Medical and research news - The Postural Tachycardia Syndrome and EDS III

The previously well recognised features of Ehlers-Danlos III (EDS III) or Joint Hypermobility Syndrome (JHS), include fragility of skin and supportive connective tissue, fatigue, soft tissue lesions and pain. However, recent clinical and research experience indicates that there are a number of previously unrecognized symptoms that are not related to the musculoskeletal system and are predominantly due to autonomic dysfunction. These relate mainly to disturbances in control of heart rate and blood pressure. Current ongoing research in our group suggests that a large number of, but not all, EDS patients have palpitations, light-headedness/dizziness near fainting, and also fainting (syncope), mainly when upright. These symptoms are related to alterations in the usual autonomic responses when standing or sometimes even sitting.

The abnormally rapid heart rate changes to positional change are part of a relatively newly recognised autonomic nervous system disorder, called the Postural Tachycardia Syndrome or PoTS. We consider it to be a transient or intermittent autonomic dysfunction, as in many, testing of the autonomic nervous system excludes autonomic failure and thus more serious disorders usually associated with damage to autonomic nerves and pathways. It appears to affect mainly young people, from the teenage years to about 50, and there are more women affected than men.

Some of the reasons for PoTS and the associated symptoms are described below. When we stand up blood shifts down to the legs due to the effects of gravity.

To ensure not too much blood pools in the legs and enough is returned to the heart, so as to safely maintain blood pressure, the autonomic nervous system directs a number of important cardiovascular responses through the sympathetic and parasympathetic nerves. An increase in sympathetic nerve activity causes heart rate to increase and also narrows blood vessels to reduce the amount of blood that flows through them and thus maintain blood pressure.

In contrast, parasympathetic nerve activity decreases, which raises heart rate. PoTS is characterised by an impaired inability to withstand the upright posture, and is a cause of orthostatic or postural intolerance. Blood pressure while standing does not fall, in contrast to patients with orthostatic (postural hypotension). However, there is an excessive increase in heart rate (greater than 30 beats/min or a heart rate above 120 beats/min) during the first few minutes of assuming the upright posture, in response to too much blood 'pooling' in the lower body. Symptoms and signs associated with PoTS include palpitations, light-headedness/ dizziness, impaired vision, a purplish or blotchy discoloration of skin over the legs as a result of excessive pooling of blood, and sometimes fainting. These usually are reversed by sitting or lying down.

In contrast, parasympathetic nerve activity decreases, which raises heart rate. PoTS is characterised by an impaired ability to withstand the upright posture, and is a cause of orthostatic or postural intolerance. Blood pressure while standing does not fall, in contrast to patients with orthostatic (postural hypotension). However,

there is an excessive increase in heart rate (greater than 30 beats/min or a heart rate above 120 beats/min) during the first few minutes of assuming the upright posture, in response to too much blood 'pooling' in the lower body.
Symptoms and signs associated with PoTS include palpitations, light-headedness/dizziness, impaired vision, a purplish or blotchy discoloration of skin over the legs as a result
of excessive pooling of blood, and sometimes fainting. These usually are reversed by sitting or lying down.

Our previous research and clinical experience in autonomic disorders had also alerted us to consider other factors in addition to standing that may make the symptoms of PoTS worse—these include dehydration, a hot environment, exercise and food. Many have a low blood pressure level, even while lying flat, which may make them more prone to symptoms when upright. We also recognise that a small subset of patients with PoTS and EDS III may also have symptoms suggesting involvement of autonomic function affecting the bladder and gut, which is studied with our specialist colleagues. The exact cause, or causes, of PoTS is not known and needs further research.

Recognition of autonomic symptoms, and diagnosis of PoTS, especially in EDS (as it may occur independently of EDS), is thus especially important.
We have 2 departments, the Autonomic and Neurovascular Medicine Unit at St Marys Hospital, Imperial College London, and the Autonomic Unit at the National Hospital for Neurology and Neurosurgery, Institute of Neurology Queen Square, that have pioneered many advances in the diagnosis, understanding of causes, and treatment of autonomic disorders over the last 3 decades. We have been utilising this experience to now help further understand the problems in PoTS to improve diagnosis and treatment.

We have focused over the years on ensuring safety during testing, using the latest non-invasive technology and with highly experienced staff. The testing also ensures that we are aware of individual differences between patients with PoTS, as this is needed in deciding which of a number of non-pharmacological and/or drug strategies are needed to reduce or ideally abolish the symptoms of PoTS, which can vary between patients. The knowledge we have in our departments in dealing with multi-system and often complex disorders over the years, as part of a team that includes clinicians, clinical scientists, autonomic nurse specialists with clinical research and support staff is of particular relevance to PoTS especially when associated with EDS III.

Our team also work closely with various colleagues in different specialties to ensure we provide comprehensive management in a condition where much still needs to be learnt.

Dr David Low, Clinical Research Lead, and Professor Christopher Mathias