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Wiskott-Aldrich Syndrome

What is Wiskott-Aldrich Syndrome (WAS)?

WAS is a rare inherited condition and is present from birth. It is inherited in an X-linked manner, which means that it only affects males, though females may be carriers.
There are three main features of (WAS). These are eczema, a tendency to bruise and bleed easily and a susceptibility to infections because of immune deficiency. The severity of these three symptoms varies between different patients.

Why does it happen?

Although tiny, every cell in your body has a structure. Different cells have different shapes and structures and rely on their cytoskeleton to keep the correct shape. In WAS, a genetic mutation means that you are unable to produce a protein which has been called WAS protein (WASP). WASP is involved in maintaining the cytoskeleton of some of your cells and in particular the cells in your blood.

What might my symptoms be?

The immune cells are affected by WAS and so the immune system may not be able to fight off infections properly. Antibodies help the immune system fight off infections, and in particular bacterial infections. In WAS the levels of antibodies are often low.
The immune deficiency and low levels of antibodies result in increased susceptibility to infections. The usual pattern is an increase in the number and severity of bacterial infections of the lungs and sinuses. The symptoms result from the infections and may include cough, sputum production, fever, tiredness and muscle pains. Recurrent chest infections can cause damage to the lungs and can results in a condition known as bronchiectasis. In bronchiectasis sputum pools in the damaged areas and causes persistent coughing and also increases the chance of future infections. It is therefore important that chest infections are treated promptly to avoid the development or worsening of bronchiectasis.
There are also problems with platelets in WAS. Platelets are needed for your blood to clot efficiently. Therefore you may have a tendency to bruise and bleed easily.
Eczema usually also occurs, for reasons which are not known. Eczema causes redness, itching and soreness of the skin. It can affect the skin on the inside of the elbow and knee joints as well as other areas of the skin.

How is WAS diagnosed?

The combination of eczema, bruising and increased rates of infections would lead to tests for WAS. However, often cases are less severe or do not have all of these features present which can lead to a delay in diagnosis.

The tests look at whether the protein WASP is present or not. Sometimes genetic tests may be performed as these can help with the diagnosis.

What treatments can be used?

Infections should be promptly treated with antibiotics. Often high dose antibiotics continued for at least 10 days are used. If the episodes of infection are frequent, you may be started on prophylactic antibiotics – this is a low dose of antibiotics taken regularly, even when you are well. The aim of prophylactic antibiotics is to prevent an infection from starting. The antibiotic chosen varies from person to person according to the individual circumstances. For example, allergies to antibiotics, the species of bacteria and the site(s) of infection will all influence the antibiotic choice

If the levels of antibody are low then it is possible to replace these. Antibodies are also known as immunoglobulins. This replaces the low levels of immunoglobulins and can be administered in 2 ways. Intravenous immunoglobulin (IVIg) is given directly into the vein, usually every 3 weeks. Subcutaneous immunoglobulin (SCIg) is given just under the skin, for example on the abdomen, usually weekly. There is a newer method of administering immunoglobulin known as facilitated SCIg (fSCIg) which allows immunoglobulin to be given subcutaneously every 3 weeks. The choice between the different ways of infusing immunoglobulin will be discussed with you and the different methods suit different people. The dose and how often the infusions need to be given varies and is tailored to each individuals requirements.

Often the bleeding does not require any specific treatments, but certain drugs such as aspirin should be avoided. Occasionally a platelet transfusion may be necessary, or sometimes the spleen is removed which helps to keep platelet numbers up.

Eczema is treated in the same way as in other patients. Treatments include moisturiser creams, steroid creams and antibiotics if the eczema becomes infected.

Complications of WAS?

There is a slightly increased incidence of lymphoma in WAS patients compared to the general public. This doesn't mean that if you have WAS you will develop lymphoma. What it does mean is that your chances are higher than that of the general population. Therefore at outpatient clinics examinations and blood tests are performed to help detect any illness at an early stage.

Further information

Please ask us in clinic any questions that you have about WAS or your treatment. If planning a family the genetics issues can be discussed further.

PID UK (Primary Immunodeficiency UK) and UKPIPS (UK Primary Immunodeficiency Patient Support) are patient groups for those affected by immunodeficiencies. Their websites are below (note that we have no editorial control for the websites and take no responsibility for their content):

www.piduk.org
www.ukpips.org.uk

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