Hematology Cancer Consortium

AMYLOIDOSIS

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- **What is Amyloidosis?**

Amyloidosis is a rare group of diseases that occurs when an abnormal protein, called **amyloid**, builds up in your organs and interferes with their normal function. Amyloid isn't normally found in the body, but it can be formed from several different types of protein. This abnormal deposition of the abnormal protein can happen systemically (though out the body) or locally (in one tissue or organ). About 30 different types of proteins are known to cause these deposits in the body.

- **Which organs can be affected by Amyloidosis?**

Amyloidosis can either be localized involving only the skin or it can be systemic when the visceral organs are affected.

Organs that may be affected by amyloidosis include

- Kidneys
- Heart
- Liver, Spleen
- Gut, Tongue
- Skin
- Bones, Joints
- Nervous system
- Blood vessels and blood clotting
- Eyes

Amyloid deposits cause disease by gradually accumulating within these organs and thereby disrupting the structure and damaging the function of the affected tissues. In some cases, previously healthy organs can be substantially replaced by extensive amyloid deposits.

- **Who can get Amyloidosis ?**

Amyloidosis is a rare disorder. There are no clear data from our country on the incidence of Amyloidosis though worldwide it is seen in eight out of one million people. This is generally a

disorder of the middle-aged and older people, although sometimes the disease can be seen in young people as well. Mostly, men are affected.

- **Why does Amyloidosis occur?**

The cause is unknown. Although it is an abnormal protein, that causes this disease, there is no recognized link between your food intake, stress, occupation or environmental exposure. It does not spread from person-to-person. Some very rare types of amyloidosis can be inherited.

Around 15% of patients who have a blood cancer called multiple myeloma can develop primary amyloidosis. Conversely, 10% of patients with primary amyloidosis can have myeloma.

- **What are the common types of Amyloidosis?**

There are many types of amyloidosis depending on the origin of the abnormal protein. The most common amyloidosis (60-70%) belongs to a type called ***primary systemic (AL) amyloidosis.*** This disorder is due to excessive proliferation of abnormal plasma cells in the blood which causes excess production of the amyloid protein leading to symptoms. As mentioned earlier, this type can be associated with a blood cancer called multiple myeloma.

There are also other rarer causes like AA amyloid (caused by long standing diseases like tuberculosis, rheumatoid arthritis etc) and chronic kidney failure. There are also very rare types of amyloidosis which are seen in families.

- **What are the symptoms of Primary Amyloidosis?**

The symptoms of this disease depend on the organs that are involved. A wide range of symptoms makes primary amyloidosis very difficult to diagnose. Some people may not have any symptoms at all. The usual symptoms include:

- Weakness, fatigue
- Weight loss or weight gain
- Swelling in the ankle and leg
- Shortness of breath
- Numbness or tingling in the hands or feet
- Dizziness or light-headedness upon standing

- Feeling of fullness in the stomach after eating small quantities of food
- Loose stools
- Easy bruisability of skin – face, chest wall, eyelids

- **How is Primary Amyloidosis diagnosed?**

Diagnosis is made by demonstrating amyloid deposits in organs and tissues in your body. In order to provide the best treatment, your doctor would like to know – what type of amyloid is deposited, where it is deposited, and how much is the organ affected due to the protein deposition. For this, the doctor may examine you and conduct a few blood and urine tests. Your doctor would also do a bone marrow examination and biopsies from other organs like rectum, abdominal fat or skin. These are minor procedures and are usually done under local anaesthesia. Occasionally, biopsies are taken from the liver, kidneys, nerves, and rarely from the heart. The doctor would also do tests to measure the extend of organ involvement like an echocardiogram, cardiac MRI etc. Once these reports are ready your doctor will be able to correctly identify the type and severity of amyloidosis and decide the best treatment for you. In addition, your doctor may also order some special tests to rule out rarer types of amyloidosis.

- **How is Primary Amyloidosis treated?**

Treatment usually involves using medicines to try to limit the production of the amyloid protein. Any other condition that may be responsible for the generation of the amyloid protein should also be identified and treated.

In primary systemic or AL amyloidosis, the abnormal plasma cells are responsible for the disease. In this disease, treatment has to be directed towards the plasma cells. The treatment is same as for multiple myeloma and includes chemotherapy in a tablet or injection form. After chemotherapy if you are medically fit, the doctor may do an autologous stem cell transplantation.

During and after treatment, measurements of protein abnormalities in the blood and urine are done on a regular basis to monitor how the treatment is affecting your condition. Treatment is successful when there is a decline in the abnormal protein. The most common protein measurement used is called monoclonal protein (M-protein).

In addition to your haematologist, the management of amyloidosis depends on which organ is involved. For example, kidney or cardiac involvement by amyloidosis will include management by a kidney specialist or a heart specialist, respectively.

Excluding primary systemic amyloidosis, in all the other types, your doctor will order tests to find out the cause for amyloidosis and will offer you treatment based on the cause.

- **Other than chemotherapy, what should I be careful of?**

Well balanced and clean food is important for your body's immune system. Ask your doctor very clearly if you need any dietary restrictions or nutritional supplements. You should also drink clean water and should avoid direct exposure to people with known infections.