SEED Dysmorphology Exam

QC (yes/no) Name:_____   STUDY ID#_____

Gender (*circle one*):   Male / Female   Date of examination: _____
<mark>Mom & Dad Race/Ethnicity:</mark>_____   Examiner: _____
Date of Birth: _____   Reviewing Physician:_____
Chronological Age: _____   Date of Review:_____

Scale QC - Use object of known weight Record weight here (including units):	Initial Scale reading with object	COMMENTS (Type of object used)

PARENT MEASUREMENTS *(Do not count as a physical anomaly for dysmorphology classification)*

Biological MOTHER	Measurement *Note Units*	Percentile	Exam Comments
Height (can be reported)		n/a	• unreliable – reason_____
Head circumference (cm)			• unreliable – reason_____
Biological FATHER	**Measurement** *Note Units*	**Percentile**	**Exam Comments**
Height (can be reported)		n/a	• unreliable – reason_____
Head circumference (cm)			• unreliable – reason_____

DYSMORPHOLOGY EXAMINATION

I. Growth Parameters	Measurement	Percentile	Exam Comments
Height (cm)			• unreliable – reason_____
Weight (kg)			• unreliable – reason_____
Head Circumference (cm)			• unreliable – reason_____
BMI and Percentile			• unreliable – reason_____

EYES	Measurement	Percentile	Photo Comments
Interpupillary distance (mm)		*(not from ABASE – need to get %ile from plotting on paper)*	Internal measurement (mm) _____ Photo #_____ • unreliable – reason_____
Inner canthal distance (mm)		*Provided by ABASE*	Internal measurement (mm) _____ Photo #_____ • unreliable – reason_____

	Right PFL		Left PFL	
	Measurement	**Photo Comments**	**Measurement**	**Photo Comments**
Palpebral fissure length (mm)		Internal measurement (mm) _____ Photo #_____ • unreliable – reason_____		Internal measurement (mm) _____ Photo #_____ • unreliable – reason_____

MOUTH	Measurement	Photo Comments
Philtrum (mm)		Internal measurement (mm) _____ Photo #_____

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		• unreliable – reason_____

EARS	**Right Ear**			**Left Ear**		
	Measure	**Percentile**	**Photo Comments**	**Measure**	**Percentile**	**Photo Comments**
Length **(mm)**			Internal measurement (mm) _____ Photo #_____ • unreliable – reason_____			Internal measurement (mm) _____ Photo #_____ • unreliable – reason_____

HANDS *From Scanner*	**Right Hand**		**Left Hand**	
	Measurement	**Scan Comments**	**Measurement**	**Scan Comments**
2nd or Index finger **(mm)**		Internal measurement (mm) _____ Scan #_____ • unreliable – reason_____		Internal measurement (mm) _____ Scan #_____ • unreliable – reason_____
4th or ring finger **(mm)**		• unreliable – _____		• unreliable – _____
Digit ratio: 2nd/4th		/ / / / / / / / / / /		/ / / / / / / / / /
	Measurement	**Percentile**	**Measurement**	**Percentile**
Middle finger **(mm)**		• unreliable – _____		• unreliable – _____
Palm **(mm)**		• unreliable – _____		• unreliable – _____
Total hand length (Middle finger + Palm) **(mm)**		• unreliable – _____		• unreliable – _____

FEET *From Paper Markings*	**Right Foot**		**Left Foot**	
	Measurement	**Percentile**	**Measurement**	**Percentile**
Foot Length **(cm)**		• unreliable – _____		• unreliable – _____

II. Minor congenital anomalies *(take additional photos if abnormal)*		**Exam Comments**	**Photo Comments**
HEAD	☐ Normal ☐ Abnormal ☐ Brachycephaly ☐ Dolichocephaly/scaphocephaly ☐ Macrocephaly ☐ Microcephaly ☐ Plagiocephaly/asymmetrical skull ☐ Other - *describe in comments*		Photo #_____ • unreliable – _____
FOREHEAD	☐ Normal ☐ Abnormal ☐ Prominent forehead/frontal bossing		Photo #_____ • unreliable – _____

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	☐ Wide forehead ☐ Narrow forehead/temporal narrowing ☐ Other - *describe in comments*		

HAIR	☐ Normal
	☐ Abnormal
	   ☐ High frontal hairline
	   ☐ Low frontal hairline
	   ☐ Widow's peak
	   ☐ Frontal upsweep
	   ☐ Double/multiple hair whorl(s)
	   ☐ Low hairline (posterior)
	   ☐ Hirsutism
	   ☐ Other - *describe in comments*
			Photo #_____
			• unreliable – _____

FACE	☐ Normal
	☐ Abnormal
	   ☐ Asymmetric face
	   ☐ Coarse facial features
	   ☐ Flat face
	   ☐ Mid-face hypoplasia
	   ☐ Small face
	   ☐ Thin/long face
	   ☐ Triangular face
	   ☐ Absent nasolabial fold (at rest)
	   ☐ Pointed chin
	   ☐ Prominent mandible/prognathism
	   ☐ Small mandible/micrognathia
	☐ Other - *describe in comments*
			Photo #_____
			• unreliable – _____

EYES	☐ Normal
	☐ Abnormal
	   ☐ Nodular eyelids
	   ☐ Ptosis of eyelids
	   ☐ Epicanthal folds (full)
	   ☐ Palpebral fissure slant (up)
	   ☐ Palpebral fissure slant (down)
	   ☐ Short palpebral fissures (measurement)
	   ☐ Wide/long palpebral fissures (measurement)
	   ☐ Periorbital skin, general abnormalities
	   ☐ Fullness of peri-orbital region
	   ☐ Deep-set eyes
	   ☐ Hypertelorism (measurement)
	   ☐ Hypotelorism (measurement)
	   ☐ Prominent eyes/proptosis
	   ☐ Other - *describe in comments*
			Photo #_____
			• unreliable – _____

EYEBROWS	☐ Normal ☐ Abnormal ☐ Sparse/decreased eyebrows		Photo #_____ • unreliable – _____

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	☐ Medial eyebrow flare ☐ Synophrys ☐ Thick eyebrows ☐ Other - *describe in comments*		

NOSE SIZE	☐ Normal
	☐ Abnormal
	    ☐ Large/long nose (by impression)
	    ☐ Small/short nose (by impression)
	    ☐ Other - *describe in comments*
			Photo #_____
			• unreliable – _____

NOSE STRUCTURE	☐ Normal
	☐ Abnormal
	    ☐ Depressed/flat nasal bridge
	    ☐ High/prominent nasal bridge
	    ☐ Short nasal septum
	    ☐ Broad nasal tip
	    ☐ Bulbous tip
	    ☐ Flat nose
	    ☐ Pinched nose
	    ☐ Upturned
	    ☐ Broad base to nose
	    ☐ Wide nasal bridge
	    ☐ Other - *describe in comments*
			Photo #_____
			• unreliable – _____

MOUTH & LIPS	☐ Normal
	☐ Abnormal
	    ☐ Long philtrum (measurement)
	    ☐ Short philtrum (measurement)
	    ☐ Simple/absent philtrum (by impression)
	    ☐ Wide philtrum (by impression)
	    ☐ Macrostomia
	    ☐ Microstomia
	    ☐ Cupid bow shape of mouth
	    ☐ Open mouth appearance
	    ☐ Thin lips
	    ☐ Thin upper lip
	    ☐ Cleft upper lip (non-midline)
	    ☐ Tented mouth
	    ☐ Wide mouth
	    ☐ Down-turned corners of the mouth
	    ☐ Prominent/everted lower lip
	    ☐ Other - *describe in comments*
			Photo #_____
			• unreliable – _____

TEETH	☐ Normal
	☐ Abnormal
	    ☐ Enamel abnormalities
	    ☐ Irregular or crowded teeth
	    ☐ Abnormally shaped teeth
	    ☐ Small teeth
	    ☐ Wide-spaced teeth
	☐ Other - *describe in comments*
			Photo #_____
			• unreliable – _____

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	R	L			
EARS Position, rotation, shape, & lobes	☐ ☐ ☐ ☐ ☐ ☐ ☐ ☐ ☐ ☐ ☐ ☐ ☐ ☐ ☐ ☐ ☐	☐ ☐ ☐ ☐ ☐ ☐ ☐ ☐ ☐ ☐ ☐ ☐ ☐ ☐ ☐ ☐ ☐	**Normal** **Abnormal** Asymmetric ears Dysplastic ears Size – Large (by measurement) Size – Small (by measurement) Position - Low set Rotation - Posteriorly Shape - Simple Shape – Lop Shape – Cupped Shape – Protuberant ears Helix - Folded Helix - Notches in helix Helix – Prominent helix Helix - Crumpled helix Helix/Lobe -Auricular pits/fistulas Lobe – Adherent/attached Other - *describe in comments*		Photos #_____ • unreliable – _____

	R	L			
HANDS Palm and fingers	☐ ☐ ☐ ☐ ☐ ☐ ☐ ☐ ☐ ☐ ☐ ☐ ☐ ☐ ☐	☐ ☐ ☐ ☐ ☐ ☐ ☐ ☐ ☐ ☐ ☐ ☐ ☐ ☐ ☐	Normal Abnormal Large hands (measurement) Small hands (measurement) Camptodactyly Clinodactyly Curved 5th finger Tapering fingers Short/hypoplastic metacarpals Short phalanges Wide phalanges Long phalanges Broad thumbs Index finger > middle finger Single transverse crease Other – *describe in comments*		Photos #_____ • unreliable – _____
NAILS (indicate hands and/or feet)	☐ ☐ ☐ ☐ ☐	☐ ☐ ☐ ☐ ☐	Normal Abnormal Hyperplastic, Includes ridged nails Hyperconvex/clubbed		Photos #_____ • unreliable – _____

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	R	L		Exam Comments	Photo Comments
	☐	☐	nails		
	☐	☐	Small/hypoplastic/deep set		
			nails		
			Short nails		
			Other – *describe in comments*		

	R	L		Exam Comments	Photo Comments
FEET Structure and toes	☐	☐	Normal		Photos #_____ • unreliable – _____
	☐	☐	Abnormal		
	☐	☐	Large feet		
	☐	☐	Small feet		
	☐	☐	Wide feet		
	☐	☐	Club foot, varus		
	☐	☐	Hallux valgus		
	☐	☐	2nd & 3rd toes as long as 1st toe		
	☐	☐	3rd toe longer than 2nd toe		
	☐	☐	Syndactyly of toes (full)		
			Syndactyly of toes (partial)		
			R - # toes _____		
	☐	☐	L - # toes _____		
	☐	☐	Short toes		
	☐	☐	Wide spaced toes		
	☐	☐	Broad toes		
	☐	☐	Over-riding toes (includes clinodactyly)		
	☐	☐	Camptodactyly/hammer toes		
	☐	☐	Toes, other		
			Other - *describe in comments*		

SKIN	☐ Normal ☐ Abnormal ☐ Cutaneous findings using **ambient light** ☐ Cutaneous findings with **Woods Lamp illumination** ☐ Other - *describe in comments*	**Record number, location, color, raised/flat and measurement(s):**	**Record number, location, color, raised/flat and measurement(s):** Photo #_____ • unreliable – _____
GAIT	☐ Normal ☐ Abnormal ☐ Toe walking (observed only) ☐ Other - *describe in comments*	*(Do not count as a physical anomaly for dysmorphology classification)*	

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OTHER OBSERVATIONS/COMMENTS:

PHOTOGRAPHS

Examiner – Take at least one photo of each listed and mark below

- ☐ Head – Anterior (face) – With Reference & for hairline
- ☐ Head – Anterior (face) – smiling for teeth
- ☐ Head – Posterior – For Hairline
- ☐ Head – Left Lateral – Profile (include entire head in shot)
- ☐ Head/Ear – Left Lateral – With Reference
- ☐ Head – Right Lateral – Profile (include entire head in shot)
- ☐ Head/Ear – Right Lateral – With Reference
- ☐ Head – ¾ view – No reference needed
- ☐ Head – hair whorls – no reference

- ☐ Hand – Left Dorsal –Non-palm Side (single hand in photo)
- ☐ Hand – Right Dorsal – Non-palm Side (single hand in photo)
- ☐ Hand (scan) – Right Ventral – Palm Side
- ☐ Hand (scan) – Left Ventral – Palm Side

- ☐ Foot – Left Dorsal – Top of Foot (single foot in photo)
- ☐ Foot – Right Dorsal – Top of Foot (single foot in photo)

- ☐ Additional photos - Note number of photos, which abnormality, and part of body:
  _____

CLINIC INTAKE QUESTIONS:

1) Was [CHILD] born with any problems in the structure of his/her body or organs (also know as birth defects)?
- ☐ No
- ☐ Yes - describe
  _____

2) Has [CHILD] had any corrective surgeries? This includes surgeries to repair findings in the abdominal or genital region (such as hernias)?
- ☐ No
- ☐ Yes - describe
  _____

3) Does [CHILD] have a clinical diagnosis of a syndrome?
- ☐ No
- ☐ Possible Dx*:_____
- ☐ Yes Dx*: _____

4) Has [CHILD] had a genetics evaluation, blood tests, or been seen by a genetic counselor?
- ☐ No
- ☐ Yes* Reason/Results:_____

*(*IF THEY HAVE NOT DONE SO ALREADY, PLEASE BE SURE TO ASK FOR THE FAMILY TO **SIGN A HIPAA MEDICAL RECORD RELEASE** FOR THIS PROVIDER.)*

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_____
_____

_____
Signature of Examiner

_____
Signature of Classifying Clinician (genetics)

_____
Signature of QC Supervisor *(if applicable)*

Skin Findings Classification System

Measurements	Criteria for Defining Feature as Dysmorphic	
	Lower limit	**Upper limit**
BMI?		
Height	≤ 10th %ile	≥ 97th %ile
Weight	≤ 3rd %ile	≥ 97th %ile
Head circumference	≤ 2nd %ile	≥ 98th %ile
Interpupillary distance	≤ 3rd %ile	≥ 97th %ile
Inner canthal distance	≤ 3rd %ile	≥ 97th %ile
Palpebral fissure length	≤ 5th %ile	≥ 95th %ile
Philtrum length	≤ 3rd %ile	≥ 97th %ile
Ear length	≤ 3rd %ile	≥ 97th %ile
Middle finger length	≤ 3rd %ile	≥ 97th %ile
Palm length	≤ 3rd %ile	≥ 97th %ile
Total hand length	≤ 3rd %ile	≥ 97th %ile
Foot length	≤ 3rd %ile	≥ 97th %ile

Hypopigmented macules
Count as dysmorphic if:
--the number is ≥ to 5 OR
--the diameter of any one macule is >50 mm

Café-au-lait spots
Count as dysmorphic if:
--the number is >= to 6 or
--the diameter of any one spot is >50 mm

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Version 1-13-09 SNC

GENETICISTS – please fill out pages 8-9

1) CLASSIFYING CLINICIAN IMPRESSION – circle one *(This is based on clinician's Gestalt and is not based on any Miles classification system)*. **Any suspected syndrome features should be listed under "List Features."**

DYSMORPHIC   NONDYSMORPHIC

_____

_____

2) Known Genetic Syndrome or Chromosome Abnormality?

NO   YES   **If Yes, Specify:**_____

3) Major malformation present?
   If Yes, check additional information box and list features under OLD Miles list.

NO   YES ⟶ ☐ Single major malformation
          ☐ Multiple major malformations (>1)

4) OLD Miles ALGORITHM RESULTS

List Features:

Final Impression by GENETICIST *(Check one box below)*	List Features
CLASSIFICATION	_____
☐ **Normal – nondysmorphic** (3 or less features)	_____
☐ **Equivocal** (4-5 features)	_____
☐ **Abnormal – dysmorphic** (6 or more features)	_____

5) NEW Miles ALGORITHM RESULTS

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Your decision flow must start at "ears" (pink box) and then proceed through a path until you reach "dysmorphic" or "non-dysmorphic." **Circle your answers for each box that you choose.** Your decision flow cannot start at "nose size," "nails" or "hair growth pattern." Your decision flow must follow until you reach "dysmorphic" or "non-dysmorphic" on the chart.

Figure 3.
Autism Dysmorphology
Measure (ADM)
Scoring Algorithm

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