

# POTS

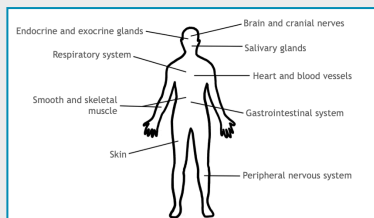
## Postural Orthostatic Tachycardia Syndrome & CHARGE Syndrome

### What is POTS?

POTS is a clinical syndrome that falls under the umbrella of dysautonomic conditions. This means that the symptoms arise from dysregulation of the autonomic nervous system. POTS is characterized by a rapid increase in heart rate upon standing up, and symptoms are often relieved by lying down again. A common “diagnostic test” is the tilt-table test, as shown in the image to the right.

### POTS Symptoms

POTS affects a number of components of the autonomic nervous system:



Common symptoms include: fatigue, weakness, increased heart rate, nausea, constipation, diarrhea, shortness of breath and poor temperature regulation.



### Why Study POTS?

Really, it’s been CHARGE families that have encouraged Dr. Blake and her team to look into POTS and dysautonomia further.<sup>1</sup> After hearing the one family’s story at the 2017 International CHARGE Syndrome Conference, Dr. Blake looked into POTS within her own practice. One case of clinically confirmed POTS provoked further investigation, and POTS is now the subject of ongoing research at the IWK Hospital in Halifax, NS. We strongly believe that understanding this condition and how it presents in CHARGE Syndrome will have a significant impact on the management of CHARGE and improve the quality of life of those who may be experiencing it.

### Preliminary Research

Following the initial case report, a questionnaire was designed in order to investigate the prevalence of POTS-associated symptoms within the CHARGE syndrome population. This questionnaire

## Original Case

A 17-year-old Caucasian female with a clinical diagnosis of CHARGE Syndrome presented to the CHARGE Clinic at the IWK with symptoms suggestive of POTS. These included:

- Physical fatigue on hot days over the last year
- Heart rate that at times elevated to 140 bpm
- Agitation and discomfort when moving from a standing to lying position
- Vomiting, loss of colour in the face, sweatiness, pre-syncope and diarrhea following standing up
- One instance severe enough that prompted a visit to the Emergency Department

At a follow-up appointment, a tilt-table test was performed to investigate the possibility of POTS. Heart rate and blood pressure were recorded throughout the test. The blood pressure did not change, but the heart rate increased by 40 beats per minute when she was positioned to 90°. Lowering her to a supine position returned the heart rate to normal.

POTS is difficult to diagnose, especially with individuals who have multi-sensory impairment. In this case, the caregiver was able to report a number of POTS-associated symptoms, assisting with the diagnosis. Treatment recommendation included awareness of the condition and associated triggers, monitoring fluid and salt intake, and taking breaks during periods of activity.

included both a summary of CHARGE related features, and a “checklist” of POTS symptoms and severity. This questionnaire was piloted at the 2019 International CHARGE Syndrome Conference in Dallas, Texas. A total of 12 individuals took part in the study, and a summary of the POTS-related results is presented below:

- 4 out of 12 reported increased heart rate, or a racing heart “sometimes” or “often”
- 3 out of 12 reported light-headedness sometimes or often
- 7 out of 12 reported fatigue sometimes or often
- 6 out of 12 reported upset stomach sometimes or often
- 8 out of 12 reported trouble passing stool
- 10 out of 12 reported that being in the heat makes them feel sick

For just 12 participants, so far, it is clear to us that there is some pattern of autonomic dysregulation among those who completed the questionnaire. The early, preliminary results have prompted a more detailed exploration of this association.

## Further Investigations

We are currently in the process of creating an online version of our POTS questionnaire for dispersal to the CHARGE community via the CHARGE Syndrome Foundation. This will allow for a wider exploration of our research question, and hopefully allow us to understand more definitively whether there is a connection between POTS/dysautonomia and CHARGE Syndrome.

We hope that our POTS research will inform further investigations into other conditions under the “dysautonomia” umbrella. We know POTS to be incredibly debilitating, especially in combination with CHARGE Syndrome, and it is our goal to understand this connection, and explore others that may exist.

If you have any questions, do not hesitate to reach out to Dr. Kim Blake, or her research student, Julia Morrison at [julia.morrison@dal.ca](mailto:julia.morrison@dal.ca).