Implementation of Newborn Screening for Critical Congenital Heart Disease (CCHD) in New York State

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Learning Objectives

- Recognize the public health significance of CCHD in NYS.
- Describe the requirements for universal CCHD screening of infants born at home or in a NYS hospital.
- Discuss the availability of NYS information resources about CCHD for parents and guardians.
- Recognize the CCHD conditions that could be associated with a low oximetry result.
- Identify the best practices process for CCHD implementation in a hospital based setting.
- Identify challenges for CCHD implementation in NYS.
Disclosure Statements

The planners and presenters do not have any financial arrangements or affiliations with any commercial entities whose products, research or services may be discussed in this activity.

No commercial funding has been accepted for this activity.

Continuing Education Credits

- Credits available: CME, CNE, and CHES
- To obtain continuing education credits, participants must complete an evaluation and score 80% of above on the post-test.
- A link to the evaluation and post-test will be available after the webinar.
- Continuing education credits are available for this webinar until February 2016.
Webinar Guidelines

- You will listen to the audio through your computer speakers. Please make sure they are turned on and turned up.
- Adobe Features you will use today:
  - Chat Box
  - Polls

- Type any questions you have into the chat box, and they will be answered at the end of session.
- Today’s session is being recorded

Disclosures

- Dr. Kacica has nothing to disclose.
Implementation of Newborn Screening for Critical Congenital Heart Disease (CCHD) in New York State

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Medical Director
Division of Family Health
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WHAT IS THE PUBLIC HEALTH SIGNIFICANCE OF CCHD?

- 2-3 out of 1,000 live births have Critical Congenital Heart Disease (CCHD)
- ~300 children in New York State are born with CCHD each year
- 17% of these children will die in the first year of life
- Universal screening may prevent up to 50 infant deaths each year in New York State
### Number/Prevalence of NYS Children with CCHD, Including Number (%) that Died in First Year Birth Year, 2007-2009

<table>
<thead>
<tr>
<th>Type of CCHD</th>
<th>Number</th>
<th>Prevalence*</th>
<th>N (%)</th>
<th>Average Annual cases</th>
</tr>
</thead>
<tbody>
<tr>
<td>Hypoplastic left heart syndrome</td>
<td>192</td>
<td>2.61</td>
<td>72 (37.5%)</td>
<td>64</td>
</tr>
<tr>
<td>Pulmonary atresia (intact septum without VSD)</td>
<td>50</td>
<td>0.68</td>
<td>8 (16.0%)</td>
<td>17</td>
</tr>
<tr>
<td>Tetralogy of Fallot</td>
<td>357</td>
<td>4.85</td>
<td>26 (7.3%)</td>
<td>119</td>
</tr>
<tr>
<td>Total anomalous pulmonary venous return (TAPVR)</td>
<td>87</td>
<td>1.18</td>
<td>14 (16.1%)</td>
<td>29</td>
</tr>
<tr>
<td>Position of Great Arteries</td>
<td>277</td>
<td>3.76</td>
<td>55 (19.9%)</td>
<td>92</td>
</tr>
<tr>
<td>Tricuspid atresia</td>
<td>41</td>
<td>0.56</td>
<td>8 (19.5%)</td>
<td>14</td>
</tr>
<tr>
<td>Truncus arteriosus</td>
<td>36</td>
<td>0.49</td>
<td>6 (16.7%)</td>
<td>12</td>
</tr>
<tr>
<td>Total by Defect**</td>
<td>1040</td>
<td>14.11</td>
<td>189 (18.2%)</td>
<td>347</td>
</tr>
<tr>
<td>Total by Child**</td>
<td>916</td>
<td>12.43</td>
<td>158 (17.3%)</td>
<td>305</td>
</tr>
</tbody>
</table>

**110 children had more than one CCHD**

### BACKGROUND

#### National Recommendations for CCHD Screening

- **September 2010**
  - Final draft of comprehensive evidence based report on CCHD presented to the federal Maternal & Child Health Bureau at HRSA
  - Secretary’s Advisory Committee on Heritable Disorders in Newborns & Children (SACHDNC) voted to add CCHD to the Recommended Uniform Screening Panel (RUSP)

- **September 2011**
  - Secretary Sebelius adopted the SACHDNC’s recommendation to add CCHD to the Newborn RUSP
NYS PUBLIC HEALTH LAW 2500-a.(a)
Effective January 27, 2014
Mandates pulse oximetry screening in NYS

- Facilities caring for infants 28 days or less and the person registering the birth of a child to test for CCHD through pulse oximetry screening

- Pulse oximetry screening should be performed after the baby is 24 hours old and < 48 hours of life

- Information to be disseminated to the parents and guardians of the infant tested

Note: Parents may object to testing based upon religious teachings/tenets
NYS Recommendations for CCHD Screening to Providers

- Document screening results in medical record
- Assure appropriate referrals for diagnostic evaluation
- Collect appropriate follow up data as to assure that all babies are screened and receive needed treatment
- Report cases of diagnosed CCHD to Congenital Malformations Registry

New York State’s Congenital Malformation Registry
Data Submission for Reporting Cases of CCHD

HCS for CMR Data Submission
NYS Recommendations Regarding Follow-Up Data for Pulse Oximetry Screening

- Data & time of screening
- Screening results (pass/fail)
- Referral to medical provider after failed screen (Y/N)
- Identification of an infant with CCHD by another means (prenatal U/S, clinical signs prior to pulse ox screening)
- Diagnostic results
- Parent refusal of screening

NYSDOH Role in the Implementation of Newborn Screening for CCHD

- Provided notice in January 2014 to health care providers (hospitals, physicians, nurse midwives) via Health Commerce System about requirements for screening and reporting of diagnosed CCHD cases

- Developed NYS tools and resources
  - Algorithm (distributed with provider notice)
  - Series of Parent information Resources
Child in well-baby nursery* at 24-48 hours of age (or shortly before discharge if < 24 hours of age)

- \(< 90 \% \text{ in RH or F}\)
  - Fail
  - Do not rescreen

- \(90-94 \% \text{ in RH and F}\) or \(> 3 \% \text{ difference between RH and F}\)
  - \(\geq 95 \% \text{ in RH or F}\)
  - \(\leq 3 \% \text{ difference between RH and F}\)
  - Repeat Screen in one hour

- \(< 90 \% \text{ in RH or F}\) and \(> 3 \% \text{ difference between RH and F}\)
  - \(\geq 95 \% \text{ in RH or F}\)
  - \(\leq 3 \% \text{ difference between RH and F}\)

**FAIL**
- Notify responsible medical practitioner about failed screen.
- PCP should conduct clinical assessment for causes of low oxygen (sepsis, pneumonia, pulmonary hypertension). In absence of clear cause for low oxygen, get echocardiogram within 24 hours. This may require transfer or telemedicine.
- If saturation is \(< 90\% in either the hand or foot, the baby should have immediate clinical assessment and refer to pediatric cardiology. In this case, do not wait and rescreen.

**PASS**
- Pass on the screen does not exclude the existence of a cardiac disorder.
- Further evaluation is otherwise indicated (e.g., clinical signs, prenatal diagnosis of critical congenital heart disease, dysmorphic features, etc., and original pulse oximetry screen.
NYS CCHD Screening Parent Resources

- General CCHD Screening Fact Sheet
- Second Fact Sheet explaining
  - What Does a Low Result Mean?
- Congenital Heart Defect Resources Fact Sheet

Congenital Heart Defects Resources

Explore these resources to find information, products, services and support for children with congenital heart defects and their families.

Cardiology Services
- Congenital Heart Information Network (CHIN): tchin.org
- It's My Heart: www.itsmyheart.org
- Kids with Heart National Association for Children's Heart Disorders: kidswithheart.org

General Information
- American Heart Association (AHA) www.heart.org
- Centers for Disease Control and Prevention (CDC) www.cdc.gov
- Mayo Clinic: www.mayoclinic.com
- National Birth Defects Prevention Study (NBDS) www nbdps.org

Care packages: Deliver gifts & supplies during a child’s hospital stay
- Mended Little Hearts www.mendedlittlehearts.org
- Saving Little Hearts www.savinglittlehearts.com
Poll Question

Is information on CCHD screening currently being given to parents or guardians by your facility or organization?

a) Yes
b) No
c) N/A
Robert Koppel M.D.

CCHD Implementation, Cardiac Anomalies Associated with CCHD and Best Practices
Physician’s Point of View

New York State Department of Health
CCHD Screening Webinar
June 9, 2014
Robert Koppel, MD
Cohen Children’s Medical Center of New York
North Shore LIJ - Hofstra University School of Medicine
Disclosures

• Dr. Koppel has nothing to disclose.

Reliability
The Cyanotic “Blind Spot”

Hemoglobin of 17.5 g/dL

Abnormal Saturation
Visible Cyanosis

83%

Abnormal Saturation
No Visible Cyanosis

95%

Hemoglobin of 13.5 g/dL

78%

95%


Reliability of Pulse Oximetry Screening for CCHD

- Meta-analysis of 13 eligible studies with data for 229,421 newborn babies

- Sensitivity: 76.5% (95% CI 67.7 – 83.5)
- Specificity: 99.9% (95% CI 99.7 – 99.9)
- False-positive rate: 0.14% (95% CI 0.06 – 0.33)

Reliability of Pulse Oximetry Screening for CCHD

- The false positive rate was particularly low when pulse oximetry was done after 24 hours from birth compared to being done before 24 hours
  - 0.05% [0.02 – 0.12] vs. 0.50 [0.29 – 0.86]
  - \( p = 0.0017 \)


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Reliability of Pulse Oximetry Screening for CCHD

- China
  - 122,738 babies
  - Sensitivity of pulse oximetry plus clinical assessment:
    - 93.2% (95% CI 87.9 – 96.2)
  - False positive rate
    - Clinical assessment: 2.7%
    - Oximetry – 0.3%

### Detection of CCHD Lesions (SpO$_2$ < 95%)

<table>
<thead>
<tr>
<th>CCHD Lesion</th>
<th>Total</th>
<th>Percent Detected</th>
</tr>
</thead>
<tbody>
<tr>
<td>DORV</td>
<td>3/3</td>
<td>100</td>
</tr>
<tr>
<td>HLHS</td>
<td>5/5</td>
<td>100</td>
</tr>
<tr>
<td>PA</td>
<td>5/5</td>
<td>100</td>
</tr>
<tr>
<td>d-TGA</td>
<td>9/9</td>
<td>100</td>
</tr>
<tr>
<td>TAPVC</td>
<td>6/7</td>
<td>85.7</td>
</tr>
<tr>
<td>Truncus</td>
<td>7/8</td>
<td>87.5</td>
</tr>
<tr>
<td>TA</td>
<td>1/1</td>
<td>100</td>
</tr>
<tr>
<td>AA/AS</td>
<td>3/4</td>
<td>75.0</td>
</tr>
<tr>
<td>TOF</td>
<td>9/13</td>
<td>69.2</td>
</tr>
<tr>
<td>AVSD</td>
<td>4/5</td>
<td>80.0</td>
</tr>
<tr>
<td>CoA</td>
<td>8/15</td>
<td>53.3</td>
</tr>
<tr>
<td>PS</td>
<td>2/6</td>
<td>33.3</td>
</tr>
</tbody>
</table>


### CCHD Screening Targets
CCHD Screening

• 7 primary targets
  • Hypoplastic left heart syndrome
  • Pulmonary atresia (with intact septum)
  • Tetralogy of Fallot
  • Total anomalous pulmonary venous return
  • Transposition of the great arteries
  • Tricuspid atresia
  • Truncus arteriosus

Hypoplastic Left Heart Syndrome
Pulmonary Atresia with Intact Septum

Tetralogy of Fallot
Total Anomalous Pulmonary Venous Return

Transposition of the Great Arteries
CCHD Screening

- 5 secondary targets:
  - Coarctation of the aorta
  - Double outlet right ventricle
  - Ebstein anomaly
  - Interrupted aortic arch
  - Single ventricle

- *Can be just as severe but not consistently detected*
- *Screening may also detect other significant medical conditions that present with hypoxemia*
Interrupted Aortic Arch

Single Ventricle

- Tricuspid atresia
- Hypoplastic left heart syndrome
- Double inlet left ventricle
- Many of the heterotaxy defects
- Some variations of double outlet right ventricle
Other Conditions Detected During CCHD Screening

35859 Deliveries

208 Admitted following positive pulse oximetry screening

17 CHD

44 Transitional circulation

140 Other significant diagnosis

9 COH

3 Serious CHD

5 Significant CHD

11 PPHN

56 Congenital pneumonia

4 Meconium aspiration

3 Pneumothorax

42 TTN requiring oxygen

3 Other Diagnoses

Figure 3 Outcomes of test positives following pulse oximetry screening.


Differential Diagnosis of Neonatal Cyanosis

A Airway

B Breathing

C Circulation

<table>
<thead>
<tr>
<th>Condition</th>
<th>Description</th>
<th>Description</th>
<th>Description</th>
</tr>
</thead>
<tbody>
<tr>
<td>Choanal atresia</td>
<td>Pneumonia</td>
<td>Oxygen carrying capacity</td>
<td></td>
</tr>
<tr>
<td>Macroglossia</td>
<td>Congenital diaphragmatic hernia</td>
<td>Polycythemia</td>
<td></td>
</tr>
<tr>
<td>Pierre Robin sequence</td>
<td>Congenital cystic adenomatoid malformation</td>
<td>Anemia</td>
<td></td>
</tr>
<tr>
<td>Laryngomalacia</td>
<td>Pulmonary sequestration</td>
<td>Methemoglobinemia</td>
<td></td>
</tr>
<tr>
<td>Vocal cord paralysis</td>
<td>Congenital lobar emphysema</td>
<td>Congenital heart disease</td>
<td></td>
</tr>
<tr>
<td>Tracheal stenosis</td>
<td>Pulmonary hypoplasia</td>
<td>Decreased pulmonary blood flow</td>
<td></td>
</tr>
<tr>
<td>Vascular sling/swing</td>
<td>Phrenic nerve palsy</td>
<td>Tricuspid stenosis</td>
<td></td>
</tr>
<tr>
<td>Cystic hygroma</td>
<td>Hypoventilation</td>
<td>Pulmonary atresia</td>
<td></td>
</tr>
<tr>
<td>Hemangioma</td>
<td></td>
<td>Pulmonary stenosis</td>
<td></td>
</tr>
<tr>
<td>Other neck masses</td>
<td></td>
<td>Tetralogy of Fallot</td>
<td></td>
</tr>
<tr>
<td></td>
<td></td>
<td>Ehlert’s anomaly</td>
<td></td>
</tr>
<tr>
<td></td>
<td></td>
<td>Inadequate mixing</td>
<td></td>
</tr>
<tr>
<td></td>
<td></td>
<td>Transposition of the great arteries</td>
<td></td>
</tr>
<tr>
<td></td>
<td></td>
<td>Persistent pulmonary hypertension</td>
<td></td>
</tr>
</tbody>
</table>

Performing the Screen

• Use an appropriate device
  • Standardized hospital grade
  • Motion tolerant
  • FDA approved
  • Proper sensors are used with the device

Supplies

• Disposable or re-usable sensors
  • Disposable - single patient use
  • Reusable
    • maintenance of infection control
    • proper cleaning between patients

• Use according to manufacturer’s instructions
## Application of the Sensor

- Apply to clean, dry skin
- Best sites: outer aspects of the palm and foot, the great toe and thumb
- The wrist is not recommended
- Light emitter and photodetector directly opposite each other

## Performing the Screen

- Secure the sensor to the infant’s right hand to obtain a pre-ductal reading and either foot for a post-ductal reading
- Turn on the oximeter
- Connect probe
- Wait for pleth wave (arterial pulse) to stabilize
- Assess HR correlation
- Assess saturation reading
- Document
Poll Question

Are staff routinely performing the pulse oximetry on the baby’s right hand and a foot for each screen?

a) Yes  
b) No  
c) N/A

Performing the Screen

- The baby should be at least 24 hours of age.
- Can be done with other newborn screening - hearing, metabolic - that is also done after 24 hours of age and prior to discharge.
- Conduct screening in quiet area and, if possible, with parent present to soothe and comfort the infant.
- Conduct screening while infant is awake and quiet.
- Avoid screening when infant is crying, cold or in a deep sleep.
Parent Education

• Explain the purpose of screening
  – to help detect some serious heart problems in well appearing infants
• Explain the screening process
• Timing of the screen
• How it is performed
• Screening does not detect all heart defects
• Warning signs of congenital heart disease
  – Sweating especially when feeding, difficulty feeding, fast breathing, poor weight gain, bluish or pale skin color.

Accuracy: Paper Algorithm v. Computer-Based Tool

![Bar graph showing accuracy comparison between paper algorithm and computer-based tool.]

Figure 2. Accuracy when using a paper algorithm vs a computer-based tool. In 20 mock scenarios for screening for CCHD using pulse oximetry, those using the computer-based tool identified the correct answer more often than those using the paper algorithm (P < .001 for paper vs computer in all comparisons).

Technical Factors

False positive and negative readings

- Poor perfusion
- Motion artifact
- Ambient light
- Partial probe detachment
- Differences between manufacturers

Physician Response to Failed Screen

- Examine the infant
  - Ensure that the baby is hemodynamically stable
  - Evaluate for cause of hypoxemia
    - Consider sepsis or pneumonia

- Any signs or symptoms of congenital heart defect should prompt rapid evaluation

- If baby is asymptomatic with no obvious cause for hypoxemia, a cardiologist or neonatologist should be consulted and an echocardiogram should be performed

- Do not discharge home until the underlying reason for hypoxemia has been identified or the hypoxemia has resolved

- Babies will often appear normal and have no clinical findings other than the low oxygen saturation, but a thorough evaluation is necessary

Managing the Positive Screen

• Unless a non-cardiac cause can be identified for a failed screen, an infant who fails the screen should have a diagnostic echocardiogram done before being discharged.

• This could involve an echocardiogram within the hospital or birthing center, transport to another institution for the procedure, or the use of telemedicine for remote evaluation.

Poll Question

Have there been any babies that have failed the pulse oximetry screen in your facility/care?

Yes or No
For Patients Who Failed Their CCHD Screens

- Confirm that the infant had a diagnostic echocardiogram
- Make sure that the patient receives appropriate follow-up, such as being seen by a cardiologist; and
- Facilitate long-term follow-up for patients diagnosed with CCHDs

Communication of Results to Primary Care

- Include screening results in discharge summary
- Include in the hand-off report to the receiving hospital if infant is transferred

**First Well Visit Post Discharge**

- Pediatrician should have access to all screening results from hospital (CCHD, Metabolic, Hearing)
- If patient not appropriately screened at birth facility, develop strategies for screening

---

**Follow-up Visit**

- Passing the newborn oxygen saturation screening DOES NOT rule out all important congenital heart disease

- It is crucial to note that an infant in a pediatric office may have severe heart disease
**Signs & Symptoms**

- Cyanosis
- Tachypnea (often with diaphoresis during feeding)
- Poor perfusion & pulses (femoral)
- Murmur – Not as pertinent
- Poor weight gain (if infant is thriving, heart failure is very unlikely)

**Case Presentation**

- 39 weeks, NSVD, Apgar 9/9
- Discharged home on Day 2
  - Oximetry screening - post-ductal SpO₂ 100%
- Day 3
  - Lethargy
  - Decreased PO intake
  - Dry diapers
  - Tachypnea
  - Evaluated by pediatrician
Case Presentation

• Referral to ED for respiratory distress
  – grunting
  – Retracting
  – unable to measure SpO₂
• Intubated
• Umbilical arterial and venous catheters inserted

Case Presentation

• ABG: 7.09/17/199/8/-23.3
• Chemistry: 143/8/104/6/63/5.98

• Echo: coarctation, DA closed
  – (history of normal fetal echo)
• Prostaglandin infusion
• Dialysis prior to CoA repair
“Swiss Cheese” Model of Accident Causation

Some holes due to active failures

Other holes due to latent conditions

SUCCESSIVE LAYERS OF DEFENSES

British Medical Journal 320 (7237): 768–770

“Swiss Cheese” Model of CCHD Screening Failure

North Shore LIJ
“Swiss Cheese” Model of Accident Causation

- Obstetric ultrasound
- Fetal echo
- Newborn physical exam
- Nursery course
- Oximetry screening

Ideal CCHD Screening Program

- **Education**
  - Nurses
    - Normal newborn
    - NICU
    - Formal screener training
    - Competency
    - Continuing education
  - Physicians
    - Pediatricians
    - Neonatologists
    - Cardiologists
    - Continuing education
Ideal CCHD Screening Program

- Education
  - Parents
    - Negative screen
    - Positive screen
    - languages
    - Literacy

Ideal CCHD Screening Program

- Standardization of algorithm
  - Minimize variation
    - Equipment
    - Supplies
Ideal CCHD Screening Program

- Universal testing
- Quality of Process
- Database
  - Local
  - Public health

**Model for Improvement**

What are we trying to accomplish?
How will we know that a change is an improvement?
What change can we make that will result in improvement?

- Act
- Plan
- Study
- Do

Ideal CCHD Screening Program

- Documentation
  - Automatic link to EHR
  - Automatic reporting to DOH
Ideal CCHD Screening Program

• Communication
  – Parents
  – Pediatrician
  – Cardiologist
Disclosures

• Adriann Combs, R.N., BSN has nothing to disclose.

Adriann Combs, R.N., BSN
Stony Brook University Hospital

Educational Events, Technical Assistance, Quality Assurance and Challenges
RPC’s Point of View
Did you host educational events for local hospitals about CCHD screening implementation?

- Initial update in 2012 with Update to Guidelines in Perinatal Care, 7th ed.
What type of technical assistance was provided?

- Strategies on incorporating screening into existing care epochs
  - With discharge bilirubin screening or test
  - With metabolic screen draw

- Clinical information regarding maximizing the infant’s condition for efficient, successful screening
  - Satiated
  - Quiet
  - Warm

Table. Failure rate and times for sensor application, for oximeter to generate and display data, and from birth to data display in delivery room resuscitation by method of sensor application

<table>
<thead>
<tr>
<th>Sensor applied to infant after connection to oximeter (n = 37)</th>
<th>Sensor applied to infant before connection to oximeter (n = 79)</th>
</tr>
</thead>
<tbody>
<tr>
<td>Failed to display data (%)</td>
<td>10 (27)</td>
</tr>
<tr>
<td>Time to apply sensor (sec)</td>
<td>20 (14-24)</td>
</tr>
<tr>
<td>Time to display data (sec)</td>
<td>41 (26-78)</td>
</tr>
<tr>
<td>Time from birth to data display (sec)</td>
<td>100 (74-157)</td>
</tr>
</tbody>
</table>

Technical assistance in identifying pulse oximeters that have a reliable signal
- Apply sensor to infant prior to attaching to cable
- Strategies to optimize signal attainment and strength
- Increased success in oximeter with signal extraction technology
What issues came up?

- Cost associated with screening
- Special circumstances: early discharge, home births
- Process for Pediatric Cardiology evaluation in settings without service
  - After a failed screen, the first step is to examine the infant to make sure the baby is hemodynamically stable, and then begin the process of evaluation for hypoxemia. Depending on the status of the baby, this could involve evaluating for sepsis or pneumonia. Any signs or symptoms of congenital heart defect should prompt rapid evaluation, including potential urgent transfer to a center with advanced care capabilities.
  - If the baby is asymptomatic and otherwise well, with no obvious cause for hypoxemia, a cardiologist or neonatologist should be consulted and an echocardiogram should be performed. Newborns should not be discharged home until the underlying reason or hypoxemia has been identified or the hypoxemia has resolved. Remember, these babies will often appear normal and have no clinical findings other than the low oxygen saturation. Still, a careful and thorough evaluation is necessary.
  - In addition, it is critical to remember that CCHD screening does not detect all cases of serious congenital heart defect. For example, coarctation of the aorta can be life threatening in early infancy, but may not be associated with hypoxemia.
Quality assurance activities

- Parent education
  - Was education provided?
  - Was education provided in the preferred language?
- Every baby screened
  - If a miss, why?
  - Do you have a policy that addresses transfers to a different level of care/another facility?
  - What if a parent refuses?
- Screeners following protocol
  - Education/training
  - Documentation
  - Knowledgeable re: process for notify LIP?
- Completeness/quality of screening data
  - Captured electronically
  - Data reviewed?
- Completeness/quality of data for CCHD positive r/t Congenital Malformation Registry
THANK YOU!
Contact

MARINA.SEPOWSKI@HEALTH.NY.GOV

Continuing Education Credits

Evaluation and post-test located here:
http://www.albany.edu/sph/cphce/mch_cchd.shtml