Primary Amyloidosis (AL amyloidosis)

Your doctor has just diagnosed that you have “primary amyloidosis”, also called AL amyloidosis.
Amyloidoses are diseases linked to deposits in various organs of amyloid substance formed through an accumulation of certain proteins in the form of insoluble fibres. These deposits impair the normal functioning of the affected organs and explain the symptoms of this disease.
The form of amyloidosis that you are affected by is called AL amyloidosis or primary amyloidosis; A for amyloidosis and L for light immunoglobulin light chain.
The term “primary” indicates that it is not secondary to an inflammation, as is the case with AA complications.

AL amyloidosis is characterised by aggregation of part of the immunoglobulin (or antibody), called a light chain, in the form of fibrils which then are deposited in tissues. Light chains are most often produced by cells in the bone marrow known as plasma cells. The role of plasma cells is to produce the antibody the body needs in order to protect itself against infections. It happens that one of these cells becomes immortal and continues to divide itself into identical cells; this is then called forming a clone. All the cells from this clone produce the same immunoglobulin, called monoclonal. This mechanism is responsible for the cancer of the blood called myeloma. In the case of AL amyloidosis, the affected cells resemble those affected by myeloma, but they don't tend to proliferate as in case of that disease. AL Amyloidosis is therefore not considered to be a cancer.

The amyloid substance appears in the form of small insoluble fibrils which form deposits within several organs and progressively impair their functioning. The organs most often affected are the kidneys, the heart, the liver and the peripheral nervous system. Deposits of amyloid substance may form in any organ in the body except the brain.
Each year, about 500 new cases of AL amyloidosis are diagnosed in France. This disease affects men a little more often than women. It occurs most often in people aged between 60 and 70, but may affect much younger patients.

AL amyloidosis is not contagious. It is not hereditary, unlike other forms of amyloidosis which are linked to genetic mutation.

Doctor's notes :

Signs of the disease

AL amyloidosis is a disease that develops slowly and which for a long time does not entail any particular sign. As the deposits of amyloid substance within an organ become significant the functioning of that organ is more and more impaired. The symptoms that then manifest themselves depend on which organ is affected. In the case of the kidneys being affected, the first consequence is the draining away of proteins via the urine, particularly albumen, entailing oedema and if not treated progressive renal insufficiency. When the latter becomes serious, it may be necessary to have recourse to dialysis.
Where the heart is affected, this basically results in fatigue, breathlessness due to the heart being unable to deliver an adequate cardiac flow because of the infiltration of the cardiac muscle. Affecting the heart may also result in abnormalities in the rhythm of the heartbeat.
In the case of the liver, the effect is most often an increase in the size of this organ, and nothing else shows.
The effect on the nerves may be a sudden loss of blood pressure when standing, pins and needles and numbness in the extremities of the limbs, or even digestive problems such as constipation or diarrhoea, also possible where the digestive system is affected.
The more significant the invasion of one or more organs by the amyloid substance is the more serious are the disorders, Hence the interest in an early diagnosis.

Certain treatments may lead to sterility, especially where men are concerned. So sperm conservation may be offered after the diagnosis.

Participating in a clinical trial

The best way to contribute to the improvement of disease management is to treat patients in the context of clinical trial. If your doctor suggests this could apply to you, he will explain its purpose, protocol, expected benefits, potential risks and will give you an information leaflet.

Participating in a trial of course means you will first have to give your written informed consent.

Useful contacts:

- **Secretarial / appointment:**
- **Nursing consultation:**
- **Consulting psychologist:**
- **Social worker:**
- **In an emergency:**

Diagnosis

AL amyloidosis is a disease which is difficult to identify given the wide range of disorders it can give rise to. Moreover, at the outset of the disease, these signs may be relatively discreet. So sometimes a certain amount of time passes between the onset of the first signs and the suspicion of a possible case of amyloidosis.

Diagnosis of the disease is based on the examination of tissues in which amyloid substance is present. To do this it is necessary to carry out several biopsies, i.e. the taking of one or more tissue samples. The biopsy may involve an affected organ, which may necessitate a surgical intervention.

But due to the way in which the deposits are disseminated around the body it is also possible to carry out a much less invasive biopsy, from the fatty tissue below the skin, from the skin itself (in an affected area) or from the salivary glands. This way the biopsy is much more straightforward and can be carried out under local anaesthetic.

The sample or samples of tissue are then examined under a microscope by a specialist doctor to identify the deposits of amyloid and determine the disease type.

Once the diagnosis has been arrived at, various tests are carried out to see the state of advancement of the amyloidosis and identify which organs are affected, for example an echocardiogram to assess the functioning of the heart.

It is also necessary to define the cells producing the monoclonal protein by examining the marrow by means of a puncture, most often carried out on the sternum, and to measure the level of monoclonal protein by blood test, with the help of various tests, the most useful being the measurement of the free light chains in the serum.

Treatment

The principal aim of the treatment for AL amyloidosis is to reduce the deposits of amyloid substance. To do this, it is necessary to reduce as much possible the production of monoclonal immunoglobulin, and therefore to destroy the clone of cells that produce it. Most often the cells are similar to myeloma cells and the treatments offered are those which are effective against myeloma. So the treatment of AL amyloidosis has greatly benefited from the significant progress made in treating this disease.

The cells responsible may also be close to those of other haematological diseases (lymphomas or chronic lymphoid leukaemia) and the treatment will then be the one used for these illnesses. The effectiveness of the treatment is measured in terms of the reduction of the level of the monoclonal light chain in the blood, which for the most part can be measured by a specific test. If this reduction is significant, deposits of amyloid substance will be progressively eliminated by the body, bringing with it a progressive improvement in the clinical state. How quickly the deposits are eliminated varies according to the organ involved. It may happen in a few months in the case of the liver, but take several years where the heart is affected.

In parallel, specific treatments to make up for the insufficiencies of the organ or organs affected are generally put in place. In cases where the heart or kidneys have been seriously affected, a transplant may be considered.

AL amyloidosis is a serious disease in which the prognosis is very harsh in the absence of effective treatment. Current treatments enable the majority of affected patients to respond; leading to a progressive improvement with regard to symptoms and transforming their life expectancy.

Patients association → *Cancer Support France* *Haute-Vienne* HELPLINE: 06 04 43 39 87
Internet site: www.cancersupportfrance.org
Association France Lymphome Espoir
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