THE NATIONAL CHILDREN'S STUDY

HEALTH GROWTH ENVIRONMENT

Development of a Visit Assessment Tool to Address Birth Defects and Dysmorphology

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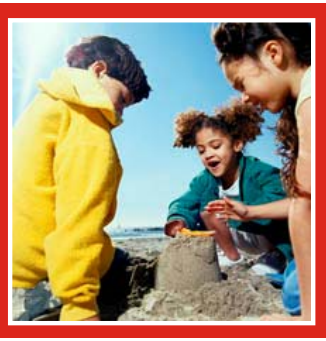
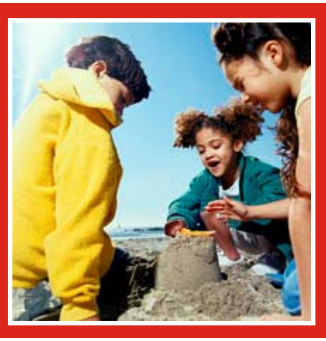
NCS Research Day, August 24, 2011

The Importance of Documentation of Structural Phenotype Variations in Children in the NCS



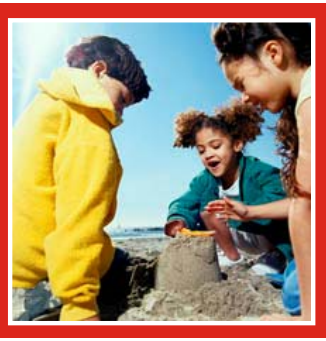
- Syndrome indicators
- Statistical association with more serious internal defects, e.g. ear tag
- Marker for identification of a teratogen
- Model for diagnostic reasoning, development of decision-making formulas, development of disease criteria
- Essential component of testing hypotheses in the NCS related to environmental exposures and genetic susceptibility

Challenges of Documentation of Structural Phenotype Variations in Children in the NCS



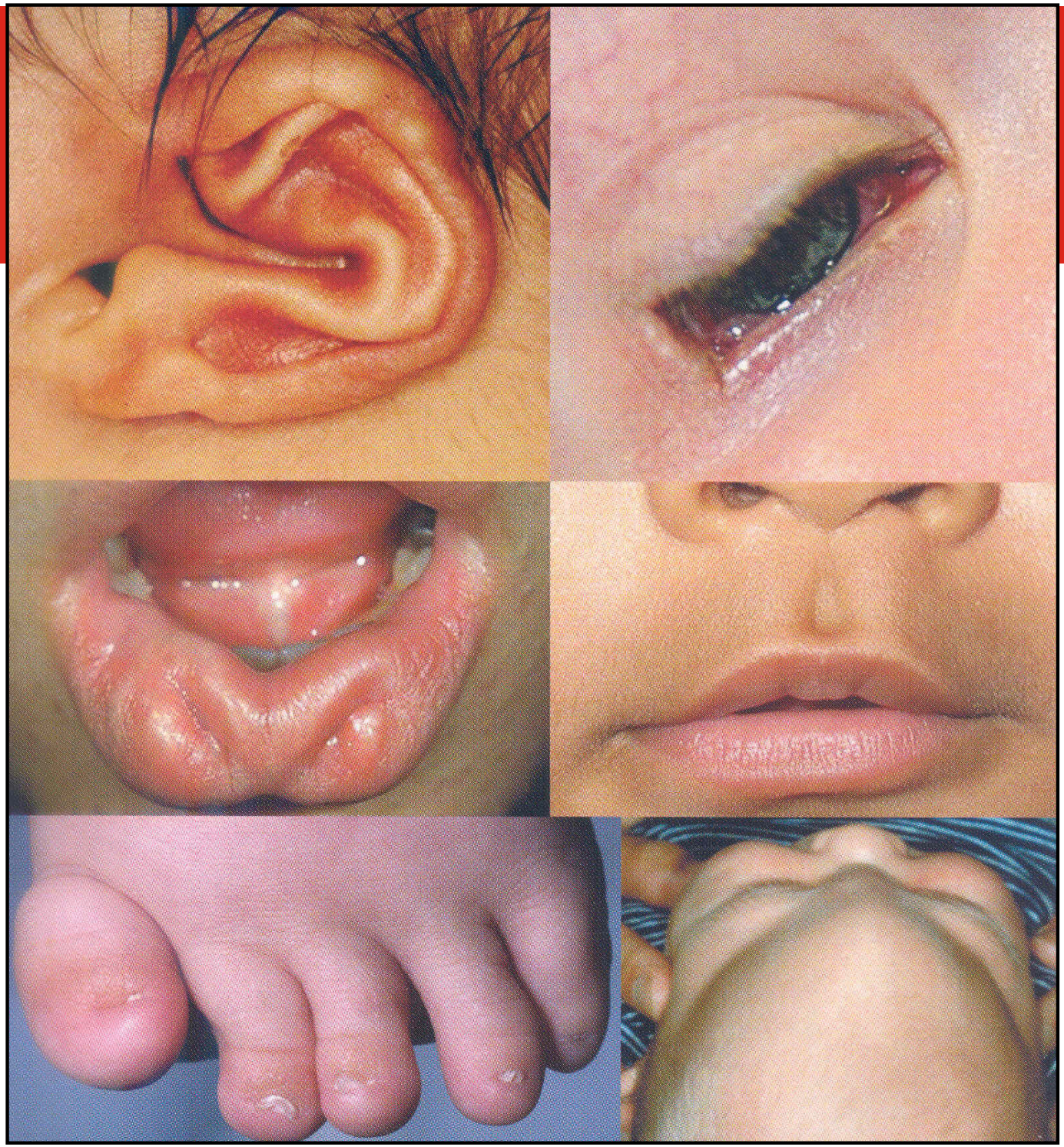
- No currently available validated protocol for assessment of a broad array of structural features in children that can reasonably be accomplished by non-specialists in a field setting
- Until recently, no standard definitions for classifying features
- No validated and feasible training paradigm for capturing this information using field staff with a wide variety of backgrounds and expertise

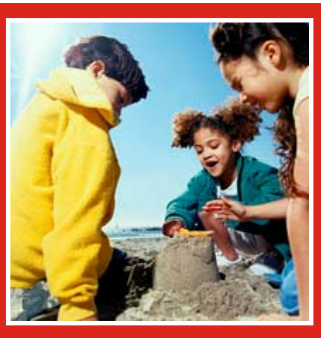
International Working Group on the Terminology and Definition of Phenotypic Findings



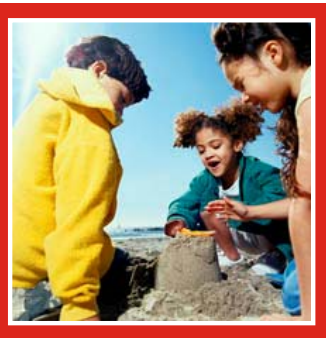
- Convened 34 clinical geneticists/dysmorphologists from nine countries in 2005
- Two formal working meetings
- Established over 400 standard definitions for morphology of face and skull, periorbital region, ear, nose and philtrum, lips and mouth, hands and feet
- Results summarized in six papers, and an introduction presented in January 2009 issue *AJMG*

- http://elementsofmorphology.nih.gov/index.cgi



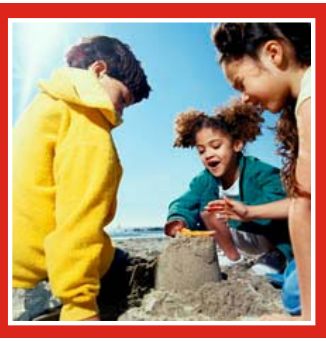


Objective of LOI



- The NCS protocol for the infant physical assessment currently includes a standardized examination and photographs at birth, 6 months and 12 months
- **The focus of this project is to refine a tool for application in the NCS main study, which can be used in field settings to standardize observation and documentation of physical variations and abnormal phenotypic features**

Methods I



- From a list of phenotypic features contained in the elements of morphology, a team of clinical geneticists from 4 NCS sites classified features on the following criteria:
  - Can the feature be captured adequately by photograph?
  - Relevance of the feature
  - Can the feature be captured by physical examination if not captured well by photo?

Methods II

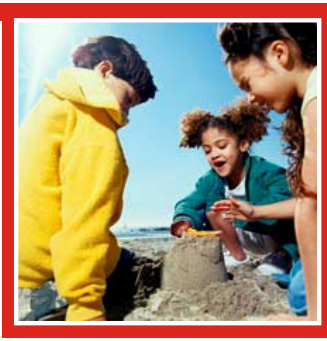
- Develop a minimum standard set of photographic views and standards of quality for those views that will allow for both quantitative and qualitative assessment of a broad range of features based on the results of Methods I
- Develop a physical examination checklist for features selected in Methods I
- Develop a training module for field staff on both of the above

Methods III

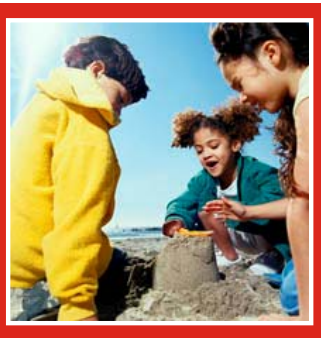
- Test the feasibility and validity of the training module in a pilot study
- Refine and update the DAI and training protocol based on experience in the pilot
- Conduct validation studies with infants at birth, 6 and 12 months of age at each of five participating NCS sites

Results

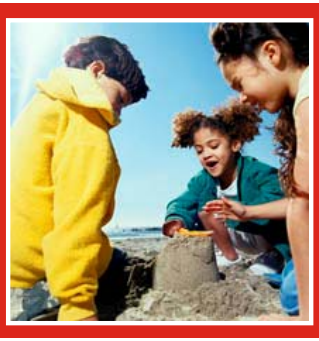
- **Categorization** of best method for assessment of features completed by expert group
- **Photographic Assessment** designed to include 15 standard photographic images (views of craniofacies and hands and feet), and three 10-second videos
- **Physical Assessment** tool developed which is comprised of those 25 features selected from *Elements of Morphology* as relevant, but that are not captured adequately by the Photographic Assessment
- **Online Training Module** that is comprised of detailed instructions for the Photographic Assessment and Physical Assessment is in development and is close to completion



Comprehensive List FEATURES: HEAD AND FACE	FEATURE INCLUSION		DAI CHECKLIST			
	PHOTO CAPTURE (✓)	DAI CHECKLIST (✓)	Feature Relevance (+/-)	Feature Trainability (+/-)	Exam Equal to Photo? (=)	Remove from DAI? (✓)
Brachycephaly	✓					
Dolichocephaly	✓					
Plagiocephaly	✓					
Trigonocephaly	✓					
Turricephaly	✓					
Occiput, Flat	✓					
Occiput, Prominent	✓					
Skull, Cloverleaf	✓					
Frontal Balding		✓	+	-		
Scalp Hair, Sparse	✓					
Hair, Frontal Upsweep		✓	+	-	=	✓
Hair Whorl, Abnormal Number		✓	+	+		
Hair Whorl, Abnormal Position		✓	+	+		
Hairline, High Anterior		✓	+	-		
Hairline, Low Anterior		✓	+	-		
Hairline, Low Posterior	✓					
Widow's Peak	✓					
Face, Broad	✓					
Face, Coarse	✓					
Face, Flat		✓	+	-	=	
Face, Long	✓					
Face, Narrow	✓					
Face, Prematurely Aged	✓					
Face, Round	✓					
Face, Short	✓					
Face, Small	✓					
Face, Square	✓					
Face, Triangle	✓					
Forehead Creases, Vertical	✓					



DAI Photo Guide: Fifteen still photos and 3 videos			
			
1. FaceFront	2. Nares	3. HeadTop	4. HeadBack
			
5. FaceLangle	6. FaceRangle	7. HeadL	8. HeadR
			
9. HandTopR	10. HandTopL	11. PalmR	12. PalmL
			Still photo of each anomaly identified on child, but not already captured in one of these photos.
13. FootTopR	14. FootTopL	15. FeetBottom	OtherAnomalies
			
1. FullbodyDorsal newborn only (10-second video)	2. FullbodyVentral newborn only (10-second video)	3. FrontFaceV (10-second video)	



Ear Pits

An ear pit is a small indentation of skin that occurs anywhere on or near the ear. There are three typical areas that ear pits can be found.

An **auricular pit** is an indentation that is seen on any surface of the portion of the ear that is visible from a lateral view. These pits are most commonly seen on the crus helix (root of the ear) as shown below.

A preauricular pit is an indentation that is seen on the cheek immediately anterior to (in front of) the ear. They are usually located nearest to the crus helix as shown below.

A posterior helical pit is an indentation seen on the posterior side of the helix. They are typically seen along the rim of the helix when the ear is folded forward in order to visualize the posterior ear.

National Children's Study

Dysmorphology Physical Assessment Data Collection Form

Birth

PART A: Administrative

Date: ☐☐ / ☐☐ / ☐☐☐☐ (m m / d d / y y y y)

Time Started: ☐☐ : ☐☐ (h h : m m) ☐ am ☐ pm

Data Collector ID: ☐☐☐☐☐☐☐☐

Time Ended: ☐☐ : ☐☐ (h h : m m) ☐ am ☐ pm

PART B: Participant Information

Child's date of birth: ☐☐ / ☐☐ / ☐☐☐☐ (m m / d d / y y y y)

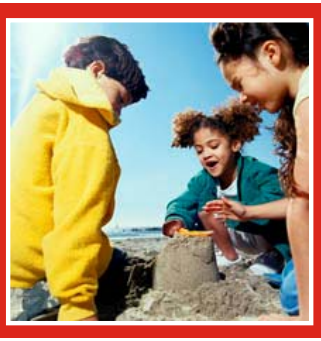
Child's gender: ☐ Male ☐ Female ☐ Both

Part C: Dysmorphology Assessment

Features of the Head and Face

	No	Yes	
1. **Metopic Ridge, Prominent** Comments: _____	☐	☐	
2. **Hair Whorl, Abnormal Number** Comments: _____	☐	☐ →	Specify Number: ☐
3. **Hair Whorl, Abnormal Position** Comments: _____	☐	☐ →	Indicate Position: *Check all that apply.* ☐ Central ☐ Other:_____
4. **Nuchal Skin, Redundant** Comments: _____	☐	☐ →	Indicate Significance: ☐ Mild ☐ Moderate ☐ Marked
5. **Neck Webbing** Comments: _____	☐	☐ →	Indicate Significance: ☐ Mild ☐ Moderate ☐ Marked

NCS Dysmorphology Assessment Instrument: Training Module



Training Module Introduction

Overview

Website last updated 8/17/11

Welcome to the Dysmorphology Assessment Instrument (DAI) Training Module.

IMPORTANT: Please read this ENTIRE Training Module Introduction before proceeding with the Lessons. Important information is shared below regarding the Module organization, computer requirements, and the Terms of Agreement.

This training module:

1. Includes 6 lessons and 6 quizzes.
2. Presents an overview of the Dysmorphology Assessment Instrument (DAI).
3. Introduces the role of the Field Trainee.
4. Provides detailed training on the use of the DAI.

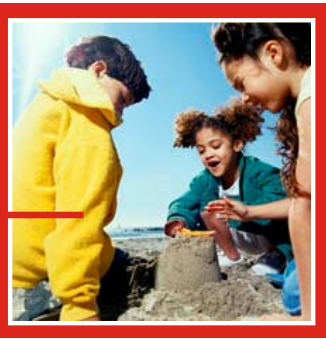
Training Module Objectives

When you have completed this module, you will be able to:

- Describe the DAI.
- Define the role of the Field Examiner.
- Take standardized craniofacial photographs and videos.
- Conduct the physical exam.
- Properly handle data.
- Ensure the safety of the subject.

Topics Below

Next Steps



- Awaiting OMB approval
- Completion of training module
- Completion of web-based interface for secure upload of photographs and physical examination results
- Training sessions at participating sites
- Pilot test, evaluation of quality of photographs and success of training for physical exam
- Field validation studies in a sample of children at birth, 6 and 12 months comparing trainee against “gold standard” expert examiner