

PATIENT JOURNEY (Sarcoidosis)

- A systemic disease with very multifaceted patient presentations.
- Need of visible/organized/accessible care within existing health systems.
- Need for new drugs for condition including drugs to manage symptoms such as breathlessness and fatigue.

Sufferers experience sarcoidosis as a systemic disease very close to rheumatoid arthritis (RA) and systemic lupus erythematosus (SLE), from the cell pathway it seems to be closer to a Th17.1 mediated disease like multiple sclerosis and Crohn's disease (CD).

Sarcoidosis is a systemic disease, but in 80% of people also the lungs are involved at some stage → is seen as a lung disease. But also 80% of sufferers have sarcoidosis activity outside the lungs and the therapy guiding organ is often NOT the lung.

Sarcoidosis is (opposed to CF and IPF) a very benign disease: 2/3 of patients will resolve without significant organ injuries.

"Watch and wait" will be appropriate for many patients, but will be a disaster for others.

1. First Symptoms

Timeline

Usually a disease for adults only, but there are also some kids with sarcoidosis.

First symptoms usually appear at age 25 to 45. It is not a purely genetic disease like CF coming into action at birth and it is not a disease of older people. It concerns people in the peak of their social and economic activity when they have young families to support.

Clinical Presentation / Symptoms (most frequent ones)

- Fatigue und unexplained exhaustion
- Memory and concentration problems
- Flu-like illness with sweat and sensation irritations
- Dry mucosa (dry mouth, dry eyes, dry cough)
- Reddish and/or elevated skin appearances, nodules and rash
- Swollen lymph nodes, glands, joints and mucosa
- Shortness of breath (dyspnea)
- Heart problems (pain, rhythm, reason for dyspnea)
- Neuropathic malfunctions
- Neuropathic autonomous malfunctions
- Sleep problems, restless muscles
- Chest pain, muscle pain, headache, stiff neck
- Red or painful eyes, cataract, blurry vision, other vision problems
- Kidney stones

Presenting phenotypes in sarcoidosis are very inhomogeneous – no two people are presenting the same – two (never identical) snow flakes are an old icon for sarcoidosis.

Maybe sarcoidosis is just a family of related syndromes showing up with the same type of non-caseating granulomas.

Sarcoidosis is often called to be the chameleon of internal medicine, because it can mimic so many other diseases. The short answer on the question "which symptoms can come from sarcoidosis" is: any. But not every symptom comes from sarcoidosis.

Around 10% of patients present with Lofgren's syndrome – a form of acute onset sarcoidosis with very optimistic outcome. But most patients experience a slow onset of unspecific symptoms which could be any disease. And often it will take a long time to make the doctor (GP or specialist) aware, that there is something unusual (and remit to a specialist).

Patient Needs

- Support for presented symptoms – look mostly like a common disease.

Ideal Outcome / Support

- **Recognize early** that there is something unusual in the patient's illness presentation, **have him seen by a specialist**
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2. Diagnosis

Timeline

Depending on the amount and/or increase rate of symptoms/presentation: from within weeks after problems onset (acute sarcoidosis) to decades - or never during lifetime.

Some already established symptom treatments might be wrong, e.g. if misdiagnosed for COPD, cancer or cardiac abnormalities.

Suspected cancer for long time.

Clinical Presentation / Symptoms

Unexplained function restriction of any organ, which is clinically better measurable than patient reported discomfort, which makes the patient to be seen by many doctors till diagnosis.

Patient Needs

- Because there is no positive clinical proof, the diagnosis of sarcoidosis is a matter of exclusion of any other disease with compatible presentation. In nontrivial cases the diagnosis should be done or be confirmed after guess by a sarcoidosis specialist center.
- Usually in non-acute presentations a biopsy is needed. Have (eg. PET) targeted biopsies from easy to reach locations (avoid probing the heart).
- **Diagnose needs to be quick, timely and well managed:** time from first symptoms to professional guess to confirmed diagnosis.
- Quality information on rare disease testing patient paths to support and accelerate diagnosis for healthcare professionals.
- Clarity about which professional team should diagnose sarcoidosis e.g. GPs, specialists, sarcoidosis specialists etc.

There is no positive test for sarcoidosis – it is always a permanent process of exclusion.

Ideal Outcome / Support

- Quicker diagnosis (full, correct and timely).
 - Use of case history to accelerate diagnosis and increase diagnostic accuracy.
 - Testing to be done by a specialist/specialist center (with a named multidisciplinary team).
 - **Defined pathway** of care from mapping the location of the sarcoidosis **to treatment including psychological and quality of life factors.**
 - Keyworkers to liaise between the different professionals involved (especially during diagnosis and initial treatment)
 - Have a designed guiding doctor also visible for consultations by the patient's primary care doctor.
 - Record QoL for treatment monitoring.
 - Inform about the value to join a patient organization.
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3. Treatment / Checkup

Timeline

Treatment is not always necessary. Some symptom treatments (e.g. pain management) might already be established, even without naming a particular disease.

Clinical Presentation / Symptoms

- Symptoms from established, remitting or progressing disease (strength and location).
- Problems from side effects of treatments
- Comorbidities

Usually doctors don't talk with the patients or talk only little with them and have therefore no chance, to judge how relevant or serious the patient's complains are. Specialists say, that e.g. for heart problems, to ask the patient is more indicative than any function test.

Patient Needs

- Information about treatment need, impacts, side-effects, adverse reactions and choices available – to prevent organ damage and improve/maintain quality of life.
- Discussion of treatment choices and options e.g. anti-inflammatory painkiller v. steroids
- Understanding risks and benefits of any treatment offered.
- Treatments available for symptoms and co-morbidities including fatigue, breathlessness, pain, small fiber neuropathy, weight gain and sleep issues.
- **Continuous diagnostic support is needed because of changing presentation and other upcoming diseases/comorbidities.**
- Information about self-management support e.g. informing about patient organizations/support groups and activities such as physical activity
- Accessible support groups
- Reduce side effects and co-morbidities: Dietetics especially for steroid side-effects, Physiotherapy and rehabilitation
- Psychological support
- How to deal with unsuccessful treatment
- Patients with longer existing disease should be seen at specialist centers – at least sometimes to guide/check the treatment procedure
- Ask/check/care for other coexisting chronic conditions

Ideal Outcome / Support

- Patients have no post-it on them indicating their course -> **ask the patient** – e.g. for heart problems – **and take complains serious.**
 - Treatment guidelines developed with good practice recommendations.
 - Shared decision making between patient and healthcare professional.
 - Access to a range of relevant therapies including psychological support, rehabilitation, nutrition, physiotherapy etc.
 - Patient-friendly information about treatment, side-effects, self-management, support groups (including risks and benefits).
 - Register supporting more research into new drugs and therapies and for quality of care measurement.
 - Support of patient groups/societies from doctors to be more efficient.
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4. Followup

Timeline

- Minimum time: a few years after end of medication.
Maximum time: as long as the patient reports problems that could be associated with sarcoidosis and have no other diagnosis.

Clinical Presentation / Symptoms

- Symptoms from established, remitting or progressing disease (strength and location)
- **Problems from side effects of treatments**
- Comorbidities

Patient Needs

- Support with living with and managing constraints due to sarcoidosis and co-morbidities
- Greater awareness by healthcare professional on potential impact on quality of life e.g. work, social life, relationships, finances, long-term effects, psychological effects, concerns.
- A clear concept of rehabilitation to be included in the care pathway.
- Care pathway to consider multi-organ stage and relapse history.
- **Clarity about which specialist should lead in the monitoring of acute and chronic development, and staging.**
- Guidance for healthcare professionals on follow-up procedures

Ideal Outcome / Support

- Clear pathway for ongoing monitoring of condition including which specialist takes the lead and standard follow-up procedures.
- Direct (and 2-way) communication between specialist and patient's family doctor (including written records) – very important for ongoing condition management.
- Greater awareness by healthcare professionals of impact on quality of life