



A look at a puzzling condition also known as The AntiPhospholipid Syndrome (APS) Hughes syndrome

Hughes syndrome (HS) is an autoimmune disease in which there is an increased risk of clotting to the blood, both in veins and arteries. As blood is needed by every area in your body to supply oxygen, HS can affect any organ, notably the brain, heart, kidney and the skin. It can therefore present itself with numerous symptoms.

Causes of Hughes syndrome

It is still not fully understood why some people develop HS. As in other autoimmune conditions such as lupus, there are strong hints of a genetic background. There could also be a trigger such as a virus attack. More research is needed before we can give a definitive reason for why such conditions develop in one person and not another.

Clinical features

As I mentioned above there are many, varied symptoms associated with HS. It is unlikely that you will have them all and they can come and go freely.

Blood clotting

These can occur almost anywhere in the body and can cause some serious events such as stroke, heart attack, pulmonary embolism and thrombosis in the limbs.

The brain

Lack of oxygen to the brain can cause a wide variety of features including migraine, epilepsy, memory loss, balance and co-ordination problems and slurred speech. Not surprisingly, many sufferers are misdiagnosed with Multiple Sclerosis as it displays similar symptoms.

Pregnancy

Possibly because of thrombosis in the placenta, the developing fetus does not receive the nutrition it requires. This can cause low birth weight babies, pre-eclampsia or, sadly, miscarriage. In many women with HS recurrent miscarriage is

the predominant feature. Often the pregnancy loss occurs late and some are even stillborn – all causing terrible distress.

Other organs

Clotting in the eye can cause visual disturbance or loss. In the skin leg ulcers may develop, and one common telltale sign is a blotchy skin rash known as 'Livedo Reticularis'.

The degree of illness can vary greatly between people. Some can suffer from most of the symptoms highlighted, yet there are others who never feel ill yet develop a clot 'out of the blue'.

Diagnosis

There are two screening tests for HS: anticardiolipin antibodies (aCL) and lupus anticoagulant (LA). These are universally available and can be carried out at your GP's surgery (don't let them tell you otherwise!).



PHOTOGRAPH COURTESY OF THE HUGHES SYNDROME FOUNDATION

Dr Graham Hughes, who first described the syndrome

Treatment

General measures include avoidance of known risk factors such as smoking and the oral contraceptive pill. The main medical treatment for this condition is

anticoagulation. For those who test positive yet have never presented any serious thrombotic symptoms, the usual form is low-dose aspirin (75-150mg daily) or an alternative anti-platelet agent such as PLAVIX).

For those with more severe disease, anticoagulation with warfarin (or heparin during pregnancy) is vital. As most readers will know the level of anticoagulation is guided by a blood test called the INR. For most cases of thrombosis, an INR of 2 is generally satisfactory. However, for more severe clotting problems, especially strokes and other neurological disorders the INR may have to be kept at 3 or even 3.5. This has inevitably proven to cause many HS sufferers problems at their local anticoagulation clinic. Many doctors, who don't understand the disease, are wary about keeping the INR high due to risk of haemorrhage. However, haemorrhaging is far less likely at an INR of 3, than clots are at an INR of fewer than 2.

Outcomes and the future

Hughes syndrome is now known as one of the most common autoimmune diseases and is potentially treatable. In obstetrics, for example, successful pregnancy is now the rule rather than the exception.

There is still much we don't know about HS, but as it is becoming increasingly recognised in the medical world, and more and more research is being carried out, we will soon hopefully start to gain a fuller picture.

Hughes Syndrome Foundation

The Foundation was set up three years ago at St Thomas' Hospital in London, to provide information and support to sufferers of Hughes syndrome and to increase awareness of the disease. Our long-term aims are to help fund medical research into this important condition.