

# **Appendix P: Dysmorphology Exam**

Protocol for Conducting Exam

Data Collection form for Exam



## PROTOCOL FOR PHYSICAL AND DYSMORPHOLOGY EXAMINATION (revised 11-14-05)

### Objective

The recommended format is intended to facilitate recording data from anthropometric and dysmorphology examination, and determine if a subject (or cohort control) is likely to have a genetic syndrome. The physical examination will include anthropometrics (height, weight, head circumference) and standardized dysmorphology examination.

### Methods

#### Measures

1. Physical anthropometric measurements according to standards described in training manual.
2. Data will be recorded for physical and dysmorphology examination in the data recording form (see Appendix).
3. Measurement and description of specified features (including face, hands, feet and others) in standardized fashion by use of digital camera and measurement software. Measurements will be recorded in datasheets.

### Procedures

#### Assessment Team

1. Qualified examiner (see below for qualifications) for dysmorphology examination and photography.
2. Pediatric clinician for supervision of physical measurements and recording data of dysmorphology examination
  1. Each center will include an experienced pediatric clinician who will train and supervise the examiner.
  2. Depending on the availability at each site, this clinician might be a Developmental or Behavioral Pediatrician, Child Neurologist, Child Psychiatrist, Pediatric Nurse Practitioner or Pediatric Geneticist.
3. Each site will have access to a Consulting Pediatric Geneticist for assessment and analysis of photographs to determine if a syndrome is likely. Please see Quality Assurance section below.

#### Photographs

1. Photogrammatic digital measurements:
  - a. Use of a digital camera (minimum 2.0 megapixels) and software program for measurement of specified dysmorphic features [web site is <http://www.kuleuven.ac.be/bio/sys/carnoy/> ].
  - b. The centers will assure that the examiner will be trained in the use of the software program.
2. Photographs of child will include:
  - a. Views of face
    - i. Profile (both sides against dark background)
    - ii. Full face portrait
  - b. Each hand with fingers spread
  - c. Feet (without shoes and socks, placed flat against a dark background)
  - d. Ear (length; from photograph – position, rotation morphology)



3. Standards for each photograph will be enumerated in the training manual.

#### Qualified Examiner:

1. **Education:** Master's level (or equivalent degree) candidate preferred, with background in working with pediatric population(s) in a clinical department (e.g., genetics, other pediatric departments) or other research project involving children.
2. **Experience:**
  - a. Previous direct clinical experience in examining children, recording data, under supervision of pediatric clinician.
  - b. Completion of written and videotaped training curriculum according to published standards of measurement.
  - c. Experience observing in a pediatric clinical genetics clinic (e.g, craniofacial clinic or others), working with geneticist and/or genetics counselor for a minimum of 6 sessions.

#### Quality Assurance

To maintain quality and consistent data collection each of the CADDRE centers will:

1. Identify a clinician who will supervise and train the examiner(s), establish reliability, and oversee quality of data collection.
2. Identify a consulting Pediatric Geneticist who will be available to assist with training curriculum and evaluate photographs of subjects to confirm clinical impressions as needed.
3. Maintain a library of standard references (see reference list below).
4. Develop a written and videotaped curriculum for standards of physical measurements and data recording. Sample examinations will be videotaped as part of a curriculum for training examiners.
5. Measures of Quality assurance
  - a. Within the center – options to be considered will include (one or both of the following)
    - i. The clinical team will periodically compare direct clinical measurements by a geneticist or experienced examiner (using sliding anthropometric caliper) with photogrammetric digital measurements. The expectation of agreement (X% of measurement vs. number of trials to establish agreement) will be determined.
    - ii. Duplicate photographs of the same child will be measured in sequence (by the same examiner) and/or by other examiners in the same program. The expectation of agreement X% of measurement vs. number of trials to establish agreement) will be determined.
  - b. Between centers – Interrater reliability measures (between centers) will be completed quarterly, with standards of agreement to be determined.

#### REFERENCES

Hall JG, Froster-Iskenius UG, Alanson JE. *Handbook of Normal Physical Measurements*. NY: Oxford University Press; Jones KL. *Smith's Recognizable Patterns of Human Malformation 5<sup>th</sup> edition*. Philadelphia, PA: WB Saunders and Company, 1989.



Jones KL. *Smith's Recognizable Patterns of Human Malformation, 5<sup>th</sup> Edition*. Philadelphia: W.B. Saunders Company, 1997.

Miles JH, Hillman RE. Value of a clinical morphology examination in autism. *American Journal of Medical Genetics* 91:245-253 (2000).

Rodier PM, Bryson SE, Welch JP. Minor malformations and physical measurements in autism: data from Nova Scotia. *Teratology*. 1997 May;55(5):319-25  
<http://download.interscience.wiley.com/cgi-bin/fulltext?ID=46014&PLACEBO=IE.pdf>

Waldrop, M. & Halverson, C. Minor physical anomalies and hyperactive behaviour in young children. In J. Hellmuth (Eds.), *Exceptional Infant. Studies in Abnormalities* (pp. 343-380). New York: Brunner/Mazel, 1971

**Dysmorphology Exam: Data Collection Form**

**STUDY ID#** \_\_\_\_\_  
 Gender: Male / Female (circle one)  
 Date of Birth: \_\_\_\_\_  
 Chronological Age: \_\_\_\_\_

Date of examination: \_\_\_\_\_  
 Examiner: \_\_\_\_\_  
 Reviewing Physician: \_\_\_\_\_  
 Date of Review: \_\_\_\_\_

**DYSMORPHOLOGY EXAMINATION**

<b>I. Growth parameters</b>	<b>Measurement</b> <i>Note Units</i>	<b>Percentile</b>	<b>COMMENTS</b> <b>(From in person exam)</b>	<b>COMMENTS</b> <b>(From photograph review)</b>
Head circumference <b>(cm)</b>				n/a
Height <b>(cm)</b>				n/a
Weight <b>(kg)</b>				n/a
Inner canthal distance <b>(mm)</b>				
Palpebral fissure length <b>(mm)</b>				

<b>Hand Measurements</b>	<b>Right</b>		<b>Left</b>		<b>COMMENTS</b> <b>(From in person exam)</b>	<b>COMMENTS</b> <b>(From image review)</b>
<i>Using copied image of palmar surface of hand</i>	<b>Size (cm)</b>	<b>%ile</b>	<b>Size (cm)</b>	<b>%ile</b>		
Palm + middle finger						
Palm						
Middle finger						
2 <sup>nd</sup> or Index finger						
4 <sup>th</sup> or ring finger						

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Public reporting burden of this collection of information is estimated to average 15 minutes per response, including the time for reviewing instructions, searching existing data sources, gathering and maintaining the data needed, and completing and reviewing the collection of information. An agency may not conduct or sponsor, and a person is not required to respond to a collection of information unless it displays a currently valid OMB control number. Send comments regarding this burden estimate or any other aspect of this collection of information, including suggestions for reducing this burden to CDC/ATSDR Reports Clearance Officer; 1600 Clifton Road NE, MS D-74, Atlanta, Georgia 30333; ATTN: PRA (0920-XXXX)

<b>II. Minor congenital anomalies</b>		<b>COMMENTS (From in person exam)</b>	<b>COMMENTS (From photograph review)</b>
<b>HEAD</b>			
Frontal Bossing	<input type="checkbox"/> Present <input type="checkbox"/> Absent		
Widow's peak	<input type="checkbox"/> Present <input type="checkbox"/> Absent		
Low hairline (posterior)	<input type="checkbox"/> Present – <b>take photo if present</b> <input type="checkbox"/> Absent		
Double/multiple hair whorl(s)	<input type="checkbox"/> Present <input type="checkbox"/> Absent		
Frontal upsweep	<input type="checkbox"/> Present <input type="checkbox"/> Absent		
Nasolabial fold (at rest)	<input type="checkbox"/> Present <input type="checkbox"/> Absent		
Epicanthal folds	<input type="checkbox"/> Present (full) <input type="checkbox"/> Pseudoepicanthic folds <input type="checkbox"/> Absent		
Nose	<input type="checkbox"/> Bulbous tip <input type="checkbox"/> Upturned <input type="checkbox"/> Wide nasal bridge <input type="checkbox"/> Normal		
Mouth	<input type="checkbox"/> Abnormal philtrum <input type="checkbox"/> Thin lips <input type="checkbox"/> Tented mouth <input type="checkbox"/> Wide mouth <input type="checkbox"/> Normal		

	<b>LEFT</b>	<b>RIGHT</b>	<b>COMMENTS (From in person exam)</b>	<b>COMMENTS (From photograph review)</b>
<b>EARS</b>				
Ear position (low +/-)	<input type="checkbox"/> Low set <input type="checkbox"/> Normal	<input type="checkbox"/> Low set <input type="checkbox"/> Normal		
Ear shape	<input type="checkbox"/> Simple <input type="checkbox"/> Lop shape <input type="checkbox"/> Normal	<input type="checkbox"/> Simple <input type="checkbox"/> Lop shape <input type="checkbox"/> Normal		
Ear shape - helix	<input type="checkbox"/> Folded helix <input type="checkbox"/> Normal	<input type="checkbox"/> Folded helix <input type="checkbox"/> Normal		
Ear shape - helix	<input type="checkbox"/> Notches in helix <input type="checkbox"/> Normal	<input type="checkbox"/> Notches in helix <input type="checkbox"/> Normal		
Ear lobes	<input type="checkbox"/> Adherent <input type="checkbox"/> Normal	<input type="checkbox"/> Adherent <input type="checkbox"/> Normal		
<b>HANDS</b>				
Nails	<input type="checkbox"/> Abnormal – <b>describe</b>	<input type="checkbox"/> Abnormal – <b>describe</b>		

	<input type="checkbox"/> Normal	<input type="checkbox"/> Normal		
Index finger > middle finger	<input type="checkbox"/> Present <input type="checkbox"/> Absent	<input type="checkbox"/> Present <input type="checkbox"/> Absent		
Single transverse crease	<input type="checkbox"/> Present <input type="checkbox"/> Absent	<input type="checkbox"/> Present <input type="checkbox"/> Absent		
Curved 5 <sup>th</sup> finger	<input type="checkbox"/> Present <input type="checkbox"/> Absent	<input type="checkbox"/> Present <input type="checkbox"/> Absent		
<b>FEET</b>				
Nails	<input type="checkbox"/> Abnormal – <b>describe</b> <input type="checkbox"/> Normal	<input type="checkbox"/> Abnormal – <b>describe</b> <input type="checkbox"/> Normal		
2 <sup>nd</sup> & 3 <sup>rd</sup> toes long as great toe	<input type="checkbox"/> Present <input type="checkbox"/> Absent	<input type="checkbox"/> Present <input type="checkbox"/> Absent		
3 <sup>rd</sup> toe longer than second	<input type="checkbox"/> Present <input type="checkbox"/> Absent	<input type="checkbox"/> Present <input type="checkbox"/> Absent		
Syndactyly of toes	# toes _____ <input type="checkbox"/> Present (full) <input type="checkbox"/> Partial <input type="checkbox"/> Absent (normal)	# toes _____ <input type="checkbox"/> Present (full) <input type="checkbox"/> Partial <input type="checkbox"/> Absent (normal)		
Short toes	<input type="checkbox"/> Present <input type="checkbox"/> Absent	<input type="checkbox"/> Present <input type="checkbox"/> Absent		
Toe spacing	<input type="checkbox"/> Normal <input type="checkbox"/> Wide spaced	<input type="checkbox"/> Normal <input type="checkbox"/> Wide spaced		
Toe walking	<input type="checkbox"/> Present <input type="checkbox"/> Absent	<input type="checkbox"/> Present <input type="checkbox"/> Absent		
<b>SKIN</b>				
Cutaneous findings suggestive of neurocutaneous disorder - <b>ambient light</b>	<input type="checkbox"/> Café au lait <input type="checkbox"/> Ash leaf spot <input type="checkbox"/> Linear nevus(i) <input type="checkbox"/> Adenoma sebaceum	<b>Record number, location and measurement(s) :</b>		
Cutaneous findings with <b>Woods Lamp illumination</b>	<input type="checkbox"/> Café au lait <input type="checkbox"/> Ash leaf spot <input type="checkbox"/> Linear nevus(i) <input type="checkbox"/> Adenoma sebaceum	<b>Record number, location and measurement(s) :</b>		

Other Observations:





**Photographs taken/ comments (Examiner please check which ones)**

- Face (frontal)
- Back of head (for hairline)
- Profile – left
- Profile – right
- Hand (volar or non-palm side) – left
- Hand (volar or non-palm side) – right
- Foot – left
- Foot – right
- Skin; note which parts of body: \_\_\_\_\_

\_\_\_\_\_  
Signature of Examiner

\_\_\_\_\_  
Signature of Reviewer