Mystery Patient Guide

A Peer-Led Guide to Navigating the Journey to Diagnosis

AiARTHRITIS
INTERNATIONAL FOUNDATION FOR AUTOIMMUNE & AUTOINFLAMMATORY ARTHRITIS

"I think I might have an AiArthritis disease, but no one believes me... what do I do?"

This guide is compiled of information from fellow patients' journeys that offer insight into different situations along the diagnosis journey. With this resource, we hope to equip undiagnosed patients with key information and support, thereby empowering them to advocate for answers.

Note: As AiArthritis disease symptoms overlap with other symptoms, you may not have one of our diseases. This guide is not a guarantee of diagnosis for any condition.

WHY THIS MATTERS

- 6 months is the ideal window of opportunity for diagnosis & treatment
- Prevent comorbidities & lead to more positive outcomes like remission
- Early intervention = higher chance for remission & lower healthcare costs
- We learn from each other when we share our experiences

HOW TO USE THIS GUIDE

Equip yourself with the knowledge and resources necessary to navigate health appointments with confidence. Gain insight into common AiArthritis symptoms, tests, and complications to manage expectations and prepare for every step of your diagnosis journey.

1. Spot potential symptoms & prepare to seek help
2. Seek help from a medical professional
3. Learn how to self-advocate

Understanding AiArthritis Diseases

Autoimmune Diseases

100+ diseases caused by adaptive immune system reactions and result from a combination of genetic and environmental influences

Autoinflammatory Diseases

About 2 dozen diseases caused by Innate Immune System (inborn) reactions and may result from genetic mutations

There are over 100 AutoImmune and AutoInflammatory diseases, but only a couple dozen include Inflammatory Arthritis as a major critical component. Those are the diseases we represent.

"**AutoImmune** Arthritis" and "**AutoInflammatory** Arthritis"
or
"**AiArthritis**"

AiArthritis Diseases

- Rheumatoid Arthritis (RA)
- Psoriatic Arthritis (PsA)
- Systemic Lupus Erythematosus (SLE)
- Sjögren's Disease (SD)
- Axial Spondyloarthritis (axSpA)
- Juvenile Idiopathic Arthritis (JIA)
- Mixed Connective Tissue Disease (MCTD)
- CAPS (Cryopyrin-Associated Periodic Syndromes)
- Undifferentiated Disease (UD)
- VEXAS Syndrome
- Scleroderma/Systemic Sclerosis
- Still's Disease (AOSD & sJIA)
- Crohn's Disease
- Sarcoidosis
- Relapsing Polychondritis (RP)
- Behçet's Disease (BD)
- Palindromic Rheumatism (PR)
- Schnitzler Syndrome
- CNO/CRMO (Chronic Nonbacterial Osteomyelitis/Chronic Recurrent Multifocal Osteomyelitis)
- FMF (Familial Mediterranean Fever)
- HIDS (Hyper-IgD Syndrome)

Recognizing Symptoms

Every diagnosis journey starts with the onset of symptoms that are different from your norm.
The following are the most common symptoms associated with AiArthritis diseases, but just because you have these does not necessarily mean you have one of our diseases.
Tip: Use a journal or log to track your symptoms and identify any patterns.

Inflammatory Arthritis

Pain in one or more joints that might be paired with swelling or redness.
This can be in common areas like knees, hips, or fingers, but also in locations you may not think of, like the neck, spine/buttocks, or even the jaw.

"Jumping" or "comes and goes" pain

Pay particular attention to pain that **comes and goes for no apparent reason or jumps locations.**

Difficulty moving after periods of rest*

Note if you experience stiffness that **worsens after rest or inactivity.**

Chronic inflammation

Even if it comes and goes, notice symptoms that occur for **6 weeks or longer**.

**Many report that their bed or pillow is suddenly uncomfortable, but it's actually pain from stiffness.*

Classic "Auto" Symptoms

These are common in all AiArthritis diseases regardless of diagnosis. Debilitating fatigue is reported in over 90% of diseases and the others are reported in over 30%.*

Fatigue that is severe, unusual, and not corrected with rest or caffeine

"Brain fog" that makes it difficult to think or concentrate

Flu-like symptoms like muscle weakness, malaise, and even nausea

Fever: low grade in autoimmune; higher grade & more common in autoinflammatory diseases

Potential Organ Involvement

While major organ involvement comes later in some cases, in others it can be a first symptom. Common organ involvement includes skin rashes & eye pain. Other organs involved may be kidneys, lungs, heart, spleen, or intestines. It's not uncommon for organ symptoms to lead you to a doctor or specialist that may not think of an AiArthritis disease as a possibility.

**Reference: Early Symptoms of AiArthritis Study (2013)*

What to Do If You Experience Symptoms

“I thought everybody was tired all the time. I thought everybody had random fevers that popped up out of nowhere like I didn't know that was unusual.“

1

Start a log or journal

Track details such as: symptom onset, length, frequency, time of day and anything that made symptoms worse or better

2

Be specific!

More detailed tracking will identify patterns that will help health practitioners get closer to a diagnosis

3

Take photos or videos

Document any swelling, redness, rashes, etc. Date them and notice if multiple symptoms happen at the same time

4

Other Things to Consider:

*Medical History
*Family History
*Any other factors -- be prepared, they'll ask!

**Average onset for our diseases is 20 - 40 years old in adults and any age in children, even at birth*

Seeking Help from a Medical Professional

Depending on your symptoms, you may first see a general practitioner, ER doctor, or specialist

Due to the complexity of these diseases, it's not uncommon to see multiple doctors before getting a diagnosis

Some may not even realize that "arthritis" isn't just osteoarthritis*

ASSESSMENT

Share a summary of your preparation work

Stick to your symptoms -- don’t quote Dr. Google

Medical review and physical exam

**Osteoarthritis often happens with age -- the average onset for our diseases is 20 - 40 years old in adults and any age in children, even at birth*

Diagnosis Journey: Tests

Blood Work

Blood work is common on the diagnosis journey. However, many AiArthritis diseases don’t need positive blood work for diagnosis (i.e. rheumatoid arthritis). It certainly can help identify the disease, including how aggressive it is, but in some AiArthritis diseases blood work is not necessary for a diagnosis.

Potential Tests

Testing may include blood work, imaging, and family history examination. Common tests for systemic inflammation may include: Erythrocyte Sedimentation Rate (ESR), C-Reactive Protein (CRP), and Antinuclear Antibody (ANA). Some of our diseases are associated with various biomarkers that, if positive, can help pinpoint the disease.

Imaging Tests

X-rays or MRIs may also be ordered to determine if there are any visible changes to joints or to locate inflammation.

**X-rays typically show damage after 10 years of disease onset or in aggressive cases. However, insurance may require them before ordering an MRI or other expensive imaging. If nothing appears on the x-rays, ask your doctor about next steps.*

Question Guide:

- What do my symptoms indicate?
- What conditions are you considering?
- Do we need to order additional tests?

**Getting a doctor to listen to you can be a huge hurdle. If tests had been ordered immediately, they would have found it right away because it wasn’t hiding.*

Diagnosis Journey:

Testing Complications

“You must be depressed or I got well, you obviously hurt yourself at the gym because I was an athlete and I kickboxed and I played sports and even they didn't believe my inflammation for a couple of reasons....

...All my blood work was ‘excellent’. I had early disease, which means if someone would have identified it and treated it I would likely have gone into remission, maybe permanently, but now I missed that window of opportunity.”

Sjogren’s Case Study

“I was tested for Sjogren's. I didn't have the biomarkers, so I literally was dismissed, it cannot be Sjogren's.”

We know now only 60 percent of people living with Sjogren's have the biomarkers.

Currently, the diagnostic criteria for Sjogren’s involves these biomarkers or a lip biopsy, which is super invasive. New biomarkers are emerging for Sjogren’s, so we predict the diagnosis criteria will be updated soon.

Diagnosis Journey:

Expectation Setting

It can take months to get in to see specialists and getting connected to a specialist who can ultimately identify and diagnose an AiArthritis disease can be an additional challenge (i.e. long wait times due to shortages or living in areas that do not have these types of specialists).

➜ Keep an Open Mind

The search for answers can be frustrating. Although we are experts of our own body, the doctors are experts in putting together the puzzle of a diagnosis.

➜ Symptom Identification Can Be Tricky

It's especially hard for patients who don't have positive blood test results, no visible damages on an X-ray or MRI, or no skin warmth and/or swelling to the touch. We all start somewhere as we start feeling the changes to our bodies - it just may take time for measurable activity to show up.

➜ Delays to Diagnosis

"I want to wait and watch you get worse." Sometimes doctors wait to investigate until symptoms worsen or will order more tests to rule out other possibilities, which prolongs the process.

➜ Patient Gaslighting

"It's probably just (fill in the blank)." i.e. stress, growing pains, an old injury, or because of your weight, lack of sleep, depression, etc.

WHAT PATIENTS WANT YOU TO REMEMBER

- Negative blood work doesn't mean you're seronegative, you just may have not had positive markers that day
- Request an MRI, if possible
- Undifferentiated Disease IS a diagnosis

Learning Self-Advocacy

“You’re too young to have arthritis.”

“Your fatigue is normal for a busy person.”

“You should try this diet before we explore further.”

“You should try to exercise more.”

“Maybe if you get better sleep, you’ll be less tired.”

Common things patients have been told during their diagnosis journey

“Your x-rays aren’t showing any damage.”

"You must have injured yourself working out."

Maybe trying yoga will help with your pain.”

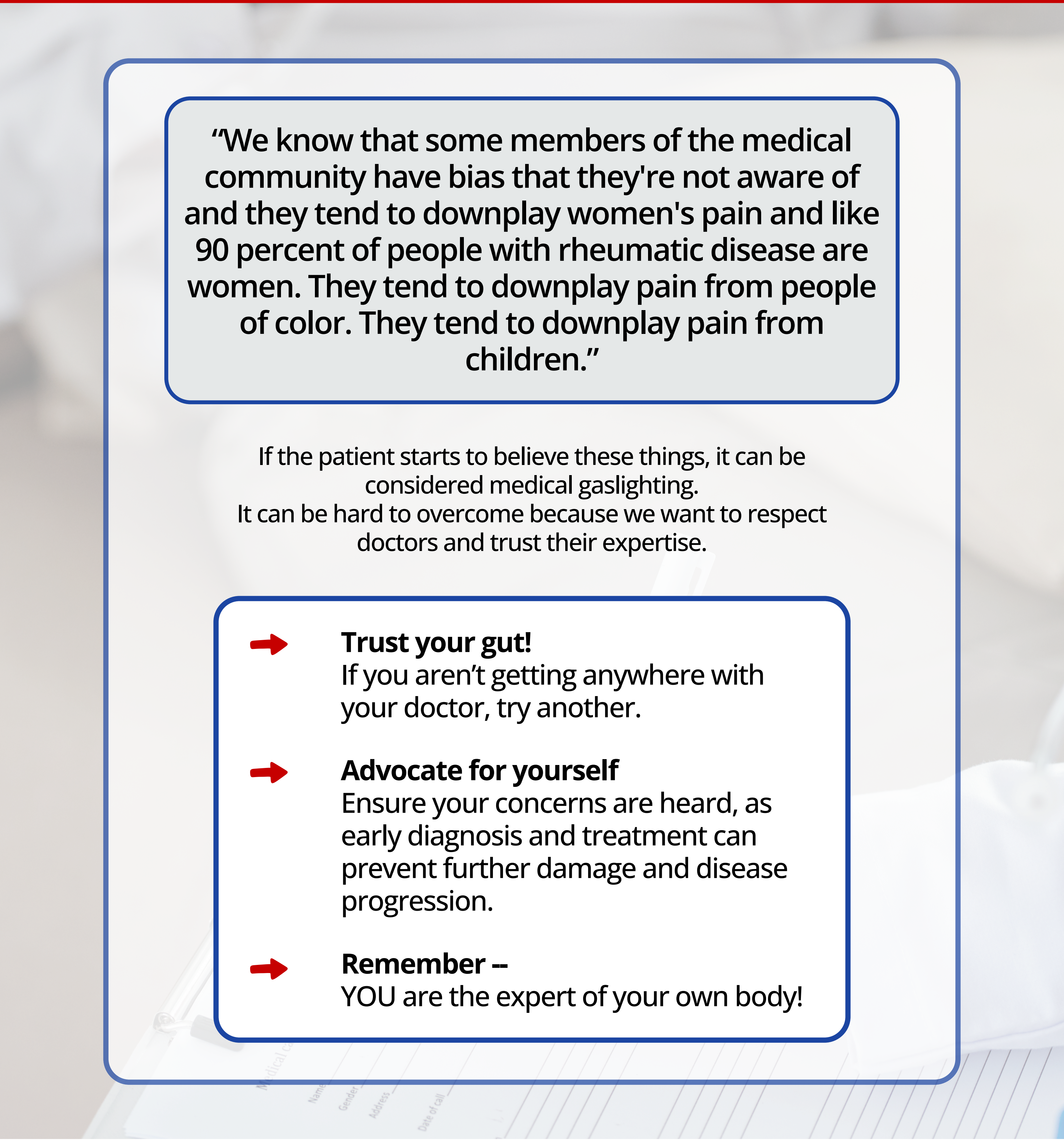
Maybe it’s depression, try seeing a therapist.”

“Your blood work is fine; nothing is wrong with you.”

Feeling dismissed and struggling to get diagnosed?

Doctors may recommend weight loss or special diets to see if symptoms improve. If you've already tried this or don't believe it will help, consider finding another doctor who listens to your concerns.

Unintentional Biases



> “We know that some members of the medical community have bias that they're not aware of and they tend to downplay women's pain and like 90 percent of people with rheumatic disease are women. They tend to downplay pain from people of color. They tend to downplay pain from children.”

If the patient starts to believe these things, it can be considered medical gaslighting.
It can be hard to overcome because we want to respect doctors and trust their expertise.

- **Trust your gut!**
  If you aren’t getting anywhere with your doctor, try another.
- **Advocate for yourself**
  Ensure your concerns are heard, as early diagnosis and treatment can prevent further damage and disease progression.
- **Remember --**
  YOU are the expert of your own body!

Help Us Help Others

Quotes from patients who went through their own diagnosis journey:

"None of us wanna be sick. We just want an answer. We wanna know what is causing this, and what we can do to have a better life."

"Because it's a rare disease, most of my doctors knew nothing about it."

"I was sitting on a chair and I stood up, and that's all it was. And I sent it to my mom and we both cried because we were so happy because there was that much of a difference just from getting the right treatment for the right condition."

"Don't give up on trying to get your diagnosis as early as possible, if I would have had treatment 7 years before I actually did, I might not have to use my cane as much as I do."

Are you currently undiagnosed or a "Mystery Patient" and would like to tell us about your frustrations? Is there something you feel we need to address in this guide? Are you someone with a diagnosis and have advice about what this guide should include?

Please complete our form and YOUR submission could be featured in the final iteration of this resource.

"There are some doctors who will be content to say it's some kind of arthritis and that's, you know, that's all you need to know."

**This is a living document, which means we will continually update it to include additional advice submitted by those diagnosed, and undiagnosed, as well as care partners/parents of juveniles.*

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Continuing Our Mission

This resource aligns with our mission to advance education, advocacy, and research for those impacted by autoimmune and autoinflammatory arthritis (AiArthritis) diseases through peer-led guidance, collaboration, and resources that are driven by patient-identified issues and patient-infused solutions.

We envision the stories of tomorrow to be those of early detection and diagnosis, individualized therapeutic intervention and access to them, improved quality of life, common remission, and a world that understands the depth of our diseases and the challenges associated with them.

Contact Us:

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Your gift helps support our mission and vision!

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Fill out a Volunteer Application today at www.aiarthritis.org/volunteer.