Patient case

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01/18/12

History of present illness

- 29 year old male- endocrinology consult due to low testosterone level
- Patient was discovered to have low testosterone after adrenal schwannoma was removed (schwannoma-benign nerve sheath tumor composed of Schwann cells, which normally produce the insulating myelin sheath covering peripheral nerves)
- At the same time he complained about joint pains and decreased libido

Past history

- US – intraabdominal mass (2008)- schwannoma,
- Initially it was believed to be gastrointestinal tumor, he had right kidney and adrenal removed
- Crohn disease (2008)

Family history

- Mother- thyroid problems
- Father- lung cancer
- One brother
- Two children who are doing well

Social history

- Marital status- single
- Tobacco use- no
- Alcohol- no
- Drug use- no
- Trauma- no
- Sexually active- female partner

Vitals and physical examination

- BP 108/76 mmHg / pulse 60 / Temp 37 C/ resp 12/ Wt 93.41 kg
- Spine- without local point tenderness on palpitation, absent thoracic kyphosis
- Neck- supple without any palpable lymph nodes, or thyromegaly
- Chest- clear without any splits, murmurs, or gallops
- Abdomen- soft, no stria
- Normal axilary, pubic hair
- Testes: 18ml R and 19 ml L, no mass

Laboratoty data

- Morning testosterone 262ng/dl (300-1000)
- Luteinizing hormone (LH) 1.88 U/L (1.84-8.16)
- Morning cortisol, ACTH, TSH - normal

- The main hypotheses for the diagnosis ?
- The cause of hypogonadism?

Assessment and plan

- Check prolactin levels to rule out prolactinoma
- Pituitary imaging
- Consider screening for hemochromatosis

- Prolactin 11.7ng/ml (1-20)
- The pituitary MRI, with and without contrast, showed a normal pituitary gland
- Ferritin 636ng/ml (18-350ng/ml)
- Transferrin saturation 65% (<45%)

Hematologist and genotypic testing 06/10

- The patient was assessed for mutation in the HFE gene and was found to be homozygous for the C282Y mutation
- Diagnosis: Hereditary hemochromatosis. Secondary hypogonadysm.

Treatment

- He was started on monthly phlebotomies (500 ml per month) and testosterone replacement (Testosterone undecanoate 250mg/ml-4ml i/m every 3 month)
- After nine phlebotomies, testosterone replacement was discontinued and the patient remained off of testosterone replacement for 8 weeks. After this treatment, the morning testosterone was 246ng/dl (300-1000), LH 2.3 IU/L (1.84-8.16) and ferritin 95ng/ml (18-350ng/ml) , Hb 13 g/l, HCT 40 %.

Clinical practice guidelines

- One phlebotomy (removal of 500 mL of blood) weekly or biweekly
- Check hematocrit prior to each phlebotomy; allow hematocrit to fall by no more than 20% of prior level
- Check serum ferritin level every 10-12 phlebotomies
- Stop frequent phlebotomy when serum ferritin falls below 50 ng/mL
- Continue phlebotomy at intervals to keep serum ferritin to between 25 and 50 ng/mL
- Avoid vitamin C supplements

Algorithm for management of HH (AASL)