Medicine for Managers

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Amyloidosis

Amyloidosis is a serious disease (or more correctly a group of diseases) of which most people have never heard but it can cause damage in the tissues and vital organs throughout the body. The incidence is thought to be about 2-4 per 100,000 people in the United Kingdom each year. Depending on the nature of the disease the symptoms can be very variable.

The nature of the disease depends on an abnormality in body proteins.

A protein molecule is simply a string of amino acids. Like all molecules these proteins form a three-dimensional shape. In amyloidosis the shape is abnormal and the molecules form what is known as a *beta-pleated sheet*. Different forms of the disease affect different proteins and different organs and tissues are involved. In Alzheimer's disease, one particular type of abnormal amyloid protein is found in the brain. In all over twenty different types of amyloid protein have been identified.

Amyloidosis may be localised or generalised. One form of the disease affects only the skin causing an irritating rash. Most other forms are widespread causing systemic effects on a range of organs resulting in a variety of symptoms and leading to progressive organ failure which is very serious.

The presenting features of amyloidosis are often insidious and diagnosis may be delayed because of their non-specific nature.

> There is a National Amyloidosis Centre at the Royal Free Hospital Foundation Trust in London

Early features include fatigue, weight loss, breathlessness, feelings of faintness and paraesthesia (tingling). The symptoms may take a lot of years to develop and the disease therefore commonly presents in older people.

If the amyloid is deposited in the heart, function will be impaired and heart failure will follow. Similarly, if deposited in the

kidneys, kidney failure will occur. Other signs include enlarged liver, bowel abnormalities with malabsorbtion or bleeding and macroglossia (an enlarged tongue) which is quite common.

The abnormality in amyloidosis is an abnormal folding of the protein molecules (sometimes referred to as light chain) and are the result of the activity of abnormal white blood cells in the bone marrow. The white cells are not cancerous in most cases but, in multiple myeloma, they are malignant.

The **A**bnormal **L**ight chains involved result in the disease often being called ***AL Amyloidosis***.

Because of the non-specific nature of the symptoms, making a diagnosis is often very difficult and, because of its relative rarity, is frequently not considered in the differential diagnosis. It should be considered in any case of renal failure, diabetes, lymphoma, cancer (particularly if unconfirmed) and sarcoidosis. Initial tests include a variety of blood tests which will detect any significant changes in the function of any specific organs.

If heart damage is a feature of the disease, changes in the electrocardiogram or an echocardiogram will assist with the diagnosis. A test called ***serum amyloid P scintigraphy*** can be used to assess the quantities of amyloid present in body organs. However, the definitive diagnosis is usually made by taking a tissue biopsy of an affected organ.

The prognosis for people with amyloidosis is not good and sufferers generally deteriorate and develop complications of the disease irrespective of any treatment that they might receive.

For 80% of patients diagnosed with the disease it is fatal and survival is not usually more than two years. The survival in multiple myeloma is poorest and is commonly less than a year from diagnosis.

Much of the treatment for the disease is supportive and directed at specific symptoms affecting specific organs; for example treating the heart failure which may develop if the amyloid infiltrates the heart muscle.

Sometimes chemotherapy is employed but its value is questionable.

Research is occurring into the disease at the Royal Free Hospital and at other centres and perhaps stem cell treatment or developments in chemotherapy will provide an effective treatment in due course.

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