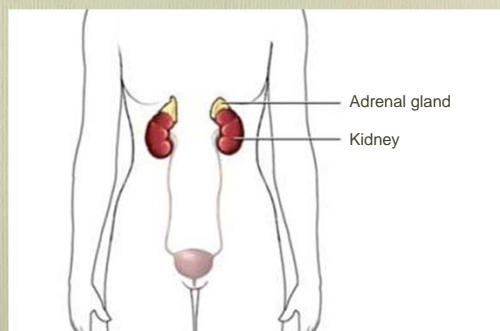


Von Hippel Lindau Syndrome and the Adrenal Glands

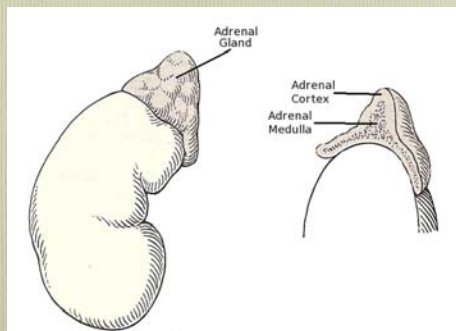
Nikolaos Stathatos, MD
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Harvard Medical School



Von Hippel Lindau Syndrome and the Adrenal Glands



Von Hippel Lindau Syndrome and the Adrenal Glands



Von Hippel Lindau Syndrome and the Adrenal Glands

Adrenal Cortex

- ◆ Glucocorticoids (cortisol: Stress hormone)
- ◆ Mineralocorticoids (aldosterone: Salt hormone)
- ◆ Sex steroids (minor)

Adrenal Medulla: Catecholamines

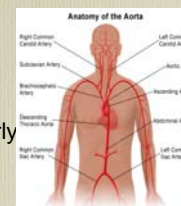
- ◆ Epinephrine (Adrenaline)
- ◆ Norepinephrine (Noradrenaline)

Von Hippel Lindau
+
Adrenal Glands
=
Pheochromocytoma

Pheo = "gray"
chromo = "color"
cyt = "cell"
oma = "tumor"

Von Hippel Lindau Syndrome Pheochromocytoma

- ◆ 10 – 20 % of VHL patients
- ◆ More common in type 2 VHL
- ◆ Mean age of diagnosis: 30 (as early)
- ◆ 5% are malignant
- ◆ Bilateral or extra-adrenal tumors also seen:
 - Neck (glomus jugulare, carotid body)
 - Around the aorta



Von Hippel Lindau Syndrome Pheochromocytoma

CLINICAL PRESENTATION

- ◆ Palpitations
- ◆ Headaches
- ◆ Episodic sweating
- ◆ Pallor
- ◆ Nausea

MOST COMMON PRESENTATION

Asymptomatic

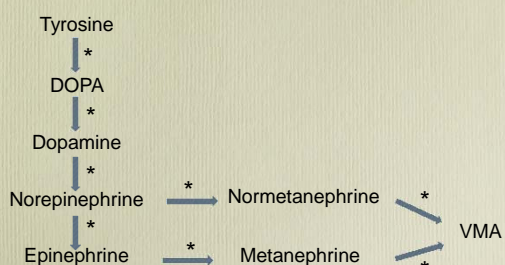
Von Hippel Lindau Syndrome Pheochromocytoma

DIAGNOSIS

Biochemical Diagnosis

- ◆ Blood test: Plasma Free Metanephrines
- ◆ Urine (24 hour collection)
 - Epinephrine
 - Norepinephrine
 - Dopamine
 - VMA
 - Metanephrine
 - Normetanephrine

Von Hippel Lindau Syndrome Pheochromocytoma



Von Hippel Lindau Syndrome Pheochromocytoma

DIAGNOSIS

Biochemical Diagnosis

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Von Hippel Lindau Syndrome Pheochromocytoma

DIAGNOSIS

Imaging Diagnosis

- ◆ CT Scan
- ◆ MRI
- ◆ Meta-IodoBenzylGuanidine (MIBG) Scan
- ◆ Octreotide Scan
- ◆ PET Scan
 - Glucose
 - DOPA

Von Hippel Lindau Syndrome Pheochromocytoma

TREATMENT

Management BEFORE surgery

- ◆ Block the effect of excess catecholamines
- ◆ Most commonly used agent: Phenoxybenzamine
- ◆ Treatment is adequate when the blood pressure is better controlled and orthostasis and/or stuffy nose develop

Von Hippel Lindau Syndrome Pheochromocytoma

TREATMENT

Surgical Management

- ◆ Adrenalectomy (laparoscopic), occasionally both sides
- ◆ For extra-adrenal tumors: depends on the location
- ◆ If both adrenals removed:

ADRENAL INSUFFICIENCY

Von Hippel Lindau Syndrome Adrenal Insufficiency

Acute Symptoms

- ◆ Unusual tiredness and weakness
- ◆ Dizziness when standing up
- ◆ Nausea, vomiting, diarrhea
- ◆ Loss of appetite
- ◆ Stomach ache
- ◆ Joint aches and pains
- ◆ **If untreated: Cardiovascular collapse and death**

Von Hippel Lindau Syndrome Adrenal Insufficiency

Chronic Symptoms

- ◆ Weight loss
- ◆ Darkened skin
- ◆ Craving for salt

Von Hippel Lindau Syndrome Adrenal Insufficiency

Treatment

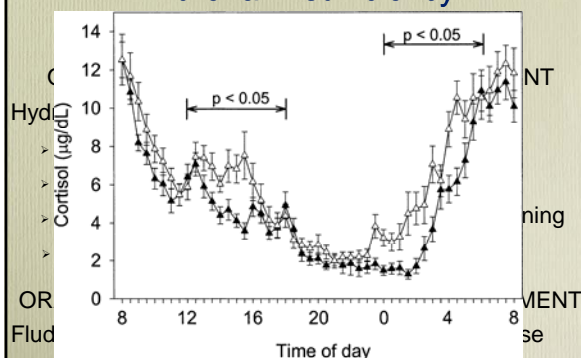
ORAL STEROIDS – CORTISOL REPLACEMENT

- ◆ Hydrocortisone
- ◆ Dexamethasone (1 mg = 20 mg of hydrocortisone)
- ◆ Prednisone (1 mg = 4-5 mg of hydrocortisone)

ORAL STEROIDS – ALDOSTERONE REPLACEMENT

- ◆ Fludrocortisone

Von Hippel Lindau Syndrome Adrenal Insufficiency



Von Hippel Lindau Syndrome Adrenal Insufficiency

Treatment

ORAL STEROIDS – CORTISOL REPLACEMENT

- ◆ No test can help decide what the correct dose is
- ◆ The person's weight is important
- ◆ The appropriate follow up is critical
- ◆ Good communication between patient and doctor

Von Hippel Lindau Syndrome Adrenal Insufficiency

Signs and Symptoms of Over-Treatment

- ◆ Weight gain
- ◆ Striae (stretch marks)
- ◆ "Moon face": rounding of the face ("Cushinoid" face)
- ◆ Hirsutism – unwanted hair growth
- ◆ Acne
- ◆ Hypertension
- ◆ High blood glucose

Von Hippel Lindau Syndrome Adrenal Insufficiency

Signs and Symptoms of Under-Treatment

- ◆ Unusual tiredness and weakness
- ◆ Dizziness when standing up
- ◆ Nausea, vomiting, diarrhea
- ◆ Loss of appetite
- ◆ Stomach ache
- ◆ Weight loss
- ◆ Darkened skin
- ◆ Craving for salt
- ◆ Joint aches and pains

Von Hippel Lindau Syndrome Adrenal Insufficiency

Treatment

ORAL STEROIDS – CORTISOL REPLACEMENT

- ◆ The human body does not need the same amount of cortisol every day !!!
- ◆ Cortisol is the "stress" hormone: when stressed (e.g. sickness, trauma), the body needs more
- ◆ The patient needs to know both WHEN and HOW to adjust their dose when sick or stressed

Von Hippel Lindau Syndrome Adrenal Insufficiency

Sick Day Rules

- ◆ Minor Illness: take 10 mg **hydrocortisone** three times a day in addition to your usual daily dose of glucocorticoid and fludrocortisone
- ◆ More severe illness: take 20 mg **hydrocortisone** three times a day in addition to your usual daily dose of glucocorticoid and fludrocortisone.
- ◆ When in doubt about whether or not to take more hydrocortisone, take more hydrocortisone.

Von Hippel Lindau Syndrome Adrenal Insufficiency

Treatment

STERIODS BY INJECTION INTO A MUSCLE

- ◆ If too sick to take pills (e.g. vomiting)
- ◆ Call your doctor or go to the Emergency Room !!!!

Von Hippel Lindau Syndrome Adrenal Insufficiency



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