The Art of Physical Examination: Is It Dying? Is it dead?

- Skill and familiarity with certain bedside maneuvers and confidence in eliciting physical signs declining
- Increased dependence on aid of radiologist or first-tier laboratory data
- Skills in getting data back and arranging for tests to be done are valued as much or more than eliciting physical findings
- Emphasis on data illustrated by "rounds" at computer, PDA, not at bedside
In the electronic era of EMRs, e-journals, and epocrates, we must remember that our patients are people, not “e-patients.”
Lawson Wilkins: the Man

- Born in 1894 to a general practitioner who worked until his death at 86 yo
- Graduated from Hopkins medical school while serving as orderly in France during WW I
- Internal medicine internship at Yale but switched to pediatrics at Harriet Lane because pediatrics was the leading branch of medicine in biochemical & metabolic approach to disease
- Practiced general peds full-time from 1922-1946, working till 8-10 PM and still doing research

Lawson, the Academic Clinician

- Research as General pediatrician included:
  - Ca and P metabolism in rickets
  - Clinical study on effect of cod liver oil in curing rickets
- Joined epilepsy clinic at Hopkins at request of Edwards Park and wrote results of studies on ketogenic diet which, Dr Park stated:
  "showed the care with which Lawson investigated the records and particularly the soundness of his conclusions. The detachment and common sense with which he approached his problems were particularly impressive"

Wilkins Medical Career

- 1922: Entered private practice
- 1935: “It was Dr Edwards Park who shepherded me back to the academic fold after 25 years in private practice- ... when he asked me to establish a pediatric endocrine clinic at the Harriet Lane Home
  - “I knew nothing about the subject and replied, 'Do you wish to make me a charlatan?'”
- 1945: Gave up private practice to work full time at Hopkins
Wilkins the Scientist

“Science”: From Latin “Sciro” To Know

“Wilkins was a skeptic of established doctrine and he was appalled to find that the literature on endocrine disease in children consisted of anecdotal reports and formulations that had little scientific basis. He, therefore, resolved to approach each endocrine disorder from a clean slate by critically seeking objective information on its pathophysiological basis." Jud Van Wyk

“His stock in trade was to plot and display graphically every objective physical finding or measurement on his patients as a function of time and treatment.”
Wilkins the Scientist

In accepting the Howland Award

... It is only the clinician...who can seek out and bring to attention the human “experiments of nature” which may present the opening key to basic problems....

No one can reproduce in the laboratory the inborn enzymatic defects which cause virilizing adrenal hyperplasia or cretinism; it was the study of patients with these disorders that led to the understanding of the steps in the normal hormonogenesis of the adrenal and thyroid glands.”
The clinical investigator “must have curiosity. If he has such curiosity, nearly every patient whom he encounters, no matter how simple and clear cut his disease may be, will call forth many, many questions of real importance which have never been answered... the clinical investigator will feel impelled to answer some of these questions by studies upon the patient.”
Taught auxology
- Upper: Lower segment
- Body proportions (e.g., Arm span)
- Sitting heights
- Dental eruption
- Carried a 6 inch ruler, which he called a phallometer
Figure 2. The figures of normal boys two and eight years respectively illustrate the change in the ratio of upper and lower skeletal segments, measured from the symphysis pubis. At birth the ratio is 1.7/1.0 and at ten years 1.0/1.0.

The hypothyroid dwarf having the height of a two year old retains the infantile proportions of two years. Dwarfs of pituitary or primordial types, however, attain the more mature proportions of their chronological age. Wilkins, L. and Fleischmann, W.: J.A.M.A. 116:(May 31), 1941.
Wilkins Laboratory was the Clinic

- Longitudinal assessment of measurements and development of growth curves for different disease states
  - Hypothyroidism
  - Hypopituitarism
  - Delayed maturation
  - Primoridal dwarfism

Guiding Principle:
Maximally use observation to evaluate patients and use lab wisely
Figure 1B. Three cases of retarded growth and development showing response to therapy. Compare types of curves with those shown on chart above.
State of the Discipline

Inferential Evaluation of Hypopituitarism
- Could not measure GH levels
- Diagnosis of hypopituitarism made on basis of growth retardation in association with:
  - Sexual infantilism in adolescents
  - PBI (protein bound iodine)
  - Insulin sensitivity
  - Inability to secrete a water load

Inferential Evaluation of Hypothyroidism
- Could not measure thyroid hormone or TSH levels
- Change in cholesterol concentrations defined adequacy of treatment
There are a number of disorders in which endocrine symptoms are due to irresponsiveness of the tissues ("end organs") to normal amounts of hormone. Examples are nephrogenic diabetes insipidus, pseudohypoparathyroidism, and the inability of some male pseudohermaphrodites with feminizing testes to develop sexual hair even when androgen is administered.

These are probably due to genetically determined defects of intracellular enzyme systems.”
SEXUAL PRECOCITY—"ALBRIGHT'S SYNDROME"

Syndrome of Polyostotic Fibrous Dysplasia, Skin Pigmentation and Precocity (in Females)

Note density of base obscuring sella

Normal 9 years  Patient (H.L.H. A 14901) age 9 years

Localized lesion in femur
Precocious Puberty

- Birth normal except for extensive unilateral pigmentation over chest, arm and face
- Normal until 3 years old when rapid growth began
- Developed breasts at 6 years, menses at 7 years
- Thyroid enlargement, hypertension noted at 8 years
- When seen at 9 years, she had a height age of 15 years and bone age 14-15 years. Dental age was 10-12 years
- “Adolescent” breasts, well-developed pubic hair, estrinization of vaginal mucosa
Precocious Puberty

Skin had dark brown flat patches on right side of trunk stopping at midline, right arm and forehead

Bone had area of rarefaction (fibrous dysplasia) right femur

Thyroid enlarged with no bruit. High strung, overactive but no other symptoms of hyperthyroidism

Pulse 100-120, BP 136/88 Cholesterol 155 mg/dL

Urinary FSH < 40 RU/24 hrs
Wilkins' Comments

- **Cause of sexual precocity, which occurs almost exclusively in females, is uncertain.**

- **Albright believes it is due to a congenital abnormality of the hypothalamus.**

- **Skeletal overgrowth and thyroid enlargement sometimes occur.**
Wilkins the Endocrinologist

“At first he concentrated on the study of children suffering from thyroid deficiency...and he used to take me into his laboratory to show me the exquisite graphic studies of his patients.

The graphs, he told me, he worked out between midnight and 2 AM.”

-Edwards Park
Photograph illustrates the characteristic *infantile proportions of skeletal segments* (measured from the symphysis pubis) and the *lack* of naso-orbital development of the cretin. The other dwarf has proportions and features normal for his age.
Note the accelerated growth, osseous development and muscular development. The penis, prostate and sexual hair develop markedly but the testes usually remain small and immature.
Unsuccessful attempts ... to suppress the secretion of androgen ... by the administration of steroids...which have a chemical structure similar to that of androgens.

Treatment of patients with cortisone ...was begun in January 1950 and preliminary reports... have been published.

This paper presents in more detail the results we have observed ... (in) 6 females and 2 males with congenital adrenal hyperplasia and includes the findings of other observers in 2 additional cases.
15 year old female observed for 11 years for virilization

1941 at age 6-1/2 years had left adrenal and part of right adrenal removed with no decrease in virilization

Urinary 17 KS measured daily for 10 days, then patient treated with IM cortisone X 15 days

After cortisone stopped, the girl had continued measurement of adrenal urinary metabolites

RESULTS: Cortisone decreased 17KS. Discontinuation of cortisone resulted in creased 17KS after 10 days

Figure 13. Males with Congenital Adrenal Hyperplasia
Macrogenitosomia Precox

Note the accelerated growth, osseous development and muscular development. The penis, prostate and sexual hair develop markedly but the testes usually remain small and immature.
Figure 16. Results of Cortisone Therapy in Congenital Adrenal Hyperplasia When Treatment Is Begun in Infancy
Prevention of Virilization; Maintenance of Normal Growth and Development

Age: 13 mos. 18 mos. 32 mos. 20 mos. 32 mos.
Height age: 12 mos. 17 mos. 20 mos. 16 mos. 36 mos.
Bone age: 22 mos. 36 mos. 30 mos. 18 mos. 34 mos.
Treated at: 3 mos. 2½ mos. 2½ mos. 3 mos. 22 mos.
Conclusions

- Cortisone decreased 17 KS and biologically active androgens in all patients

- The rapidity of the decrease depended on the dose
  - 50 mg resulted in max decrease in 4-5 days
  - 25 mg had maximum decrease in 10 days

- After discontinuation of cortisone 17 KS began to increase at 10 days and returned to pre-treatment levels by 3 weeks

- More experience is needed to determine optimal dose

- Clinical effect, not steroid excretion should be used to determine treatment efficacy
In salt-wasters, cortisone probably has salt-retaining activity

- Not known if DOCA also needed

- Estrogen effect occurred post-treatment and cortisone may prevent virilization and permit normal feminization in female pseudohermaphrodites

- Acne and hyperpigmentation decreased with cortisone treatment

- No change in carbohydrate metabolism
Clinical Recommendations

- Should give the minimum amount of cortisone needed to maintain 17 KS excretion <8 mg/day
  - Higher values associated with recurrence of androgen effects
  - Need to individualize

- Cortisone 25 mg causes growth retardation in infants
  - Cortisone 5 mg didn’t suppress growth
  - High dose cortisone doesn’t suppress growth in older patients

- Follow patients with
  - Growth velocity
  - Bone age
  - Urinary 17 KS
Patient was a hairless woman with testes and breasts
Surgery revealed
- No uterus
- Testes lined by Sertoli cells, nests of hypertrophied Leydig cells, no germinal cells

17 KS 15-20 pre castration, 7-10 mg/day post castration
Ques: Were these androgens biologically active?
- Urine sent to Dorfman's lab and the androgens were biologically active

Estrogen was elevated pre-castration and non-measurable post-castration
- Thus, estrogen came from testes

Patient did not respond to testosterone
- Testosterone was rubbed on mons pubis for 1 month but no hair grew
- Methyltestosterone 50 mg daily given for 1 month did not increase clitoral size, hair growth or result in voice change

Conclusion: Patient had resistance to androgens
Case

An 8-1/2 year old female was given cortisone 100 mg daily for 6 days followed by 25 mg daily to quickly and more effectively suppress 17 KS because the previous subject didn’t have adequate (acute) suppression when begun on 25 mg daily. Breasts developed and vaginal smear showed estrogen effect 1 month after cortisone treatment was initiated.
Clinical Features of Gonadal Dysgenesis

- Defined short stature and major and minor congenital malformations
- Auxologic measurements codified
  - Short stature
  - Lymphedema
  - Hands, feet
  - Webbed neck

- Streak ovary was different than a newborn ovary

- One of the first to describe the pathology of gonadal dysgenesis
- Gonads never developed beyond the stage of genital ridge

Wilkins L, Fleischman W. 1944
Ovarian Agenesis: pathology, associated clinical symptoms and the bearing on the theories of sex differentiation. J Clin Endocrinol 4:357-75
Emulating his mentor, Lawson Wilkins, David Smith was instrumental in developing the new field of dysmorphology, and his pioneering reference book of 1970 is now in its 6th edition, the continued effort of his trainees.
Lawson the Mentor

“A teacher affects eternity; he can never tell where his influence stops”

The Education of Henry Adams
Lawson’s studies have been based on the most meticulous investigations of his patients, not of the literature...

All data collected are represented visually on running charts which show their qualitative, quantitative, and temporal relationships.

They are accompanied by timed photographs of the patients themselves.

The whole produces a statistical record, a visual library of endocrinology.

His years of practice have “caused him to approach the problems of his patients primarily as a physician, secondarily as a scientist”

"Observe, record, tabulate, communicate. Use your five senses....Learn to see, learn to hear, learn to feel, learn to smell, and know that by practice alone you can become expert."

- Sir William Osler
The Art of Physical Examination: Where We Are

Advances in laboratory and imaging technology, along with the pace of modern medicine, have resulted in the physical examination being abbreviated, undervalued, and viewed as redundant.

Commentary by George R. Thompson III, MD, and Abraham Verghese, MD. Physical Diagnosis: A Lost Art? 2006; AHRQ M&M on-line
REMEMBER THE PATIENT!
"The diagnostic computer says that you have the flu, you're 27 pounds overweight, and that you'll probably vote Republican in November."