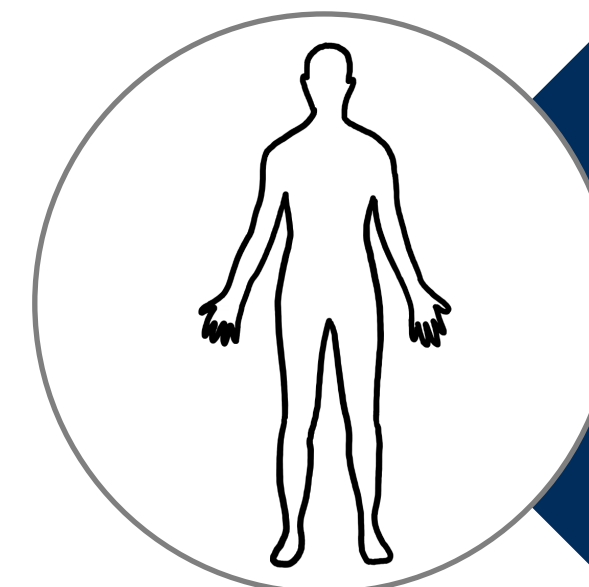



What is POTS?

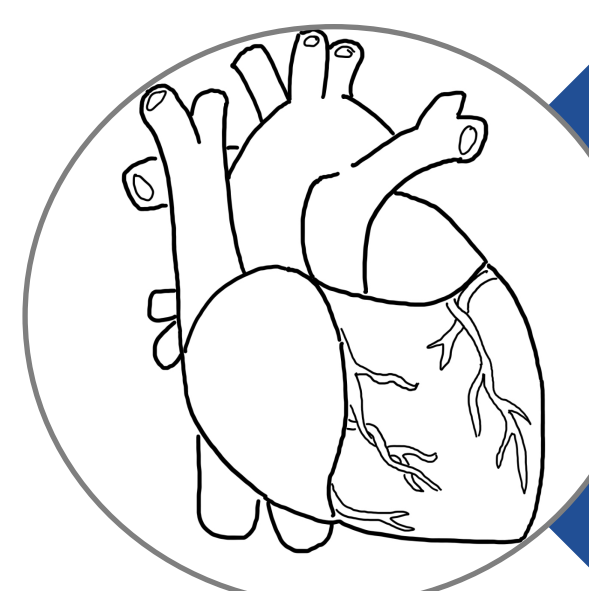
POTS is a debilitating syndrome predominantly affecting young women. People with POTS present with a variety of symptoms in a variety of body systems, as shown below. POTS is characterized by a rapid increase in heart rate upon standing up, and symptoms can often be relieved by lying back down.



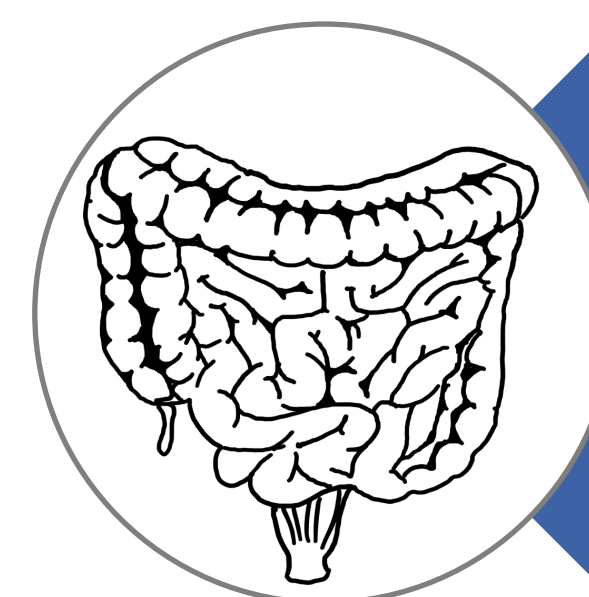
Fatigue, weakness
Muscle pain, exercise intolerance
Tremors



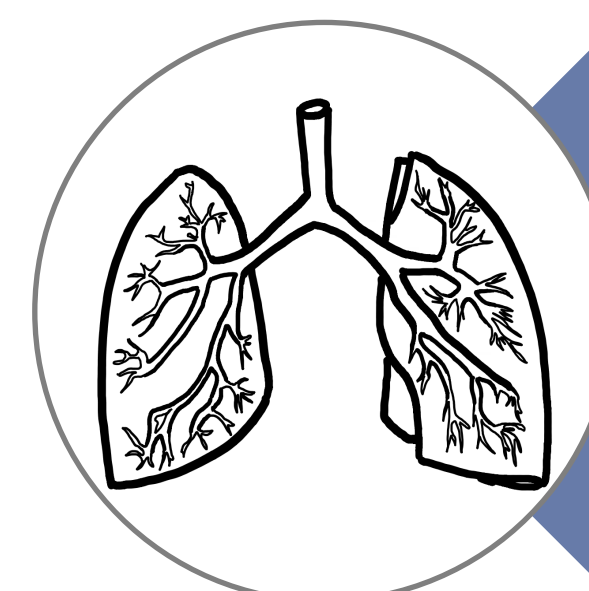
Feeling worried, anxious or depressed
“Brain fog”
Headache, dizziness




Increased heart rate, especially when standing up
Chest pain or discomfort



Feeling nauseous
Getting diarrhea or constipation
Feeling bloated
Getting cramps in your belly



Rapid breathing
Shortness of breath



Poor temperature regulation
Increased or decreased sweating

POTS Diagnosis and Treatment

- The “gold standard” of POTS diagnosis is a tilt-table test. POTS can be diagnosed when there is an increase in heart rate by 30 beats per minute within 10 minutes when moving from lying to standing.
- Treatment of POTS is typically focused on increasing fluid and electrolyte intake. Other treatment options include things like compression stockings and certain medications (i.e. beta blockers, fludrocortisone, midodrine or pyridostigmine).

A Case Report of POTS in CHARGE

- POTS has been diagnosed in one individual with CHARGE syndrome from the Atlantic Provinces in Canada (see below for presentation), and there is clinical suspicion of POTS in another individual.
- A 17-year-old individual with CHARGE who presented to the CHARGE Clinic at the IWK with symptoms of POTS underwent a tilt-table test. The results of this test aligned with a diagnosis of POTS.

Table 1: Symptoms of POTS and symptoms reported by a 17-year-old individual with CHARGE Syndrome

Symptom	Present in patient
Cardiac symptoms	
Increased heart rate on standing	Yes
Palpitations	No*
Light-headedness	Yes
Dyspnea	No
Non-cardiac symptoms	
Syncope	Near syncope
Fatigue	Yes
Weakness	Yes
Sweating	Yes
Tremor	No
Nausea	Yes
Anxiety/change in mood	Yes
Blurred vision	Possible
Headache	Yes
Abdominal pain	No*
Diarrhea	Yes
Vomiting	Yes
Change in colour	Yes (loss of colour in face)



Figure 1. Tilt-table test being performed

Why are we studying POTS?

- The *CHD7* mutation present in individuals with CHARGE Syndrome has an impact on many organs in the body. Many of these are also organs of the autonomic nervous system (ANS), which is the system that gets dysregulated in dysautonomic conditions like POTS.

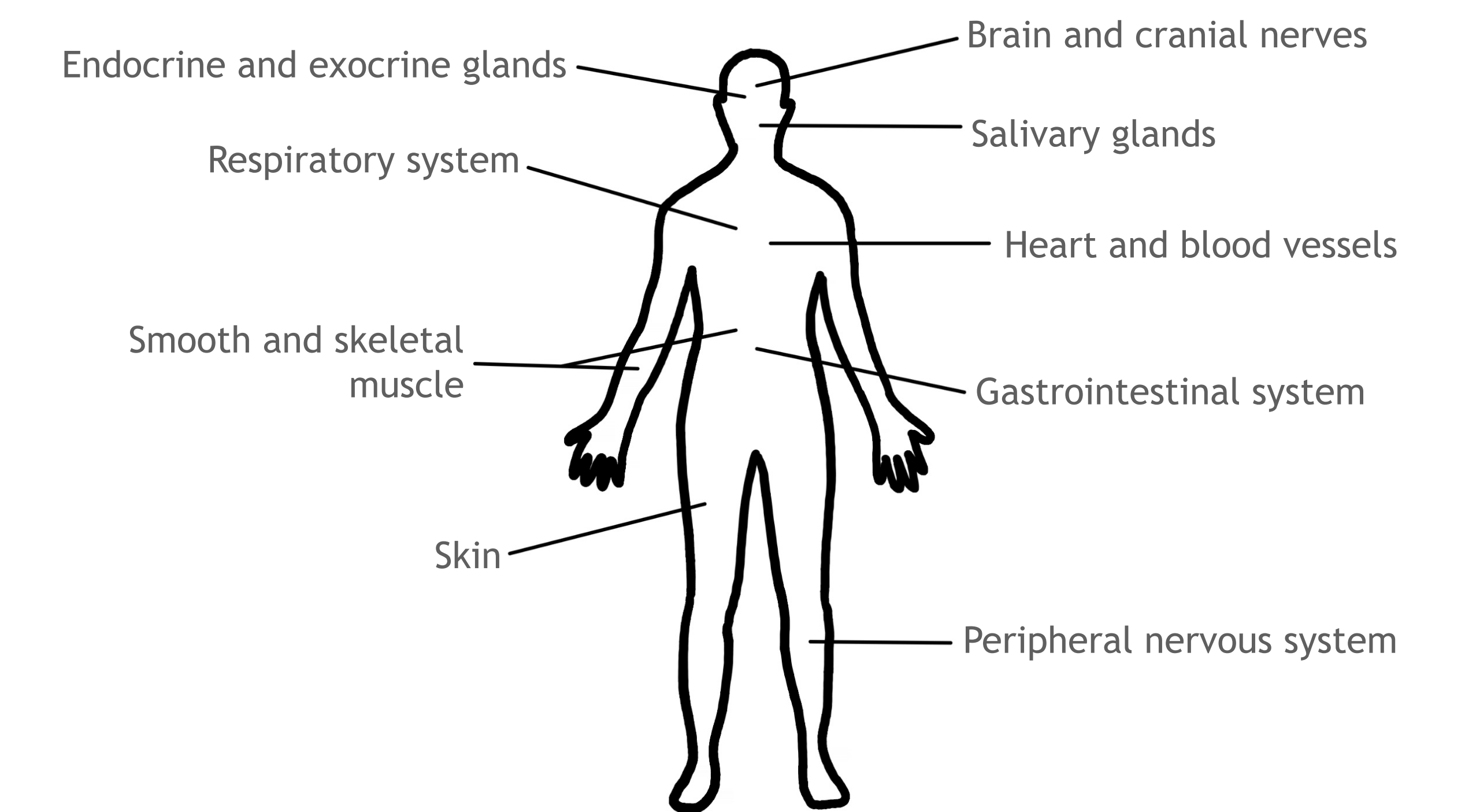


Figure 2. ANS affected organs in CHARGE Syndrome

- GI issues are known to be a factor in both CHARGE and POTS, and this can be related to nervous system dysfunction in both syndromes.
- POTS can induce substantial disability, with data suggesting 25% of patients file for disability and 50% interrupt their education. We believe the combination of POTS and CHARGE can be even more debilitating.
- Future work is planned to increase the understanding and treatment of POTS in CHARGE as well as pursuing connections with the vagus nerve.
- Identifying an association between these conditions is important in the management of adolescents and young adults with CHARGE Syndrome, and awareness will enhance clinical care.

Funding and References

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This poster can be found on Dr. Blake’s website: <https://www.drkimblake.com>

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