITIONERS

Multiple specialties:
 Rheumatologist
 Infectious disease
 Immunologist
 General practitioner
 Cardiologist
 Pulmonologist
 Dermatologist
 Gastroenterologist
 Neurologist
 Haematologist
 ENT
 Physiotherapist
 Endocrinologist
 Ophthalmologist
 Nephrologist
 Gynecologist
 Urologist

Diagnostic Challenges



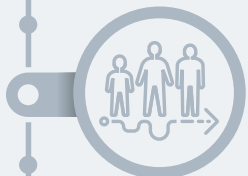
SYMPTOMS



Diagnostic



TREATMENT



FOLLOW-UP & AGING



DIAGNOSIS ISSUES

Diagnostic timeframe ranges from 3 years for children to 14+ years for adults

.....

Diagnosis through to incidental finding

.....

The odyssey begins after diagnosis

.....

Colchicine trial may be helpful to confirm the diagnosis

.....

Genetic testing can be key during the diagnostic process



MISCONCEPTIONS

Only people of certain ethnic origins can have FMF (blond and blue-eyed people can have FMF too).

.....

Two mutations are needed for an FMF diagnosis (single carriers face diagnostic delays).

.....

Elevated inflammatory markers are needed for a diagnosis (not all patients have elevated markers).

.....

High fevers must present in all FMF patients (fever is not common in adults).



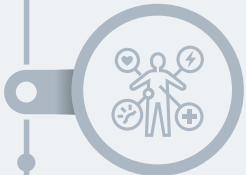
CLINICAL CHALLENGES

Bloodwork may not capture inflammation.

.....

Adult patients often outgrow their fevers.

Diagnostic Challenges



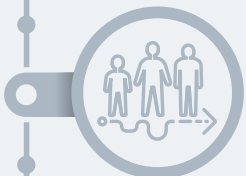
SYMPTOMS



Diagnostic



TREATMENT



FOLLOW-UP & AGING



MULTIPLE DOCTORS & MISDIAGNOSES

On average, a patient seeks help from 7 to 10 doctors

Multiple misdiagnoses: mental illness, appendicitis, asthma, osteoarthritis, fibromyalgia, chronic intestinal inflammation/ IBS, chronic fatigue, chronic pain syndrome, drug seeker/ addict, COPD, colon cancer, anorexia, assumed stomach ulcers, accused of ingesting poison, diabetes, celiac disease

Patient remains unsupported

Difficult identifying comorbidities



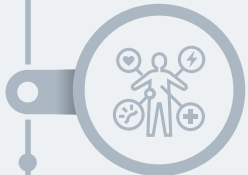
GASLIGHTING

Patients not taken seriously

Often sent to psychiatric counselling

Hypochondriac diagnosis

Diagnostic description



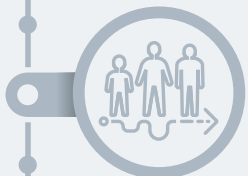
SYMPTOMS



Diagnostic



TREATMENT



FOLLOW-UP & AGING



ACCESS TO MEDICAL CARE AND SUPPORT

To have extensive medical care

To have medical explanation of FMF

To have access to labs during a flare

To have a medical explanation of lab results

To have access to telehealth during a flare

To have an appropriate FMF trained clinician



ACCESS TO MEDICAL TREATMENT

To have limited access to biological medications due to prohibited cost or availability within a country

To trial multiple colchicine brands



COMMUNITY & INFORMATION

To have access to medical information on FMF

To have access to a supportive community of like patients

Treatment description



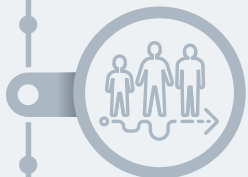
SYMPTOMS



DIAGNOSTIC



Treatment



FOLLOW-UP & AGING

DISEASE MEDICATIONS

Colchicine
(try several brands)
Kineret
Ilaris
Actemra
Cortisone
Riloncept
(US only)



PAIN MEDICATIONS

NSAIDs:
Ibuprofen
Mefenamic acid
Novalgin

Opioids:
Tramadol
Palexia
Oxycodone
Morphine



REQUIRED PRACTITIONERS

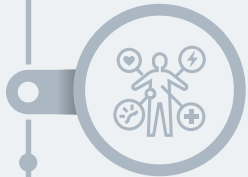
Rheumatologist

Immunologist

General practitioner

Socio-therapist
(offers advocacy support to patients)

Treatment Challenges



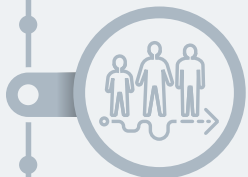
SYMPTOMS



DIAGNOSTIC



Treatment



FOLLOW-UP
& AGING



LIFE ISSUES

Lack of understanding by employers and authorities of the disease

Despite treatment, patients are often unable to lead a normal life

Patients living on a limited budget are often required to travel long distance to see a specialist; these costs are not covered



MEDICATION ISSUES

Lack of pain management

Doctors often under-medicate patients especially when in pain

Difficulty accessing biological medications



DISEASE PROTOCOL & GUIDELINES

Lack of medical documentation to support the flare-cycle severity of the disease

Incorrect treatment protocol or not enough medication based on lacking medical research

No medical team collaboration

Treatment Needs



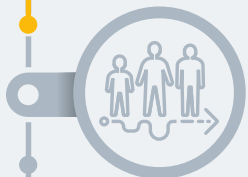
SYMPTOMS



DIAGNOSTIC



Treatment



FOLLOW-UP
& AGING



MEDICAL EDUCATION

Requires specialists with the necessary knowledge and experience

To have a doctor who listens



TREATMENT

Pain medications are often not strong enough to manage the patient's flare

Need additional and effective treatments developed for FMF

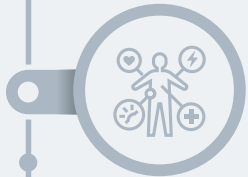


DISEASE BURDEN

Financial burden of over-the-counter medications not covered by insurance

Triggers need to be identified and reduced (avoid physical exertion and stress)

Follow up & Ageing description



SYMPTOMS



DIAGNOSTIC

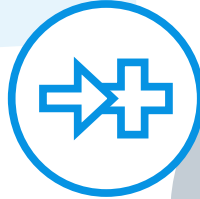


TREATMENT



Follow-up & Aging

EVERY 3-6 MONTHS FOLLOW-UP



DISEASE MAY MANIFEST DIFFERENT OVER TIME



TRANSITIONING FROM PEDIATRICS TO ADULT DOCTORS



FOR SEVERE FLARES, A&E



REQUIRED PRACTITIONERS

Rheumatologists

Immunologists

General practitioners

A&E

Follow up & Ageing Challenges



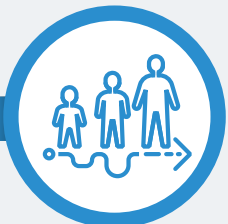
SYMPTOMS



DIAGNOSTIC



TREATMENT



Follow-up
& Aging



**DISEASE
PRESENTATION
VARIABILITY**

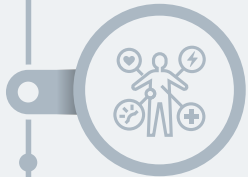
Proving that FMF is variable
and can become worse with
aging



PATIENT BURDEN

Often long-distance travel
for follow-up of medical
appointments

Follow up & Ageing Needs



SYMPTOMS



DIAGNOSTIC



TREATMENT



Follow-up & Aging



LACK OF SPECIALISTS

Not enough rheumatologists or immunologists treating FMF

Finding a doctor with the necessary knowledge and close to where the patient lives



FLARE MANAGEMENT

To have an emergency plan

Updated emergency guidelines for FMF



OTHERS

More awareness

More knowledgeable doctors

More research

Better collaboration

More understanding

Resources

Awareness, advocacy, patient support

Patient organisations

Self-support groups

Research resources

PubMed

Scientific journals

Webinars