

Facilitating a Public Private Partnership – Creation of Data Sets by the CAH/DSD Endocrine Work Group of the Region 4 Genetics Collaborative (R4GC)

*Newborn Screening Translational Research Network
Disease Specific Workgroup Meeting
February 25-26, 2010 Bethesda, Maryland*

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Data Sets

CAH Registries

- CAH 21-OHD enrollment infant/child
- CAH 21-OHD enrollment adolescent
- CAH 21-OHD interval infant/child
- CAH 21-OHD interval adolescent
- CAH 3b-HSD enrollment infant/child
- CAH 3b-HSD enrollment adolescent
- CAH 3b-HSD interval infant/child
- CAH 3b-HSD interval adolescent
- CAH 11b-OHD enrollment infant/child
- CAH 11b-OHD enrollment adolescent
- CAH 11b-OHD interval infant/child
- CAH 11b-OHD interval adolescent
- CAH 17-OHD/17.20 lyase enrollment infant/child
- CAH 17-OHD/17.20 lyase enrollment adolescent
- CAH 17-OHD/17.20 lyase interval infant/child
- CAH 17-OHD/17.20 lyase interval adolescent
- CAH Lipoid enrollment infant/child
- CAH Lipoid enrollment adolescent
- CAH Lipoid interval infant/child
- CAH Lipoid interval adolescent

DSD Psychological

- Psych enrollment infant/child
- Psych enrollment adolescent

DSD Urologic

- Urology enrollment infant/child
- Urology enrollment adolescent
- Urology interval infant/child
- Urology interval adolescent

DSD Registries

- Turner enrollment infant/child
- Turner enrollment adolescent
- Turner interval infant/child
- Turner interval adolescent
- Klinefelter enrollment infant/child
- Klinefelter enrollment adolescent
- Klinefelter interval infant/child
- Klinefelter interval adolescent
- 46,XX DSD - partial androgen effect enrollment infant/child
- 46,XX DSD - partial androgen effect interval infant/child
- 46,XX DSD - partial androgen effect enrollment adolescent
- 46,XX DSD - partial androgen effect interval adolescent
- 46,XX DSD - no androgen effect enrollment infant/child
- 46,XX DSD - no androgen effect interval infant/child
- 46,XX DSD - no androgen effect enrollment adolescent
- 46,XX DSD - no androgen effect interval adolescent
- 46,XY DSD - partial androgen effect enrollment infant/child
- 46,XY DSD - partial androgen effect interval infant/child
- 46,XY DSD - partial androgen effect enrollment adolescent
- 46,XY DSD - partial androgen effect interval adolescent
- 46,XY DSD - no androgen effect enrollment infant/child
- 46,XY DSD - no androgen effect interval infant/child
- 46,XY DSD - no androgen effect enrollment adolescent
- 46,XY DSD - no androgen effect interval adolescent
- Other DSD - enrollment infant/child
- Other DSD - enrollment adolescent
- Other DSD - interval infant/child
- Other DSD - interval adolescent

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Region 4 Genetics Collaborative

- Public-private partnership created in 2003
- Specialists, laboratorians, pediatricians, genetic counselors, state public health staff and parents of children
- Seven Midwest states (Illinois, Indiana, Kentucky, Michigan, Minnesota, Ohio, and Wisconsin).

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Goals of the R4GC

- Facilitate data collection
- Provide a forum to share best practice models
- Improve diagnosis, treatment, care-coordination and long term outcomes
- LTFU – Inborn Errors of Metabolism

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Leo Fung Center for CAH/DSD

- Public private partnership opens in June 2005
- 1st multidisciplinary CAH/DSD center in the country where
 - Onsite providers operate from single clinic location
 - Direct participation from Minnesota Dept of Health
 - Public health nurse and NBS follow-up specialist

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Leo Fung Center for CAH and DSD

- **University of Minnesota**
 - Department of Pediatrics
 - Division of Ped Endo
 - Division of Genetics and Metabolism
 - Department of Urology
 - Division of Pediatric Urology
 - Dept. of Family Medicine & Community Health
 - Program in Human Sexuality
 - Dept. of OB-GYN and Women's Health
- **Minnesota Department of Health**
 - Newborn Screening Program
- **UMN Amplatz Children's Hospital**
 - Genetic Counseling
 - Child-Family Life Services

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Public-Private Partnership

Mayo Clinic

Leo Fung Center for CAH/DSD

Minnesota Department of Health

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Public-Private Partnership

Mayo Clinic

Leo Fung Center for CAH/DSD

Minnesota Department of Health

4 year old
17-OHP : 21,000
Bone age: 13 years
Missed by NBS

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Print Form

False Negative CAH Report for the Newborn Screening Program
Fax completed report to 651.291.3477

Please include patients born between and inclusive of 2004-2009

Patient Identified with Congenital Adrenal Hyperplasia (CAH)

Child's Name: Last First DOB

Parent/Guardian Name: Last First

Diagnostic Information on Patient:

Was the patient diagnosed by Newborn Screening? Yes No

CAH Form: 21-OHD 11-OHD 3-hydro DSD 17-OHD (17, 20 form) S/MAR

CAH 21-OHD subtype: Ash-Weisinger Non-classical Simple Virilizing Not determined

What was the age at diagnosis?

Confirmatory 17-OHP level: male female

ACTH Stimulation Performed? Yes No If yes, please attach a copy of the results

Genetic Testing Completed? Yes No Mutations: and

Please check patient's presenting symptoms:

<input type="checkbox"/> Abnormal Sodium - High	<input type="checkbox"/> Abnormal Renin - High	<input type="checkbox"/> Other
<input type="checkbox"/> Abnormal Sodium - Low	<input type="checkbox"/> Abnormal Renin - Low	
<input type="checkbox"/> Abnormal Potassium - High	<input type="checkbox"/> Abnormal Glucose	
<input type="checkbox"/> Abnormal Potassium - Low	<input type="checkbox"/> Abnormal Electrolytes	
<input type="checkbox"/> Growth Acceleration	<input type="checkbox"/> Family Enlargement	
<input type="checkbox"/> Pubic Hair Development	<input type="checkbox"/> Hypertension	
<input type="checkbox"/> Amblyopia/Strabismus	<input type="checkbox"/> Bone Age Advancement	

* Thank you for helping Minnesota better screen for Congenital Adrenal Hyperplasia!

MDH
Minnesota Department of Health
655 West County St., Suite 800, St. Paul, MN 55104
Phone: 651-291-3477 Fax: 651-291-3478 or 800-644-3774
www.mnhs.gov www.mn.gov/healthscreening

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- ## Region 4 Genetics Collaborative
- Creating a CAH/DSD Endocrine Work Group
 - Developing long term follow-up data sets
 - Providing the infrastructure, the Region 4 facilitated:
 - Teleconferences
 - Face to face meetings
 - Distribution of material
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- ## Fundamental Considerations for Creating the CAH Data Sets
- 21-OHD vs. including other forms of CAH
 - Register patients or capture current practices
 - Sub-divide by specific disorder and age
 - Multidisciplinary and long-term approach
 - Collecting data from metabolic specialists vs. endocrinologists
 - Provider settings
 - Input from the Departments of Health and PHL
 - AHRQ guidelines
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- ## Fundamental Considerations in Creating the CAH Data Sets
- 21-OHD vs. including other forms of CAH
 - "Register" patients or capture current practices
 - Are current practices based on the consensus statements?
 - Consensus statements – Are they followed?
 - > 20% patients referred to Leo Fung Center with previous CAH DX have their DX change to different 21-OHD sub-type
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Fundamental Considerations in Creating the CAH Data Sets

- Sub-divide by specific disorder and age
 - Single catch-all CAH survey – too long

- Multidisciplinary and long-term approach
 - Endocrine/Genetics/NBS
 - Psychological
 - Urologic

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Fundamental Considerations in Creating the CAH Data Sets

- Collecting data - metabolic specialists vs. endocrinologists
 - Metabolic centers
 - University based
 - Fewer centers per state (some with 1 or none)
 - All inborn errors are rare
 - Endocrine centers
 - University, hospital, private practice based
 - Higher numbers per state
 - Not all endocrine disorders are rare

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Fundamental Considerations in Creating the CAH Data Sets

- Provider settings
 - University-based and clinics
 - Private hospital-based clinics
 - Private practice clinics

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Fundamental Considerations in Creating the CAH Data Sets

- Input from the Departments of Health and PHL
- AHRQ guidelines
 - Guidelines for understanding and creating registries are available for free on the web
 - <http://www.ahrq.gov/>

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Fundamental Considerations for Implementing the CAH Data Sets

- Centralized administrative support
- Data extraction
- Retrospective data

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Fundamental Considerations for Implementing the CAH Data Sets

- Centralized administrative support
 - Wide spectrum of provider settings
 - Clinical workload of endocrinologists
 - Getting someone to sign on does not mean any data will get collected
- All regulatory paperwork initiated and followed-up
- Potential projects, NIH grant announcements, assist with grants, etc

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Fundamental Considerations for Implementing the CAH Data Sets

- Data extraction
 - Heterogeneity of data
 - Site retention
 - Retrospective data

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Fundamental Considerations for Implementing the CAH Data Sets

- Retrospective data
 - To collect or not to collect . . .
 - Feasibility

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The Minnesota Experience

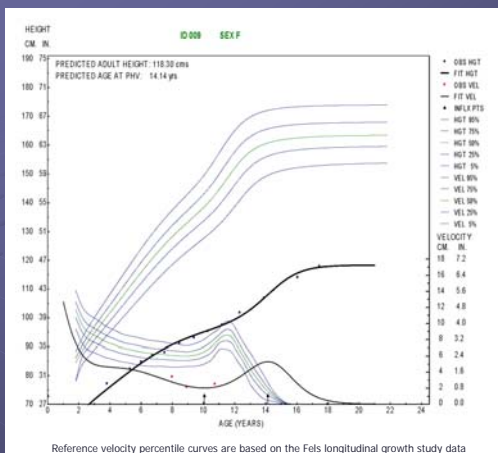
- Obesity in CAH
 - University of Minnesota
 - The Mayo Clinic
 - Children's Hospitals of Minnesota

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Growth and Obesity in CAH

- ~ 282 CAH cases collected throughout Minnesota
- Dating back to 1965
- Many individual cases span 18 years

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UMN Statistical Model for CAH

- Statistical model being developed with Dr. John Himes (Chair of group that created CDC growth charts)
- Bayesian statistical methods
- Can be used for irregular, incomplete, longitudinal, or fragmented data
- Height data at maturity not required
- 3 stages of logistic growth (triple logistic regression) detecting growth velocity at:
 - Early childhood
 - Mid childhood
 - Adolescence
- Can potentially be used to create CAH specific growth charts when more data is collected regionally and nationally

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Growth Parameters

Other obtainable summaries from model include:

- Age at peak height velocity (y)
- Peak height velocity (cm/y)
- Growth during peak height velocity (cm)
- Age at Mid growth velocity spurt
- Mid growth spurt velocity
- Stature gained during mid growth spurt
- Predicted or measured (if the data are there) adult height (cm)

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Partners

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