Postural tachycardia syndrome (POTS) in a patient with familial adenomatous polyposis (FAP): A genetic cause for autonomic dysfunction ?

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LEARNING OBJECTIVE

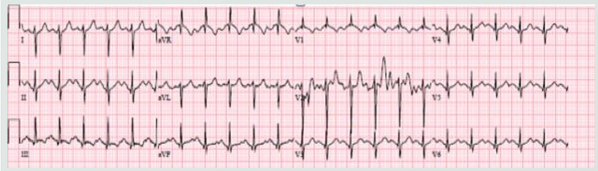
- Diagnose POTS
- Highlighting possible association between FAP and autonomic dysfunction

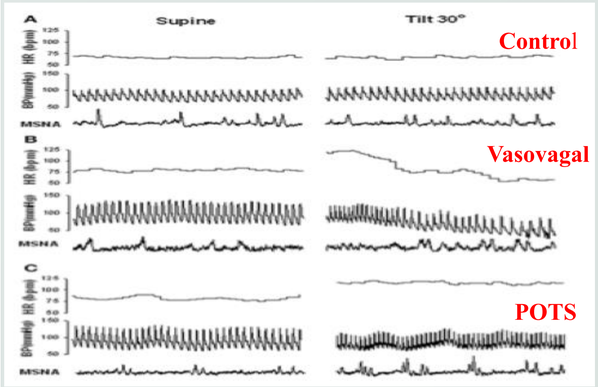
INTRODUCTION

- POTS: Type of autonomic dysfunction defined as an increase in HR by > 30 bpm on standing without significant hypotension (< 20/10 change)
- Affects 1-3 million Americans; 25% are disabled by symptoms)
- APC suppressor protein plays a role in neuronal nicotinic synapse assembly and signaling
- We report a case of POTS associated with familial adenomatous polyposis.

CASE

- A 32-year-old man with PMH of FAP s/p total colectomy in 1999 and multiple duodenal polyps s/p Whipple procedure in 2014 complicated by LE DVT & Pulmonary embolism 4 weeks prior to his admission.
- He presented with recurrent severe palpitations when he gets out of bed or chair associated with chest pain, lightheadedness and pre-syncope.
- His symptoms started more than 2 years ago yet it became more frequent recently.
- BP 110 /75 and supine HR of 85.
- **Orthostatics showed a HR of 145 immediately after standing and BP of 105/ 80 which were unchanged** after an additional 2 minutes of standing.
- Physical exam did not show signs of dehydration.
- Cardiac structure and function as assessed by ECG and ECHO were normal (No RV strain).
- Thyroid and adrenal functions were normal.
- **On telemetry, he had repeated episodes of sinus tachycardia that correlated with him getting out of bed (Bed monitor sign !).**





- POTS was confirmed with tilt table testing.
- Treatment was initiated with low dose beta blocker. On follow-up the patient was improved.

DISCUSSION

- POTS is The most common disorder amongst patients referred to centers specializing in the autonomic nervous system disease.
- **Diagnostic criteria** :
  - **HR increase > 30 bpm**, within the first 10 minutes of standing, **in the absence of orthostatic hypotension.**
- Often diagnosed by a Tilt Table Test.
- **Symptoms**: Palpitations, chest pain, SOB, fatigue, headaches, lightheadedness, palpitations, exercise intolerance, nausea, pre-syncope, acral cyanosis.
- **Pathophysiology**: **(Theories)** hypovolemic, neuropathic, hyperadrenergic and autoimmune (10% have AChR antibodies).
- **Primary vs secondary** (DM, Amylodosis, Sarcoidosis, paraneoplastic syndromes)
- **Treatment**: Fluid intake > 2 liters , salt intake 3-5 gm, compression stockings, beta blockers (in small doses), midodrine, desmopression, pyridostigmine, and octreotide **(off-label)**.
- **Could FAP be another cause of secondary POTS ?**
- Germline mutation in the adenomatous polyposis (*APC*) gene →Classic FAP
- **APC organizes a multi-molecular complex that is essential for targeting α3*nAChRs to neuronal nicotinic synapses which plays a central role in normal autonomic function.**
- APC brings together EB1 + IQGAP1 + 14-3-3 adapter protein at nicotinic synapses→ unites with α3-subunit of nAChR surface membrane → multi-molecular APC complex→ stabilizes the local microtubule and F-actin cytoskeleton and links postsynaptic components to the cytoskeleton (essential for the synapse stabilization).
- **Studying the autonomic manifestations in a case series of patients with FAP might reveal another extra-intestinal manifestation.**

REFERENCES

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