

Extra articular manifestations of hypermobility

RA Hughes

Dermatological



Papyraceous



Haemosiderin scar deposition

Depressed scars



Molluscoid
pseudotumour of the
heel





Multiple ecchymoses and
hematomas

Dysautonomia

2002 – Rodney Grahame Am J Med

- Study of 48 hypermobile EDS Type 3

Symptoms related to the autonomic nervous system, such as syncope and presyncope, palpitations, chest discomfort, fatigue, and heat intolerance, were significantly more common among patients.

Orthostatic hypotension, postural orthostatic tachycardia syndrome, and uncategorized orthostatic intolerance were found in 78% (21/27) compared with in 10% (2/21) of controls.

Patients with the syndrome had a greater mean (\pm SD) drop in systolic blood pressure during hyperventilation than did controls (-11 ± 7 mm Hg vs. -5 ± 5 mm Hg, $P = 0.02$) and a greater increase in systolic blood pressure after a cold pressor test (19 ± 10 mm Hg vs. 11 ± 13 mm Hg, $P = 0.06$).

Patients with the syndrome also had evidence of α -adrenergic (as assessed by administration of phenylephrine) and β -adrenergic hyperresponsiveness (as assessed by administration of isoproterenol).

Semin Arth Rheum 2013

- 39 females with EDS-HT and 35 age-matched controls underwent autonomic function testing.
- *Resting autonomic tone* was assessed using heart rate variability (frequency domain) and baroreflex sensitivity analysis (cross correlation).
- *Autonomic reactivity* was assessed using the Autonomic Reflex Screen test battery. Factors suspected to contribute to dysautonomia, e.g., neuropathy, medication use, decreased physical activity, depression, pain-induced sympathetic arousal, and connective tissue laxity, were quantified using validated questionnaires, the Beighton score, and measurement of skin extensibility.

Mechanisms of dysautonomia

- Neuropathy – sympathetic neurogenic dysfunction
- Connective tissue laxity
- Vasoactive medication

- There are two causes for Dysautonomia in JHS

Autonomic Nervous System dysfunction, characterized by:

- Orthostatic hypotension or POTS (tachycardia).
- Symptoms: Palpitations, lightheadiness, tiredness, dizziness, somnolence, poor thermostat regulation, chronic fatigue, presyncope or syncope.

Increased venous pool derived from collagen weakness of the venous wall..

Dysautonomia

- Low blood pressure is desirable
- Dysautonomia often goes undiagnosed and can lead to large effects on Q of L and happiness

Aggravating factors

- Dehydration
- Hot and prolonged baths.
- High altitude.
- Standing for too long.
- Getting up suddenly from a bed or a chair (as in church).
- Acute pain, associated with anxiety (eg injections)

Treatment

General measures

Avoid standing for too long, without moving the feet.

Avoid walking slowly.

When sitting for too long is necessary to move hands, feet and knees frequently and to get up and walk.

Rest for 15 to 30 minutes after lunch or when too tired.

Drink 2 to 3 liters of fluids a day.

Add 6 to 9 grams of salt a day (if there are no contraindications).

Use elastic stockings.

Practice Yoga, Pilates, Tai Chi or moderate aerobic exercise.

Treatment - drugs

A. Mineralocorticoid: Fluudrocortisone 0.1 mg tablets.

Effect lasts 24 to 36 hours

B. Beta adrenergic blocker: Midodrine 2.5 and 5 mg tablets

Effect lasts only 4 hours

C. Propanolol is used in cases of POTS (Postural Orthostatic Tachycardia).

Effects of treatment

- Dysautonomia is very frequent in hypermobility and causes poor quality of life, sometimes for years.
- The treatment of Dysautonomia is very effective, the problem is that Dysautonomia is usually overlooked.
- Adolescents feel much better and improve their grades at school.
- Not only the patient benefits from treatment, but also the people around him/her.

Pitfalls of dysautonomia

The diagnosis is usually made in adolescent or young females, with time, people end up believing that tiredness is a part of themselves

Most patients do not have syncopal episodes.

The ones that do, seek medical attention, but the condition is seldom diagnosed and they get multiple exams and see multiple specialists without getting the proper diagnosis and treatment.

Hypertensive patients can have Dysautonomia, but the treatment gets more complicated.

Mucosal problems

- Mucosal involvement is common in JHS/EDS-HT.
- Xerostomia, xerophthalmia, and vaginal dryness are frequent
- Together with hypohidrosis, mucosal xerosis could be a remote consequence of autonomic dysregulation
- Blue sclerae are overrepresented among JHS/EDS-HT patients and are likely caused by more visible uveal blood vessels through thinner sclerae.
- Focal blue-purple discolorations of the oral mucosa are not uncommon in JHS/EDS-HT,

Gastro-intestinal

- Chronic (slow transit) constipation, hiatus hernia, Crohn's disease faecal incontinence, rectal evacuatory dysfunction, and functional gastrointestinal disorder
- Typical gastrointestinal features include gastroesophageal reflux (74%) with or without hiatus hernia, chronic/recurrent gastritis (48%), symptoms of delayed gastric emptying, recurrent abdominal pain (68%), and constipation/diarrhea (72%)

Gynae / urological

- Pelvic prolapse is the most debilitating gynecologic feature of JHS/EDS-HT

Clinical manifestations mainly include

- Urinary stress incontinence
- Uterine prolapse
- Faecal incontinence.