Appendix P: Dysmorphology Exam

Protocol for Conducting Exam

Data Collection form for Exam

Center for Autism and Developmental Disabilities Research and Epidemiology

Pennsylvania CADDRE

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)

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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 5th edition*. Philadelphia, PA: WB Saunders and Company, 1989.

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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
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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

Form Approved
OMB NO. _____
Exp. Date _____

STUDY ID# _____ Date of examination: _____
Gender: Male / Female (*circle one)* Examiner: _____
Date of Birth: _____ Reviewing Physician:_____
Chronological Age: _____ Date of Review:_____

DYSMORPHOLOGY EXAMINATION

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

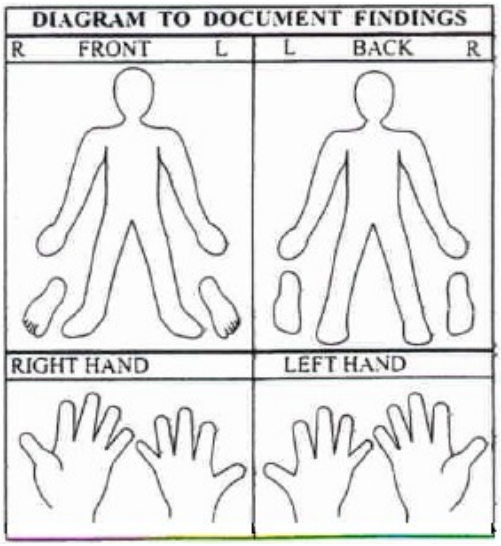
Hand Measurements	**Right**		**Left**		**COMMENTS (From in person exam)**	**COMMENTS (From image review)**
Using copied image of palmar surface of hand	**Size (cm)**	**%ile**	**Size (cm)**	**%ile**		
Palm + middle finger						
Palm						
Middle finger						
2nd or Index finger						
4th or ring finger						

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

	LEFT	**RIGHT**	**COMMENTS (From in person exam)**	**COMMENTS (From photograph review)**
EARS				
Ear position (low +/=)	☐ Low set ☐ Normal	☐ Low set ☐ Normal		
Ear shape	☐ Simple ☐ Lop shape ☐ Normal	☐ Simple ☐ Lop shape ☐ Normal		
Ear shape - helix	☐ Folded helix ☐ Normal	☐ Folded helix ☐ Normal		
Ear shape - helix	☐ Notches in helix ☐ Normal	☐ Notches in helix ☐ Normal		
Ear lobes	☐ Adherent ☐ Normal	☐ Adherent ☐ Normal		
HANDS				
Nails	☐ Abnormal – **describe**	☐ Abnormal – **describe**		

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

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