

Newborn screening and follow-up of children with endocrine disorders: current successes and challenges

David Allen, MD
Professor, Division Chief, and Fellowship Program Director
Department of Pediatrics
University of Wisconsin School of Medicine and Public Health



Wisconsin Newborn Screening (NBS) Panel

Hemoglobinopathies (5)

- Beta Thalassemia Major
- Hemoglobin S-Beta Thalassemia
- Hemoglobin SC Disease
- Hemoglobin E-Beta Thalassemia
- Sickle Cell Disease

Organic Acidemia (12)

Example: Proponic Acidemia

Other Disorders (5)

- Biotinidase Deficiency
- Galactosemia
- Cystic Fibrosis
- Severe Combined Immune Deficiency (SCID)
- Spinal Muscle Atrophy (SMA)

Fatty Acid Oxidation Disorders (12)

Medium Chain Acyl-CoA Dehydogenase Deficiency (MCAD)

Amino Acid Disorders (11)

Example: Phenylketonuria (PKU)

Endocrine Disorders (2)

- Congenital Adrenal Hyperplasia
- Congenital Hypothyroidism

Wisconsin Newborn SCREENING PROGRAM

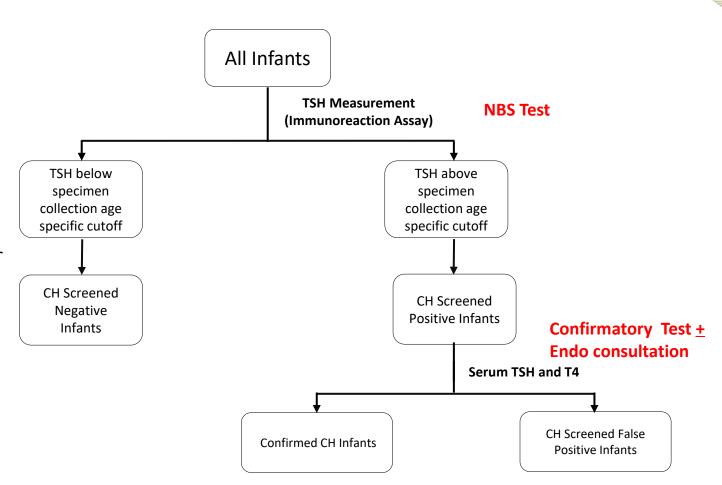
Babies are covered by the umbrella of Wisconsin's Newborn Screening Program

NBS for CH in Wisconsin



Congenital hypothyroidism

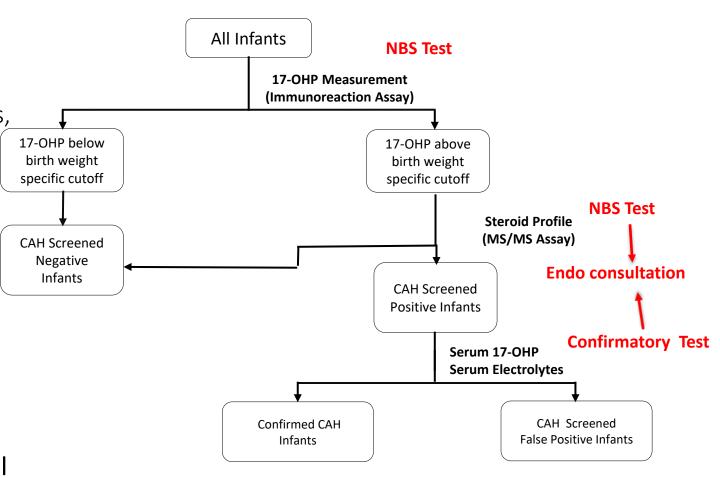
- ~ 1:2000 live births (~35/year WI)
- Most common preventable cause of cognitive disability
- NBS TSH predominant
- Continuum of severity –
 permanence established by 3y
- Therapeutic objective FT4 upper normal, TSH lower normal
- Follow-up
 - Lab q3-6 months
 - Frequent dose adjustment
 - Ped Endo visit 1-2 x/year
 - Telemedicine adaptable



NBS for CAH in Wisconsin

Congenital Adrenal Hyperplasia (210HD +)

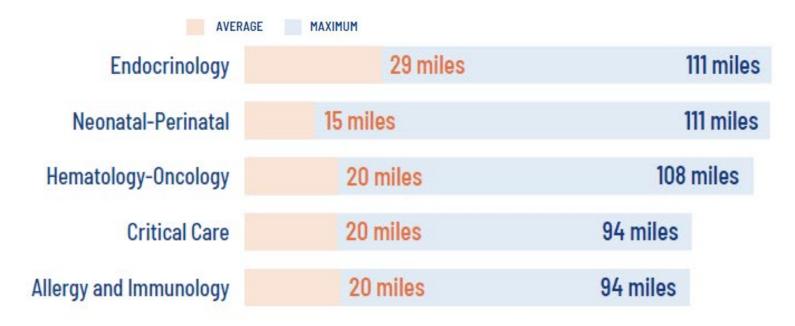
- AR, ~1:10,000-15,000 births
- Cortisol <u>+</u> aldosterone deficiency, female genital and CNS virilization
- Pre-NBS salt-wasting adrenal crises, sex mis-assignment
- NBS filter paper 170HP ——
- High false positive -> second tier strategies evolving
- Treatment cortisol and mineralocorticoid replacement
- Follow-up
 - Lab, PE, growth monitor q3-6m
 - Frequent stress-dose management for illness/injury
 - Puberty complex psychosexual and medical management





Specialty care for children with CH and CAH: shortage and maldistribution of pediatric endocrinologists

HOW FAR DOES A CHILD IN WISCONSIN NEED TO DRIVE FOR PEDIATRIC SUBSPECIALTY CARE?



Ten states have fewer than 1 PE per 100 000 children





NOTES FROM THE ASSOCIATION OF MEDICAL SCHOOL PEDIATRIC DEPARTMENT CHAIRS, INC.



Sustaining the Pediatric Endocrinology Workforce: Recommendations from the Pediatric Endocrine Society Workforce Task Force

David B. Allen, MD¹, Tandy Aye, MD², Charlotte M. Boney, MD³, Erica A. Eugster, MD⁴, Madhusmita Misra, MD, MPH⁵, Kanakadurga Singer, MD⁶, Diane Stafford, MD², Selma F. Witchel, MD⁷, and Philip Zeitler, MD, PhD⁸

What Evidence Raises Concern about the Future Pediatric Endocrinology Workforce?

Dwindling recruitment:

- Total PE fellows declined from 254 (2012) to 243 (2018)
- Applicant/position = 0.7 -> 41/108 positions unfilled (2020)



Declining recruitment to Pediatric Endocrinology

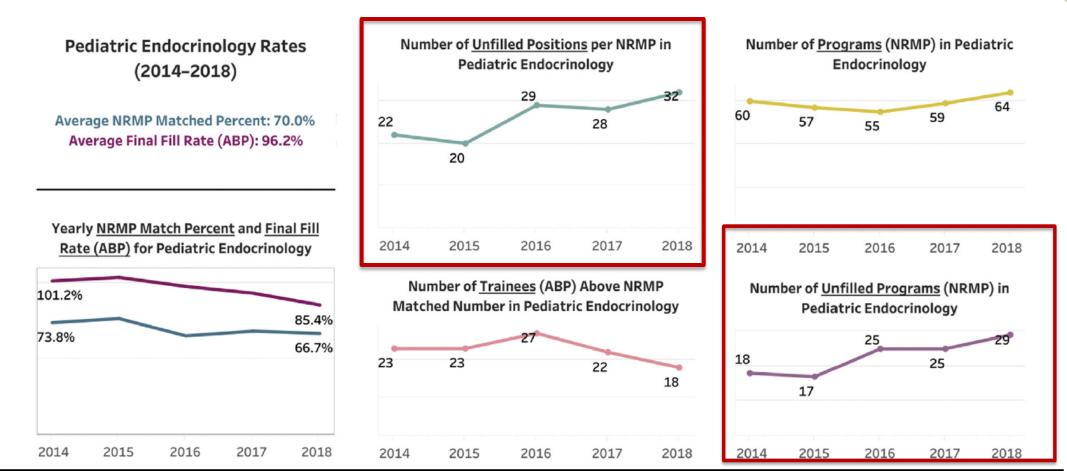


Figure. Pediatric endocrinology fellowship match and program fill rates from 2014 to 2018. Source: National Resident Matching Program (NRMP) data and American Board of Pediatrics (ABP) data; https://www.abp.org/content/comparison-abp-data-nrmp-match-data.





Dwindling recruitment:

- Total PE fellows declined from 254 (2012) to 243 (2018)
- Applicant/position = 0.7 -> 41/108 positions unfilled (2020)

Declining and under-diverse current workforce

- Aging 21% >60 years old
- Early career 80% female (many part-time)
- URM 23% of trainees, 5.5% black

Growing patient population

- Increasing T1DM and (especially) T2DM and obesity-morbidity
- Transgender, cancer survivors, retained complex young adult pts

Synchronous trends— \downarrow fellowship recruitment, \uparrow patient numbers -> shortage of trained pediatric endocrinologists



What Factors and Perceptions Threaten the Pediatric Endocrinology Workforce Pipeline?

Lack of early subspecialty exposure and mentorship

- Not required medical school rotation
- Typical 3rd year residency exposure post-career decision

Financial concerns

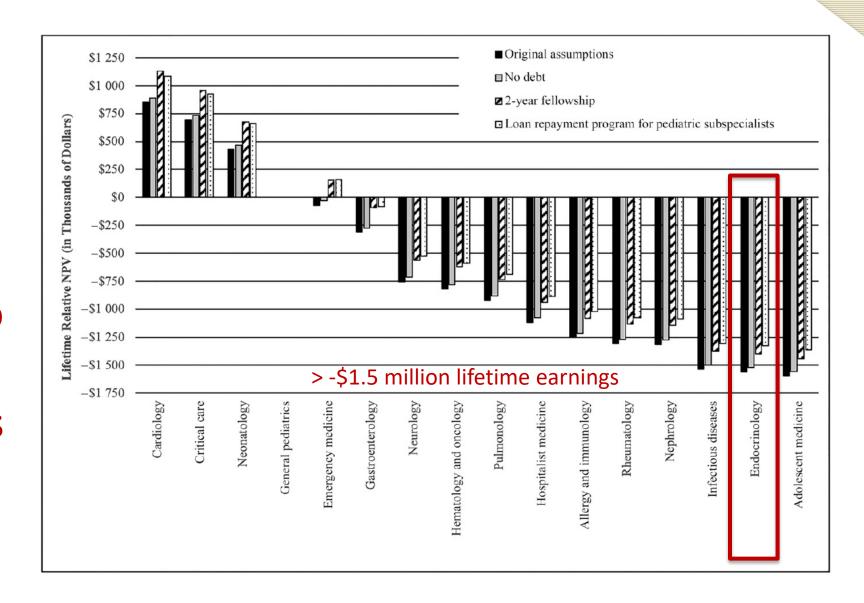
- Medical student debt (averaging \$232K) dissuades additional training
- Relatively low average salary of PE

Negative professional QOL perceptions

- Personal/professional life boundaries perceived unpredictable for PE
- Competition from shift-scheduled specialties (e.g. hospitalist)

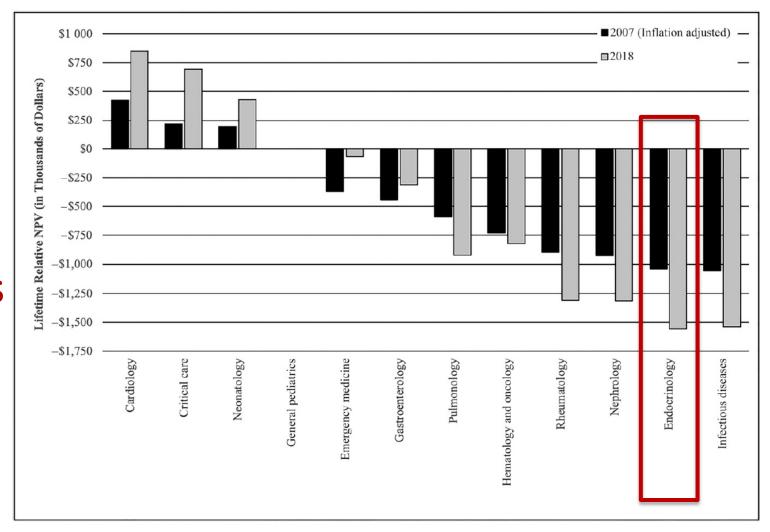


Lifetime earnings of pediatric specialists compared to general pediatricians





Pediatric endocrinology: largest relative decline in lifetime earnings among pediatric specialists 2007 -> 2018



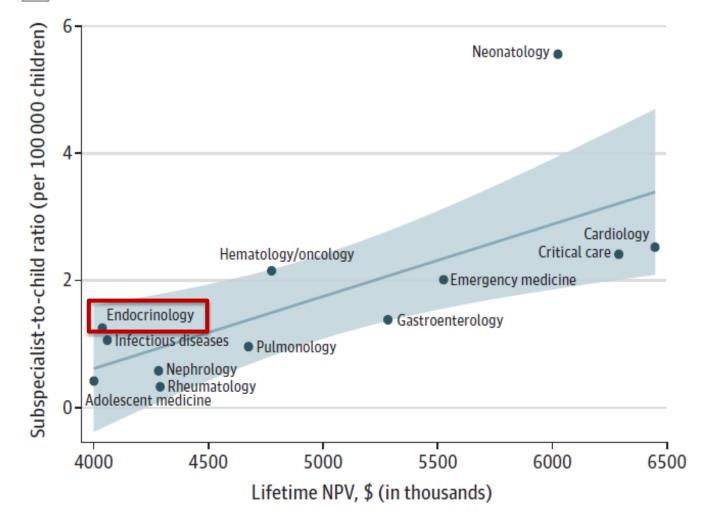


Lifetime earning potential

predicts:

- distance to subspecialists
- # subspecialists per regional pediatric population
- increase in # specialists/population growth
- fill rates for fellowship training spots







What Efforts Are Needed to Increase the Numbers and Diversity of the Pediatric Endocrinology Workforce?

Increase early positive exposure to Pediatric Endocrinology

- Outpatient subspecialty exposure in core rotations
- Emphasize exposure to enthusiastic fellow and faculty mentors
- ACGME/CoPS/APPD support for early residency exposure to nonprocedural subspecialties
- Professional society medical student recruitment initiatives





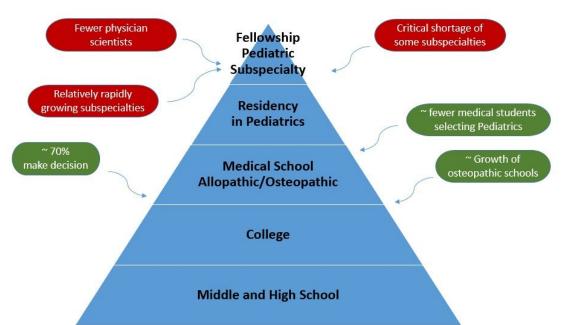
Lessening barriers

- Financial
 - Expand loan forgiveness for work in underserved areas and lower-paid specialties
 - Fund a targeted Loan Repayment Program for non-procedural specialties
 - Implement shared-care models that value non-procedural PE expertise
- Duration of training
 - Re-evaluate 2 year program and modify if deemed appropriate
- Perceived lifestyle detractors
 - Expand utilization of care extenders
 - Embrace technology to improve work/personal life balance



Pediatrics 2025: AMSPDC Workforce Initiative

Workforce Pipeline



Objective: attract trainees to undersubscribed pediatric subspecialties:

- Change medical education paradigm
- Early exposure and marketing
- Address economics
 - Lessen financial burden
 - Equalize Medicare and Medicaid revenue streams
 - Equalize compensation

Steps to ensure follow-up for endocrine disorder NBS

- Early life close collaboration with NBS program
- Recruitment to strengthen viability of the PE specialty
- Assistance from care extenders (PA, NPP)
 - Effective for CH >> CAH
 - Private practice >> academic institutions
- Adult medicine collaboration/early transition
 - Limited necessity for CH
 - CAH transition complicated and thus far not satisfactory
- Technology (propelled forward by COVID-19)
 - Improves patient access (>50% CH follow-up visits)
 - Does not address provider shortage



Thank you for this opportunity to present