Dysautonomia

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Persons with identified mitochondrial disorders may have features of autonomic dysfunction. This may include the following problems:

1) gastrointestinal dysmotility with resultant constipation and pseudo-obstruction, and in some circumstances intermittent diarrhea and rectal incontinence;
2) body temperature dysregulation, with inappropriately decreased or increased sweating;
3) postural hypotension, including symptoms of ligthheadedness, fainting or near-fainting spells that may or may not be associated with exercise, brief loss or dimness of vision, sensation of weakness;
4) cardiac conduction defects;
5) sexual dysfunction including impotence, loss of tumescence in females, loss of libido, or retrograde ejaculation (the loss of ability to achieve orgasm has not been studied);

Outside of the purview of the mitochondrial physician, the general physician should consider and treat common medical issues such as blood volume depletion (dehydration), anemia, Addison disease, thyroid disease, complications of diabetes, thermodilation, deconditioning (prolonged lack of exercise or prolonged bed rest), alcohol use, use of diuretics, cimetidine, medications used to treat hypertension, nitrates, potassium depletion and the vasovagal reaction. Adolescent vasovagal and positional fainting are common and not serious medical disorders. Dysautonomia symptoms are common after infections, which may be due to deconditioning and volume depletion although mitochondrial dysfunction has been suspected (but not proven) as a contributing factor. Mitochondrial dysfunction has been explored in many patients but no pattern of lab results or clinical study has shown this to be an identifiable factor. Therefore, we cannot recommend exploring mitochondrial dysfunction in post-infectious states unless the patient is part of a controlled clinical trial. When the cause of dysautonomia is not identifiable, and when there are no other signs or symptoms involving other organ systems, the term "primary dysautonomia" is used. The term "primary" can also be exchanged for the term "idiopathic", which consists "idio" meaning “by itself” or "unknown" and "pathic" meaning "disease." The truth is that there are many causes for the primary dysautonomias and - as we learn more - the term primary dysautonomia will disappear. Regardless, we cannot recommend exploring mitochondrial dysfunction in primary dysautonomia unless the patient is part of a controlled clinical trial.

If dysautonomia is accompanied by typical mitochondrial features, such as PEO, neuropathy or myopathy with objective abnormalities on NCV/EMG, cardiomyopathy, liver dysfunction, high frequency hearing loss, retinitis pigmentosa, epilepsy, or dementia, then it is reasonable to consider a focused mitochondrial evaluation. In reviewing the literature, there are patients with dyasautonomia who likely had a mitochondrial disease, but whose diagnosis was primary dysautonomia because these reports were written before mitochondrial disorders became known or because the authors did not recognize the underlying mitochondrial disease. These include patients

with symptoms of progressive external ophthalmoplegia (PEO), neuropathy and myopathy.

Using current methods of assessment, including the use of muscle biopsy and genetic testing, we have not been able to identify mitochondrial dysfunction in evaluating patients diagnosed with POTS (Postural Orthostatic Tachycardia Syndrome) or with primary dysautonomia and cannot recommend at this time that such investigation be undertaken. There is also no evidence that standard mitochondrial supplements are helpful in patients with primary dysautonomia. The decision to evaluate any patient must rest in the hands of the physician and the patient, but we advise the primary care doctor to screen patients for the common causes of dysautonomia and to perform a full neurological examination. This statement does not conflict with the belief of many mitochondrial doctors that there may be a mitochondrial component to the post-infectious dysautonomia syndrome or to the primary dysautonomias. However, at this point in time, this is mere speculation.

The treatment of dysautonomia, whether due to mitochondrial dysfunction or not, is best left to the care of the appropriate specialist. Some mitochondrial doctors feel comfortable treating dysautonomia symptoms in patients with mitochondrial disease. However, the treatment of these symptoms falls outside the level of training and comfort of most mitochondrial doctors. In some medical centers, there are dysautonomia experts in the departments of neurology or cardiology. Consultations with gastroenterologists, cardiologists, urologists or other specialists is often required.

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