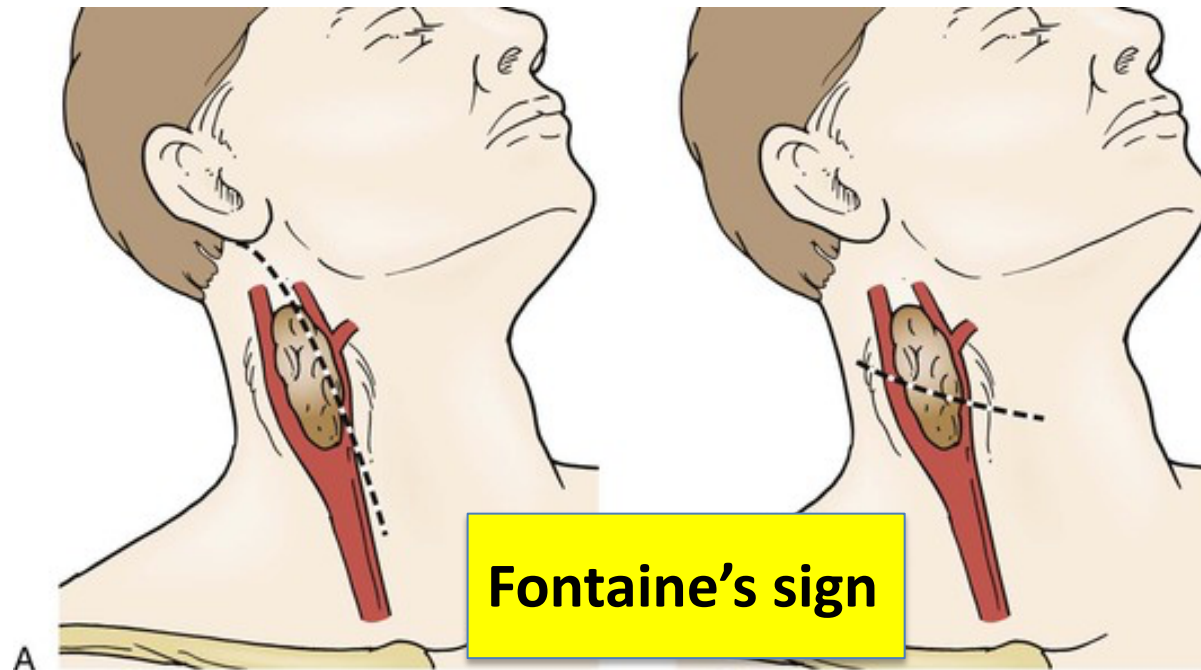


Case 2

Lesson learnt

Bilateral carotid body tumors suspected *SDHD* mutation





Preoperative Endocrine Assessment

- **All patients** (including those with tympanic PGLs) should have **plasma metanephrine levels** measured at presentation (including adrenaline, normetadrenaline, and 3-methoxytyramine)
- **Only 1 test is required** to exclude a secretory lesion
- 24-hr urinary metanephrine measurement may also be used, but this is less sensitive

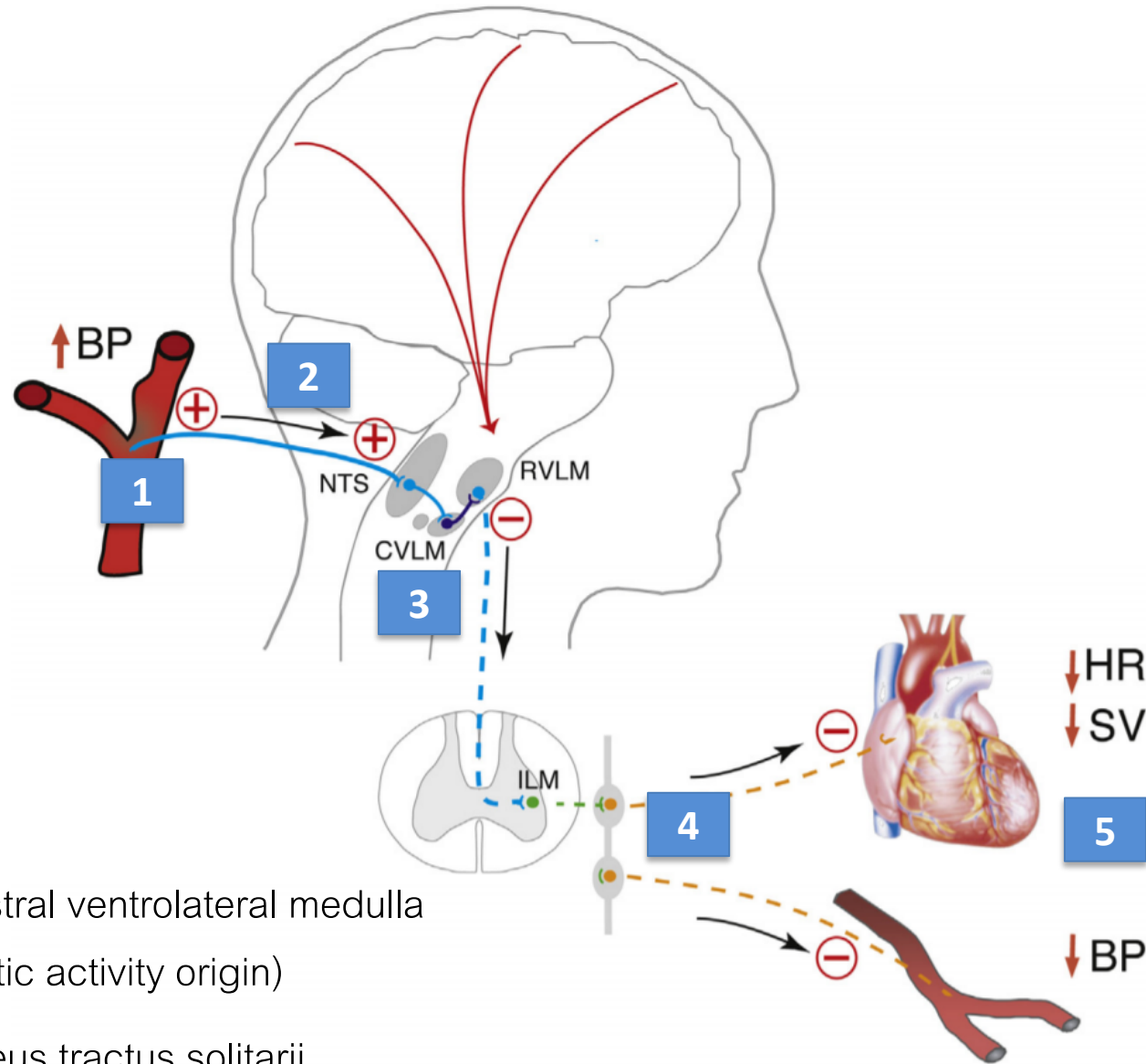


SDHD associated PPGL: PGL1

- Most common type of familial paraganglioma syndrome
- Maternally imprinted gene (**paternally inherited mutation**)
- **92% had PGL** (22% sympathetic and 84% **parasympathetic** PGL)
- 56% had multiple PGLs
- Penetrance of PCC/PGL in *SDHD* mutation carriers has been estimated to 86% by the age of 50 years
- **50-80% of Carotid body paraganglioma** has *SDHD* mutation

80-90% present with head and neck paraganglioma

Baroreflex



1. **Elevated BP** activate baroreceptor in Carotid sinus

2. **Afferent** signals are sent through the **glossopharyngeal nerve (IX) to activate NTS**

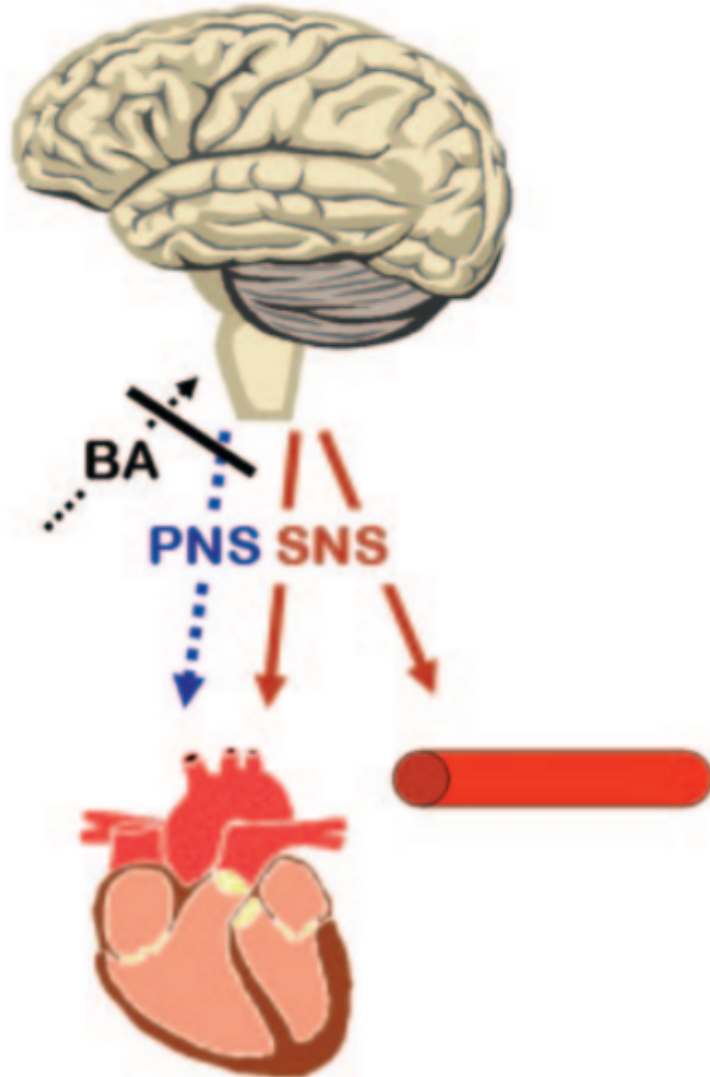
3. **Inhibitory input** to the RVLM

4. **Decrease** in **sympathetic tone** to heart and blood vessel

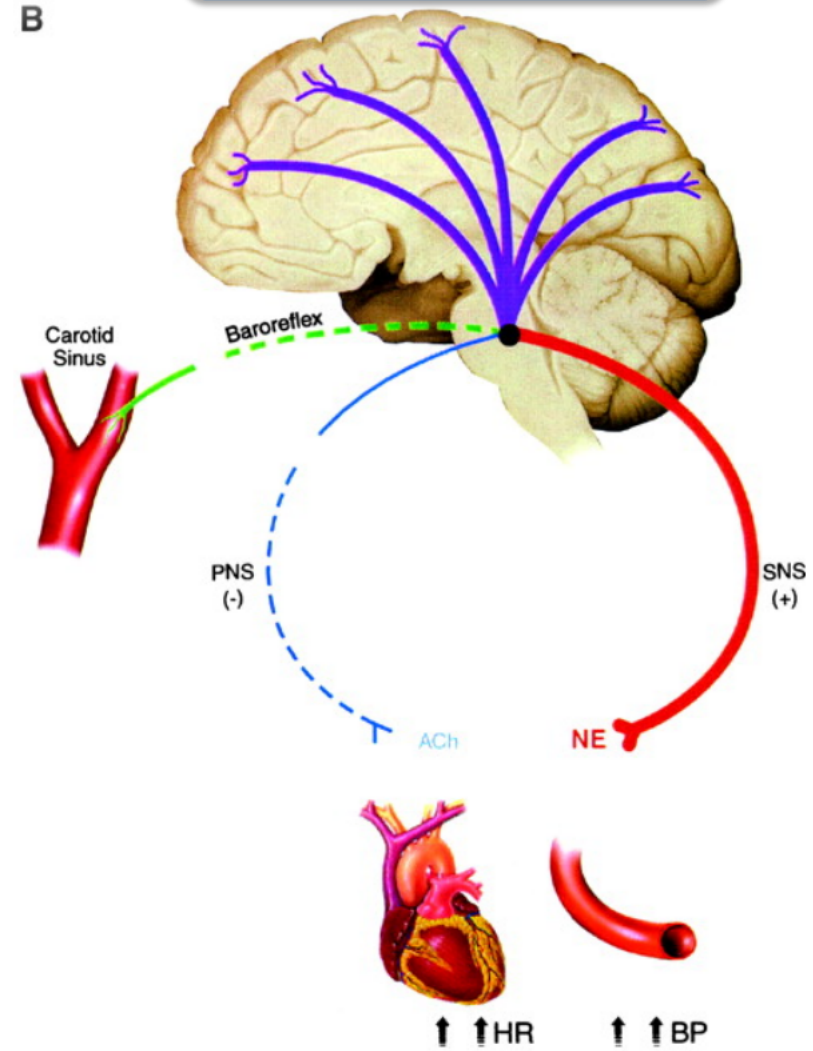
5. **Decrease BP and HR**

- RVLM: Rostral ventrolateral medulla (sympathetic activity origin)
- NTS: Nucleus tractus solitarii

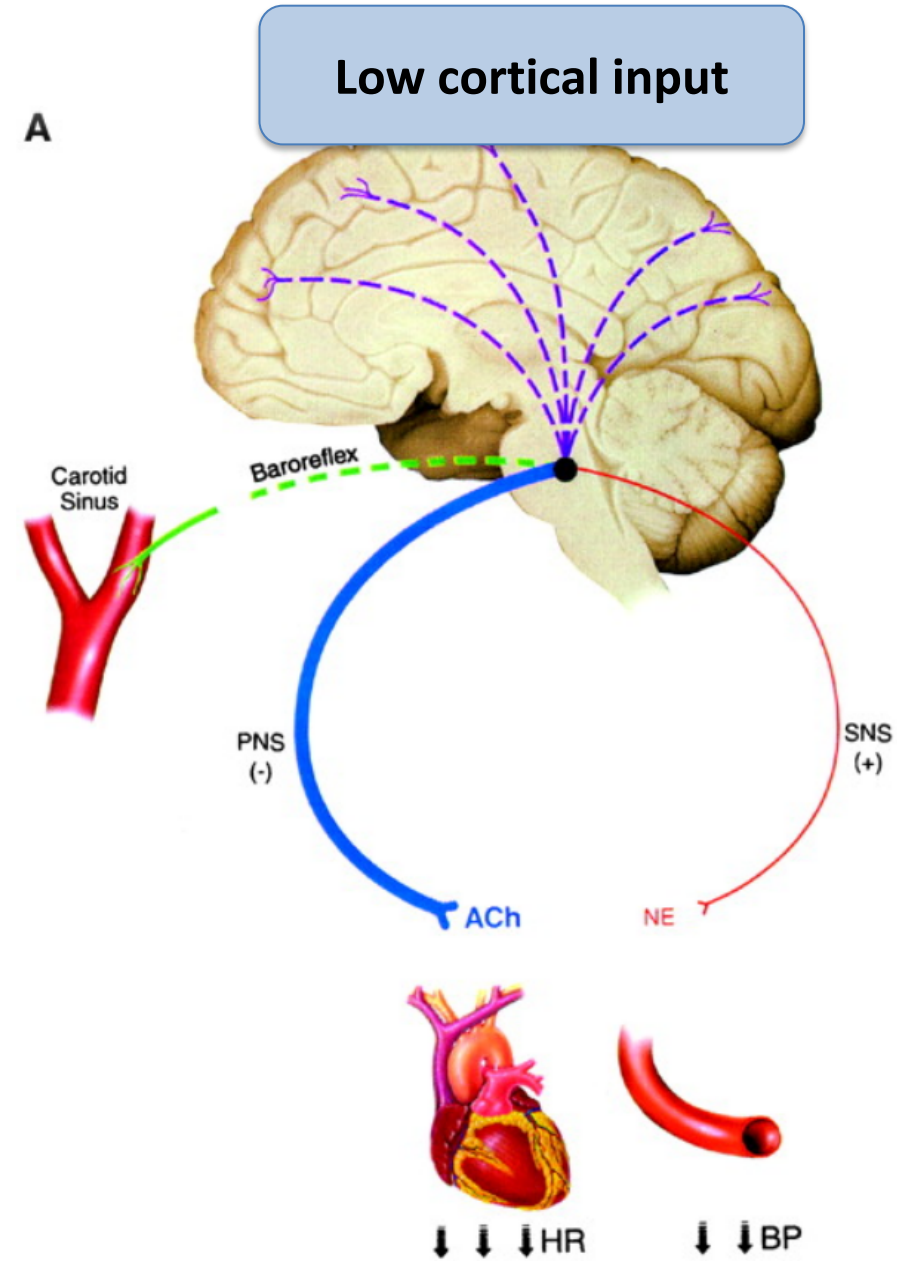
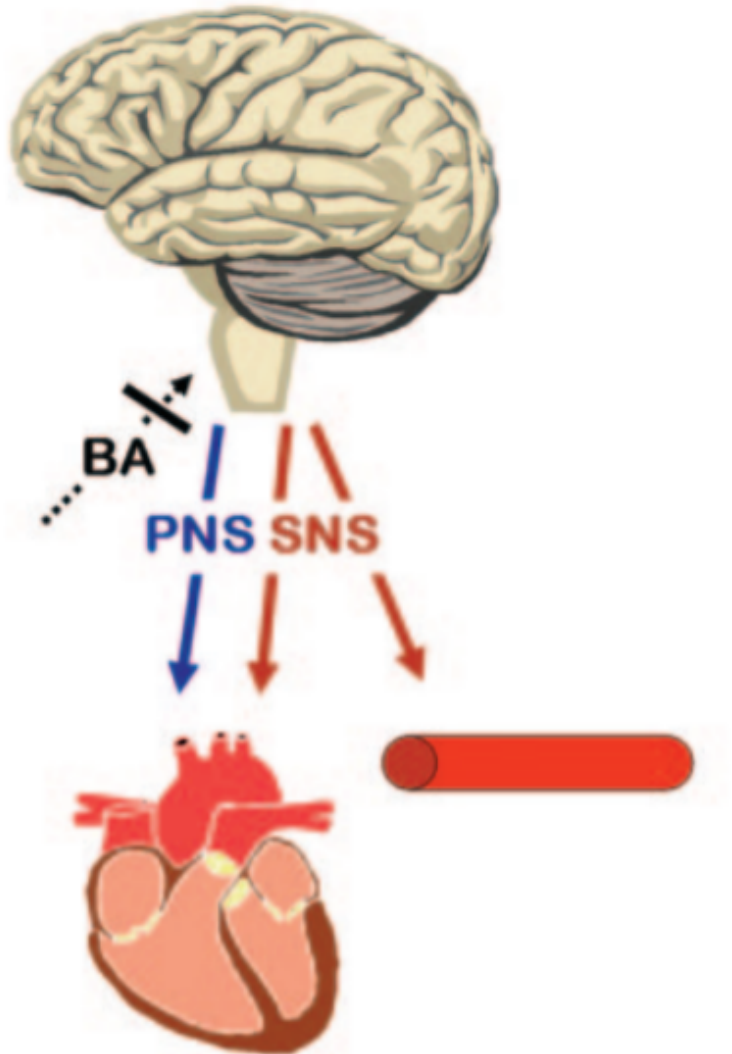
Nonselective Baroreflex Failure



High cortical input



Selective Baroreflex Failure



Hypertension. 2005;45:834-839.

Circulation. 2002 May 28;105(21):2518-23



Characteristics of Baroreflex Failure

- Hypertensive crisis --> Tracking of blood pressure and heart rate
- Volatile labile hypertension
- Alternating with episodes of hypotension--> severe malignant vagotonia
- Orthostatic tachycardia



Differential Diagnosis: Baroreflex Failure

Pheochromocytoma	Carcinoid
Syncope	Intracranial lesions
Paroxysmal tachycardia	Alcohol withdrawal
Orthostatic intolerance	Cerebral vasculitis
Pure autonomic failure	Page syndrome
Hyperthyroidism	Pseudopheochromocytoma
Renovascular hypertension	Migraine
Medications/drugs	Psychological disorders e.g. panic attack
Mast cell activation disorder	



Management: Baroreflex Failure

TABLE 1 General Guidelines for the Management of Baroreflex Failure

1. Accept the fact that **we will not normalize blood pressure.**
2. **For labile hypertension**
 - Do not “chase” blood pressure (i.e., wait for blood pressure to increase to treat individual hypertensive surges); do not use short-acting clonidine.
 - Use **longer-acting central sympatholytic drugs (methyldopa, guanfacine, clonidine patch)** at the lowest dose that prevents the biggest blood pressure surges; do not try to normalize blood pressure.
 - For breakthrough hypertension episodes, add clonidine, alpha blockers.
 - For hypertensive crises triggered by stress: consider biofeedback, benzodiazepines, cannabinoids.
 - For “background” essential hypertension: ARB, ACE inhibitors.
 - For cardioprotection: beta-blockers (added to above, not as initial treatment).
3. For **hypotensive episodes or orthostatic hypotension**
 - Oral water bolus, physical stimulation, abdominal binder.
 - Midodrine as needed.
 - **Fludrocortisone** in otherwise treatment-resistant cases (but may promote cardiovascular damage).
4. In severe cases: “clamp” blood pressure with high-dose central sympatholytic drugs and “rescue” midodrine

ACE = angiotensin converting enzyme; ARB = angiotensin receptor blockers.



Take Home Message

- Lateral neck mass with positive Fontaine's sign is highly suggestive of carotid body tumor
- The presence of carotid body tumor should raise suspicion of SDHD mutation
- Baroreflex failure should be considered when hypertensive episode develops after bilateral carotid body tumor resection