SARCOIDOSIS - INFORMATION FOR THE PATIENT

**What am I sick with?**

You have just found out that you have sarcoidosis, or your doctor suspects that you have sarcoidosis. Sarcoidosis is a chronic inflammatory disease. This means that clusters of inflammatory cells have formed in your body, forming the so-called sarcoid granulomas, and the substances secreted by these cells maintain inflammation. The lesions most often occur in the lymph nodes located in the chest (lung hili and mediastinum), but other organs may be affected, such as the skin, other lymph nodes, heart, eyes, abdominal organs, bones, salivary glands, gastrointestinal tract, breasts, sexual organs, and other internal organs. Sarcoidosis is a rare disease. It can appear at any age, but it most often affects young adults.

**Why am I sick?**

The cause is unknown. The disease is influenced by genetic factors.

It has not been proven that performing a specific profession, staying in a specific environment or exposure to specific harmful factors caused the disease. Therefore, there is no way you could have prevented the disease by leading a different lifestyle or practicing a different profession.

**What are the symptoms of sarcoidosis?**

Many patients do not report any symptoms. The disease may be detected accidentally.

Many patients report unusual symptoms, such as joint and bone pain, weakness, apathy, increased sweating, low-grade fever, and insomnia.

The most common symptoms caused by changes in the respiratory system are non-productive, paroxysmal cough, shortness of breath on exertion, chest pain, and discomfort, usually unrelated to exercise.

In every fifth patient, sarcoidosis begins with acute symptoms - fever, pain and swelling of the ankle joints, tender or painful red-colored subcutaneous tumors (the so-called erythema nodosum). Lesions most often occur on the lower legs and feet. These symptoms create the so-called Lofgren syndrome and most often appear in young people. The prognosis for these patients is very good.

In patients with involvement of other organs, symptoms characteristic of a specific organ or system may come to the first plan, e.g. heart, nervous system, visual disturbances, and general symptoms. For this reason, sarcoidosis patients often go first to specialists other than pulmonologists - to rheumatologists, laryngologists, ophthalmologists, cardiologists, general surgeons, oral surgeons, dermatologists, and other specialists.

**What tests should be done to confirm sarcoidosis?**

Sarcoidosis is diagnosed based on the typical clinical picture, radiological changes typical of this disease (on a chest X-ray or computed tomography), and a biopsy examination in which a pathologist under a microscope confirms the presence of so-called epithelioid granulomas.

There are many sites from which specimens can be taken, including mediastinal lymph nodes, lung, bronchus, skin, peripheral lymph nodes, oral mucosa, salivary glands, and other organs, depending on availability. The most recommended method is a needle biopsy of the mediastinal nodes performed during bronchofiberoscopy (inspection of bronchi)combined with an ultrasound examination (EBUS examination). It is used in patients with enlarged mediastinal lymph nodes. In a situation where the disease mainly affects the lungs, it is recommended to perform a biopsy from the peripheral parts of the lung through the bronchial wall, using biopsy forceps or freezing (so-called cryobiopsy). These procedures are performed under the so-called intravenous premedication, i.e. after intravenous administration of sedative and anxiety-reducing drugs.

During bronchofiberoscopy, we can also perform a biopsy of the bronchial mucosa, which does not cause any significant complications, but the diagnostic effectiveness of this method is low. We often suggest patients perform the so-called bronchoalveolar lavage (BAL). During bronchoscopy, we administer small portions of physiological saline to one of the eighteen so-called lung segments. The result of this test allows us to confirm deviations in the cellular composition typical of sarcoidosis. However, this test does not replace a biopsy.

An equally important element of diagnostics is the performance of several additional tests aimed at assessing the presence of lesions in organs other than the lymph nodes in the chest or lungs. These tests include, among others: ophthalmological examination, ECG, abdominal ultrasound, laboratory tests, including serum calcium concentration, serum creatinine concentration (assessment of kidney function), blood count, and liver function tests.

**What awaits me if I do not consent to the biopsy?**

In certain situations, sarcoidosis can only be diagnosed based on the clinical picture and evaluation of a chest X-ray or computed tomography. This applies to patients with typical clinical symptoms and when the assessment of the X-ray or computed tomography does not raise any doubts. Biopsy may also be omitted if radiological changes have been observed for a long time and are stable. In each situation, the decision about biopsy is made by the doctor together with the patient, after presenting the arguments for and against. The patient always has the right to refuse to undergo invasive tests.

In a patient in whom biopsy is not performed, there is a greater risk of incorrect diagnosis. This may have serious consequences because sarcoidosis requires differentiation from diseases such as blood cancers, metastatic lesions in the lungs, tuberculosis, and other interstitial lung diseases.

An important element of diagnostics is the assessment of lung function. Using spirometry, we assess lung volume and measure diffusion - the ability of respiratory gases to pass between the pulmonary alveoli and the vessel lumen. The doctor may also suggest performing a simple exercise test, the so-called 6-minute walk test, which assesses the impact of physical exercise on breathing parameters.

**What are the risks associated with diagnostic procedures?**

The biopsy techniques described above (lymph node biopsy, lung forceps biopsy, cryobiopsy) are safe. The most common complication of lung biopsy is pneumothorax (damage to the membrane covering the lung called the pleura, as a result of which causes air to accumulate between the lung and the chest wall. This complication occurs in approximately 10% of patients undergoing cryobiopsy and approximately 6% of patients undergoing forceps biopsy. Most often, pneumothorax absorbs itself within a few days, less often a thin tube (drain) needs to be inserted into the pleural cavity and the air should be sucked out of it. A rarer complication, more common in patients undergoing cryobiopsy, is bleeding from the respiratory tract. The technique of performing this procedure is designed to minimize this risk or to quickly stop the bleeding if it occurs.

Bronchoalveolar lavage (BAL) is a safe procedure. After the test, you may experience fever and flu-like symptoms. These symptoms disappear no later than the next day and are alleviated by taking paracetamol or another antipyretic drug.

**Will I be treated once the diagnosis is made?**

Treatment of sarcoidosis involves the administration of steroids, immunosuppressive drugs (which lower the natural immunity) or so-called biological drugs. Treatment is usually long-term and most often continued for several months.

The decision to initiate treatment is made by the doctor and the patient. Lung lesions usually tend to go away on their own (without treatment). Therefore, the most common approach is to wait and see and only observe the course of the disease during subsequent visits. Your doctor will suggest you start treatment when your symptoms are very severe and make everyday functioning difficult, when he or she detects changes in organs that may result in serious, irreversible consequences, or when changes in the lungs intensify and cause the appearance or exacerbation of symptoms (e.g. shortness of breath, worsening exercise tolerance) and worsening of the results of tests assessing lung function.

**Will the treatment be well tolerated?**

Side effects associated with long-term use of glucocorticosteroids (steroids) include increased appetite and weight gain, gastritis, gastric and duodenal ulcers, diabetes, osteoporosis, mood disorders, sleep disorders, and increased tendency to infections. In people with contraindications to their use, treatment with an immunosuppressive drug (e.g. methotrexate) may be proposed, simultaneously with or without a lower dose of steroids.

Treatment with immunosuppressive drugs is associated with the risk of infection, liver dysfunction and bone marrow damage.

For this reason, treatment must be systematically monitored for side effects.

**What will happen to me if I don't start treatment?**

Using medications when there are no indications for treatment is a mistake and may cause side effects. In most cases of sarcoidosis, as already mentioned, treatment is unnecessary.

However, abandoning treatment when there are indications for its implementation may result in permanent, irreversible changes in the affected organs, leading to damage to the organ's functions. For example, failure to treat ocular sarcoidosis may result in loss of vision, and failure to treat calcium metabolism disorders or renal sarcoidosis may result in failure of this organ and the need for dialysis.

**What symptoms indicate the progression of the disease?**

In the respiratory system, symptoms of advanced changes in the lungs or their intensification over time include shortness of breath during exercise, worsening of exercise tolerance, and worsening cough. Objective assessment involves a comparative analysis of the results of radiological examinations and functional tests (spirometry, diffusion), which will demonstrate the deterioration of the measured parameters.

Symptoms of disease progression in other organs will be specific to a given organ location.

**How often should I visit my doctor?**

Typically, visits to a specialist clinic should be scheduled initially every 3 or 6 months. In situations where the disease is mild and previous tests have not shown any worsening of the results, visits can be scheduled less frequently, for example once a year. During the visits, selected tests are repeated, most often a chest x-ray, ECG, ophthalmological examination and blood tests.

**When can I stop visiting my doctor regularly?**

If your doctor determines that the disease has resolved or its course is stable, periodic visits to a specialist clinic should be continued for at least 2 years. If the course of sarcoidosis was severe, relapses occurred or treatment was carried out, periodic specialist assessment should be continued for several years, and in the most severe cases for life.