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Images of Endocrine Disorders

M2 - Endocrinology Sequence
A. Kumagai & T. Giordano

NOTE: BEST VIEWED AS SLIDE SHOW

Winter 2009
Images of Endocrine Disorders

Because endocrine disorders are manifested by unusual and varied clinical findings, diagnosis often involves developing a “gestalt” -- an overall picture of the individual with a possible endocrine disorder -- and matching of this picture with characteristic clinical features.

The purpose of the following photographs is to give you a “gallery” of these characteristic features to help you to remember elements of several different disease processes. The material is meant to reinforce information provided in lectures, handouts and discussion groups (and hopefully to have some fun while doing it.) Please refer to required materials for details.
Endocrine Images: Acromegaly

Picture of wrestling star Andre the Giant and Skull X-ray of man with acromegaly. Notice the characteristic prominent supraorbital ridge (“frontal bossing”), large jaw, and dental malocclusion with underbite (x-ray).
Individual with acromegaly photographed over a 37-year span. Ages in years are in lower left corner of each photograph.

Note that the changes occurring with acromegaly may be very gradual and go completely undetected by the patient or his or her family for many years. It is often only thorough the comparison with old photographs or complaints involving complications of acromegaly, such as sleep apnea, diabetes or dental problems that acromegaly is suspected.
Endocrine Images: Acromegaly

Hands of individual with acromegaly (left) compared to hand of non-acromegalic adult (far right).
Foot X-ray of Patient with Acromegaly.
Notice the unusually thick “pad” of soft tissue overlying the calcaneus (double arrow). It is said that a good clinical sign of acromegaly is the inability to feel the calcaneus when pressing on the heel.
Endocrine Images: Acromegaly

Clinical Findings in Acromegaly.

Symptoms & Signs:
• Excessive sweating, snoring.
• Arthalgias, carpal tunnel syndrome.
• Change in ring/glove or shoe size.

Signs:
• Dental malocclusion and widely spaced teeth.
• Macroglossia.
• Large hands and feet.
• Large heart (may see signs of heart failure).

Laboratory results:
• Impaired glucose tolerance or diabetes.
• Elevated IGF-1.
• Enlarged cardiac silhouette on chest x-ray.
Graves Ophthalmopathy (Exophthalmos).

Graves ophthalmopathy is due to autoimmune-mediated inflammation and edema of the extraocular muscles. Graves eye disease may be asymmetrical and often progresses independently of hyperthyroidism and may lead to diplopia, corneal dryness, ulceration, and blindness. Severe cases may require surgical decompression. Exophthalmos is specific to Graves disease. On the other hand, “lid lag,” in which the eyelids do not closely follow downward gaze, may be seen in all forms of hyperthyroidism and is due to hyperstimulation of the orbicularis oculi muscles.
Endocrine Images: Graves Disease

This photo was taken from Dr. Koenig’s thyroid lecture and is meant to highlight the eye findings in Graves disease: the classic “stare” of hyperthyroidism and a prominent goiter. Notice in Graves that the thyroid is symmetrically enlarged and “plump.” This is because the entire thyroid is being stimulated by thyroid stimulating immunoglobulin (TSI), which causes constitutive activation of the TSH receptor in the absence of TSH. Auscultation of the goiter of an individual with active Graves disease may reveal a thyroid bruit, due to the hypervascularity of the overactive gland. This bruit must be distinguished from cardiac (or carotid) bruits by localizing its source over the thyroid.
Graves dermopathy is also known as “pretibial myxedema,” which is an unfortunate term, since “myxedema” usually refers to hypothyroidism. The term “myxedema” describes the “doughy” or “peau d’orange” texture of the skin. Graves dermopathy involves inflammation and mucopolysaccharide deposition most prominently in the pretibial regions of the legs. It is a relatively uncommon--albeit, classic--finding in Graves disease and affects approximately 5% of patients with Graves.
Endocrine Images: Graves Disease

Clinical Findings in Graves Disease.

Symptoms & Signs:
• Heat intolerance, excessive sweating.
• Anxiety, “hyperkinesis.”
• Sleep disturbances.
• Weight loss *despite* increased appetite.
• Hyperdefecation (not diarrhea).

Signs:
• Tachycardia, wide pulse pressure.
• Warm, moist skin.
• Exophthalmos may be present.
• Symmetrical, “plump” goiter.
• Fine tremor of outstretched hands.
• Brisk reflexes.
Endocrine Images: Hypothyroidism

This pair of photographs illustrates some general physical features of congenital hypothyroidism and severe hypothyroidism in an adult. The face is has a puffy, “doughy” appearance (hence, the term “myxedematous”). Periorbital edema may be present. The skin is dry and cool, and the hair is coarse. The affect is blunted and apathetic. The child is short and has mental retardation.
Clinical Findings in Hypothyroidism.

Symptoms & Signs:
- Depression.
- Cold intolerance.
- Weight gain *despite* unchanged appetite.
- Constipation.

Signs:
- Bradycardia, diastolic hypertension.
- “Myxedematous facies” with coarse hair.
- Distant heart sounds.
- Delayed relaxation phase of achilles reflex.

Laboratory results:
- Anemia: either macrocytic or normocytic.
- Hyponatremia (due to decreased free water clearance by the kidney).
- Elevated TSH, low free T4 (primary hypothyroidism). [Note: since free T3 may remain normal until hypothyroidism is severe it is useless in the diagnosis of hypothyroidism.]
Prominent physical findings in Cushings syndrome include round “moon facies,” supraclavicular and supracervical fat pads (“buffalo hump”), central obesity and purple abdominal striae. If the result of a pituitary adenoma (Cushings Disease), hyperpigmentation may be present. If an adrenal cortical carcinoma is the cause, there may be hirsuitism and virulization. (Adrenal carcinomas may produce DHEA sulfate, a potent adrenal androgen.) Adrenal carcinomas also grow more rapidly than adrenal adenomas and tend to be larger: almost always > 5 cm in diameter on an abdominal CT scan.
Abdominal Striae in Cushings Syndrome.

Classically, these striae are purplish in color and appear on the abdomen, thighs, upper arms and axillae. They are distinguished from silver striae seen in postpartum women or pink striae seen with significant weight loss.

Excessive steroid action on skin also may lead to skin fragility and easy bruising during routine activities.
This slide of identical twins is from Dr. Hammer’s lecture and is meant to emphasize the hyperpigmentation and thin body habitus that is often seen in primary adrenal insufficiency (the woman with adrenal insufficiency is on the right). Hyperpigmentation may also be seen in the extensor surfaces of the limbs (knuckles, elbows, knees), in newly formed scars and in palmar creases and buccal mucosa. (What’s the cause?)
Clinical Findings in Addison’s Disease.

Symptoms & Signs:
• General malaise, fatigue.
• Weakness and difficulty climbing stairs, arising from sitting, combing or shampooing hair.
• Salt craving.

Signs:
• Orthostatic hypotension.
• Hyperpigmentation of extensor surfaces of skin, buccal mucosa, palmar creases.
• Weakness of proximal muscle groups.

Pertinent routine laboratory results:
• Normocytic anemia
• Neutropenia (mild) with eosinophilia.
• Hyponatremia, hypokalemia and “non-gap” metabolic acidosis.
• Mild hypoglycemia (may be pronounced in infants).
Endocrine Images: Addison’s Disease

Hyperpigmentation in Addison’s Disease.

In primary Addison’s disease, one often sees hyperpigmentation of extensor surfaces of the limbs (knuckles, elbows, knees), of the areolae of the breasts, of newly formed scars, and of the buccal mucosa. In this photograph, one may see darkening of the face, fingertips and gingiva as well. (What’s the mechanism?)
Hyperpigmentation in Addison’s Disease.

This is a (presumably) postmortem drawing from Addison’s original paper of an individual with primary adrenal insufficiency. In Addison’s day, the primary cause was not autoimmune adrenalitis, but tuberculosis.
The great British novelist Jane Austin also suffered from Addison’s disease and died prematurely of its complications. If you look closely, you can see areas of hyperpigmentation on her cheeks...but again, this might be the product of an over-worked endocrinologist’s imagination...
Endocrine Images: Lipodystrophy in Diabetes

Lipodystrophy (lipohypertrophy and lipoatrophy) may be seen as a side effect of insulin therapy.

- Lipohypertrophy is common and occurs in areas of frequent insulin injections. It is caused by hyperplasia and hypertrophy of subcutaneous fat from prolonged exposure to high insulin concentrations and may affect insulin absorption and action. Lipohypertrophy may be seen in individuals with type 1 or type 2 diabetes.

- Lipoatrophy is much less common (especially with the current use of recombinant human insulins) and is seen in individuals with type 1 diabetes. Its cause appears to be an autoimmune reaction and is characterized by complement deposition in subcutaneous tissue.

- Generalized lipoatrophic syndromes are rare and may occur independently from insulin-treated diabetes.
Endocrinologists spend a great deal of time looking at people’s feet. Foot problems are very common in diabetes and may result from diabetic sensory neuropathy, a lack of normal sweat response and dry feet (from compromised sweat gland innervation), and poor circulation from peripheral vascular disease. Sensory neuropathy leading to permanently “numb” or “insensate” feet predispose the feet to unrecognized trauma and abnormal weight-bearing. This in turn may lead to cracks, fissures, and ulcers or the development of so-called “Charcot Feet,” which involve bony changes and gradual, severe skeletal deformation. Ulcers may be very slow to heal and if deep, may lead to osteomyelitis, cellulitis and gangrene.

Remember: Diabetes is the leading cause of non-traumatic lower extremity amputation in the United States.
The term “Charcot feet” used to describe bony deformation in diabetes comes from the great French neurologist (and Sigmund Freud’s former professor) Dr. Jean-Martin Charcot of the Hôtel Dieu Hospital in Paris. Charcot originally described similar lower extremity changes in another disease…tabes dorsalis associated with neurosyphilis….

In addition to a 128-Hz tuning fork, a 10-gram monofilament (similar to a straight piece of fishing line) is frequently used to assess the severity of diabetic peripheral neuropathy. Failure of the patient to feel the touch of the filament is associated with a significantly increased risk of developing foot ulcers.

This instrument was actually originally developed to assess the peripheral neuropathy seen in another disease…“Hansen’s Disease” or leprosy….
Necrobiosis lipodica diabetorum.
Indurated, nontender ulcerations of the lower extremities occasionally seen in individuals with type 1 diabetes.
A Gallery of Endocrine Images: Famous Names in Endocrinology
Famous Names in Endocrinology

Acromegaly

• Robert Wadlow, the “Alton Giant” 1918-1940

Robert Wadlow, the “Alton Giant” is said to be the tallest human in history, stood at 8’ 11 ½” and died at age 22 from an infected leg ulcer. He was very spiritual, was a Boy Scout, and briefly attended college until his death.

• Lurch of the Original Addams Family

• Andre the Giant

• “Jaws” of the James Bond Movie Fame
Endocrine Images: Addison’s Disease


In the first-ever televised presidential debate, John F. Kennedy was the apparent winner over Richard M. Nixon, a win which helped him in his narrow victory over Nixon in the presidential election of November, 1960. Many observers attributed Kennedy’s “telegenic” character to his youthful, dynamic, tanned (i.e., “hyperpigmented”) appearance...
Endocrine Disorders and World History


Endocrine disease has clearly affected the course of world history. Kennedy’s year-around “tan” (from his Addison’s disease) helped him win the presidency and lent a youthful air to “Camelot,” as the Kennedy White House was known, whereas Tito’s death from complications from type 2 diabetes in 1980 eventually led to the break-up of the Yugoslavian federation and the bloody Balkan wars and “ethnic cleansing” of the 1990’s.
Famous Names in Endocrinology

Graves Disease

Marty Feldman, star of a series of classic movies by Mel Brooks had Graves disease. Hint: Feldman is the guy on the right. The man on the left is Gene Wilder. He has no endocrine disorder but is just pretty wacky...
Famous Names in Endocrinology

Graves Disease

Both Bushes were diagnosed with Graves disease: Barbara had Graves ophthalmopathy and George presented with atrial fibrillation. Their dog, Millie, had lupus. No kidding. Must be the water...
Addison’s Disease

We now all know about John F. Kennedy, but the novelist Jane Austen was also afflicted with adrenal insufficiency. Her Addison’s disease worsened as she grew older, and she finally succumbed to it at the age of 41 in Winchester, in Central Hamshire (UK) in 1817.
Famous Names in Endocrinology

The beleaguered looking beagle is one of several dogs whose pancreatectomies were instrumental in experiments leading to the discovery of insulin in 1922.

Type 1 Diabetes

The beleaguered looking beagle is one of several dogs whose pancreatectomies were instrumental in experiments leading to the discovery of insulin in 1922.
Famous Names in Endocrinology

• Mary Tyler Moore
• 1999 Miss America Nicole Johnson
• 1996 Atlanta Olympic Gold Medalist Gary Hall, Jr.

Type 1 Diabetes: Recent

Both actress Mary Tyler Moore and the 1999 Miss America, Nicole Johnson, have type 1 diabetes. Both women have done much to publicize the issue of diabetes awareness in general, and of type 1 diabetes in particular. Gary Hall, Jr. won gold and silver at the 1996 Summer Olympic Games in Atlanta.
Famous Names in Endocrinology

Type 1 Diabetes: Before 1922

A “trick” question, since before the invention of insulin, no one with type 1 diabetes lived long enough to become famous.....
We now all know about B.B. King, but the jazz great Ella Fitzgerald also had type 2 diabetes. Ms. Fitzgerald suffered from neuropathy and peripheral vascular disease, which necessitated lower extremity amputation prior to her death…We miss you Ella...