

SEED Dysmorphology Exam

QC (yes/no) Name: _____

STUDY ID# _____

Gender (circle one): Male / Female

Date of examination: _____

Mom & Dad Race/Ethnicity: _____

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 <i>Note Units</i>	Percentile	Exam Comments
Height (can be reported)		n/a	• unreliable – reason _____
Head circumference (cm)			• unreliable – reason _____
Biological FATHER	Measurement <i>Note Units</i>	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)		<i>(not from ABASE – need to get %ile from plotting on paper)</i>	Internal measurement (mm) _____ Photo # _____ • unreliable – reason _____		
Inner canthal distance (mm)		<i>Provided by ABASE</i>	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 # _____			

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

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

FEET <i>From Paper Markings</i>	Right Foot		Left Foot	
	Measurement	Percentile	Measurement	Percentile
Foot Length (cm)		• unreliable – _____		• unreliable – _____

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

	<input type="checkbox"/> Wide forehead <input type="checkbox"/> Narrow forehead/temporal narrowing <input type="checkbox"/> Other - <i>describe in comments</i>		
HAIR	<input type="checkbox"/> Normal <input type="checkbox"/> Abnormal <input type="checkbox"/> High frontal hairline <input type="checkbox"/> Low frontal hairline <input type="checkbox"/> Widow's peak <input type="checkbox"/> Frontal upsweep <input type="checkbox"/> Double/multiple hair whorl(s) <input type="checkbox"/> Low hairline (posterior) <input type="checkbox"/> Hirsutism <input type="checkbox"/> Other - <i>describe in comments</i>		Photo # _____ • unreliable - _____
FACE	<input type="checkbox"/> Normal <input type="checkbox"/> Abnormal <input type="checkbox"/> Asymmetric face <input type="checkbox"/> Coarse facial features <input type="checkbox"/> Flat face <input type="checkbox"/> Mid-face hypoplasia <input type="checkbox"/> Small face <input type="checkbox"/> Thin/long face <input type="checkbox"/> Triangular face <input type="checkbox"/> Absent nasolabial fold (at rest) <input type="checkbox"/> Pointed chin <input type="checkbox"/> Prominent mandible/prognathism <input type="checkbox"/> Small mandible/micrognathia <input type="checkbox"/> Other - <i>describe in comments</i>		Photo # _____ • unreliable - _____
EYES	<input type="checkbox"/> Normal <input type="checkbox"/> Abnormal <input type="checkbox"/> Nodular eyelids <input type="checkbox"/> Ptosis of eyelids <input type="checkbox"/> Epicanthal folds (full) <input type="checkbox"/> Palpebral fissure slant (up) <input type="checkbox"/> Palpebral fissure slant (down) <input type="checkbox"/> Short palpebral fissures (measurement) <input type="checkbox"/> Wide/long palpebral fissures (measurement) <input type="checkbox"/> Periorbital skin, general abnormalities <input type="checkbox"/> Fullness of peri-orbital region <input type="checkbox"/> Deep-set eyes <input type="checkbox"/> Hypertelorism (measurement) <input type="checkbox"/> Hypotelorism (measurement) <input type="checkbox"/> Prominent eyes/proptosis <input type="checkbox"/> Other - <i>describe in comments</i>		Photo # _____ • unreliable - _____
EYEBROWS	<input type="checkbox"/> Normal <input type="checkbox"/> Abnormal <input type="checkbox"/> Sparse/decreased eyebrows		Photo # _____ • unreliable - _____

	<input type="checkbox"/> Medial eyebrow flare <input type="checkbox"/> Synophrys <input type="checkbox"/> Thick eyebrows <input type="checkbox"/> Other - <i>describe in comments</i>		
NOSE SIZE	<input type="checkbox"/> Normal <input type="checkbox"/> Abnormal <input type="checkbox"/> Large/long nose (by impression) <input type="checkbox"/> Small/short nose (by impression) <input type="checkbox"/> Other - <i>describe in comments</i>		Photo # _____ • unreliable – _____
NOSE STRUCTURE	<input type="checkbox"/> Normal <input type="checkbox"/> Abnormal <input type="checkbox"/> Depressed/flat nasal bridge <input type="checkbox"/> High/prominent nasal bridge <input type="checkbox"/> Short nasal septum <input type="checkbox"/> Broad nasal tip <input type="checkbox"/> Bulbous tip <input type="checkbox"/> Flat nose <input type="checkbox"/> Pinched nose <input type="checkbox"/> Upturned <input type="checkbox"/> Broad base to nose <input type="checkbox"/> Wide nasal bridge <input type="checkbox"/> Other - <i>describe in comments</i>		Photo # _____ • unreliable – _____
MOUTH & LIPS	<input type="checkbox"/> Normal <input type="checkbox"/> Abnormal <input type="checkbox"/> Long philtrum (measurement) <input type="checkbox"/> Short philtrum (measurement) <input type="checkbox"/> Simple/absent philtrum (by impression) <input type="checkbox"/> Wide philtrum (by impression) <input type="checkbox"/> Macrostomia <input type="checkbox"/> Microstomia <input type="checkbox"/> Cupid bow shape of mouth <input type="checkbox"/> Open mouth appearance <input type="checkbox"/> Thin lips <input type="checkbox"/> Thin upper lip <input type="checkbox"/> Cleft upper lip (non-midline) <input type="checkbox"/> Tented mouth <input type="checkbox"/> Wide mouth <input type="checkbox"/> Down-turned corners of the mouth <input type="checkbox"/> Prominent/everted lower lip <input type="checkbox"/> Other - <i>describe in comments</i>		Photo # _____ • unreliable – _____
TEETH	<input type="checkbox"/> Normal <input type="checkbox"/> Abnormal <input type="checkbox"/> Enamel abnormalities <input type="checkbox"/> Irregular or crowded teeth <input type="checkbox"/> Abnormally shaped teeth <input type="checkbox"/> Small teeth <input type="checkbox"/> Wide-spaced teeth <input type="checkbox"/> Other - <i>describe in comments</i>		Photo # _____ • unreliable – _____

	R	L			
EARS Position, rotation, shape, & lobes	<input type="checkbox"/>	<input type="checkbox"/>	Normal		Photos # _____ • unreliable – _____
	<input type="checkbox"/>	<input type="checkbox"/>	Abnormal		
	<input type="checkbox"/>	<input type="checkbox"/>	Asymmetric ears		
	<input type="checkbox"/>	<input type="checkbox"/>	Dysplastic ears		
	<input type="checkbox"/>	<input type="checkbox"/>	Size – Large (by		
	<input type="checkbox"/>	<input type="checkbox"/>	measurement)		
	<input type="checkbox"/>	<input type="checkbox"/>	Size – Small (by		
	<input type="checkbox"/>	<input type="checkbox"/>	measurement)		
	<input type="checkbox"/>	<input type="checkbox"/>	Position - Low set		
	<input type="checkbox"/>	<input type="checkbox"/>	Rotation - Posteriorly		
	<input type="checkbox"/>	<input type="checkbox"/>	Shape - Simple		
	<input type="checkbox"/>	<input type="checkbox"/>	Shape – Lop		
	<input type="checkbox"/>	<input type="checkbox"/>	Shape – Cupped		
	<input type="checkbox"/>	<input type="checkbox"/>	Shape – Protuberant ears		
	<input type="checkbox"/>	<input type="checkbox"/>	Helix - Folded		
	<input type="checkbox"/>	<input type="checkbox"/>	Helix - Notches in helix		
	<input type="checkbox"/>	<input type="checkbox"/>	Helix – Prominent helix		
	<input type="checkbox"/>	<input type="checkbox"/>	Helix - Crumpled		
<input type="checkbox"/>	<input type="checkbox"/>	helix			
<input type="checkbox"/>	<input type="checkbox"/>	Helix/Lobe -Auricular			
		pits/fistulas			
		Lobe – Adherent/attached			
		Other - <i>describe in</i>			
		<i>comments</i>			
	R	L			
HANDS Palm and fingers	<input type="checkbox"/>	<input type="checkbox"/>	Normal		Photos # _____ • unreliable – _____
	<input type="checkbox"/>	<input type="checkbox"/>	Abnormal		
	<input type="checkbox"/>	<input type="checkbox"/>	Large hands (measurement)		
	<input type="checkbox"/>	<input type="checkbox"/>	Small hands (measurement)		
	<input type="checkbox"/>	<input type="checkbox"/>	Camptodactyly		
	<input type="checkbox"/>	<input type="checkbox"/>	Clinodactyly		
	<input type="checkbox"/>	<input type="checkbox"/>	Curved 5 th finger		
	<input type="checkbox"/>	<input type="checkbox"/>	Tapering fingers		
	<input type="checkbox"/>	<input type="checkbox"/>	Short/hypoplastic		
	<input type="checkbox"/>	<input type="checkbox"/>	metacarpals		
	<input type="checkbox"/>	<input type="checkbox"/>	Short phalanges		
	<input type="checkbox"/>	<input type="checkbox"/>	Wide phalanges		
	<input type="checkbox"/>	<input type="checkbox"/>	Long phalanges		
	<input type="checkbox"/>	<input type="checkbox"/>	Broad thumbs		
	<input type="checkbox"/>	<input type="checkbox"/>	Index finger > middle		
	<input type="checkbox"/>	<input type="checkbox"/>	finger		
	<input type="checkbox"/>	<input type="checkbox"/>	Single transverse crease		
			Other –		
		<i>describe in comments</i>			
NAILS (indicate hands and/or feet)	<input type="checkbox"/>	<input type="checkbox"/>	Normal		Photos # _____ • unreliable – _____
	<input type="checkbox"/>	<input type="checkbox"/>	Abnormal		
	<input type="checkbox"/>	<input type="checkbox"/>	Hyperplastic,		
	<input type="checkbox"/>	<input type="checkbox"/>	Includes ridged		
	<input type="checkbox"/>	<input type="checkbox"/>	nails		
<input type="checkbox"/>	<input type="checkbox"/>	Hyperconvex/clubbed			

	<input type="checkbox"/>	<input type="checkbox"/>	nails Small/hypoplastic/deep set nails Short nails Other – <i>describe in comments</i>		
	R	L		Exam Comments	Photo Comments
FEET Structure and toes	<input type="checkbox"/>	<input type="checkbox"/>	Normal		Photos # _____ • unreliable – _____
	<input type="checkbox"/>	<input type="checkbox"/>	Abnormal		
	<input type="checkbox"/>	<input type="checkbox"/>	Large feet		
	<input type="checkbox"/>	<input type="checkbox"/>	Small feet		
	<input type="checkbox"/>	<input type="checkbox"/>	Wide feet		
	<input type="checkbox"/>	<input type="checkbox"/>	Club foot, varus		
	<input type="checkbox"/>	<input type="checkbox"/>	Hallux valgus		
	<input type="checkbox"/>	<input type="checkbox"/>	2 nd & 3 rd toes as long as 1 st		
	<input type="checkbox"/>	<input type="checkbox"/>	toe		
	<input type="checkbox"/>	<input type="checkbox"/>	3 rd toe longer than 2 nd toe		
	<input type="checkbox"/>	<input type="checkbox"/>	Syndactyly of toes (full)		
	<input type="checkbox"/>	<input type="checkbox"/>	Syndactyly of toes (partial)		
	<input type="checkbox"/>	<input type="checkbox"/>	R - # toes _____		
	<input type="checkbox"/>	<input type="checkbox"/>	L - # toes _____		
	<input type="checkbox"/>	<input type="checkbox"/>	Short toes		
	<input type="checkbox"/>	<input type="checkbox"/>	Wide spaced toes		
	<input type="checkbox"/>	<input type="checkbox"/>	Broad toes		
	<input type="checkbox"/>	<input type="checkbox"/>	Over-riding toes (includes clinodactyly)		
	<input type="checkbox"/>	<input type="checkbox"/>	Camptodactyly/hammer toes		
	<input type="checkbox"/>	<input type="checkbox"/>	Toes, other		
	<input type="checkbox"/>	<input type="checkbox"/>	Other - <i>describe in comments</i>		
SKIN	<input type="checkbox"/>	<input type="checkbox"/>	Normal	Record number, location, color, raised/flat and measurement(s):	Record number, location, color, raised/flat and measurement(s): Photo # _____ • unreliable – _____
	<input type="checkbox"/>	<input type="checkbox"/>	Abnormal		
	<input type="checkbox"/>	<input type="checkbox"/>	Cutaneous findings using ambient light		
	<input type="checkbox"/>	<input type="checkbox"/>	Cutaneous findings with Woods Lamp illumination		
	<input type="checkbox"/>	<input type="checkbox"/>	Other - <i>describe in comments</i>		
GAIT	<input type="checkbox"/>	<input type="checkbox"/>	Normal	<i>(Do not count as a physical anomaly for dysmorphology classification)</i>	
	<input type="checkbox"/>	<input type="checkbox"/>	Abnormal		
	<input type="checkbox"/>	<input type="checkbox"/>	Toe walking (observed only)		
	<input type="checkbox"/>	<input type="checkbox"/>	Other - <i>describe in comments</i>		

OTHER OBSERVATIONS/COMMENTS:

Empty box for other observations and 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.)

STUDY ID# _____
 CLASSIFYING CLINICIAN: _____

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

 Signature of Examiner

 Signature of Classifying Clinician (genetics)

 Signature of QC Supervisor (if applicable)

Skin Findings Classification System

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

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

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**

Final Impression by GENETICIST (Check one box below)
CLASSIFICATION
<input type="checkbox"/> Normal – nondysmorphic (3 or less features)
<input type="checkbox"/> Equivocal (4-5 features)
<input type="checkbox"/> Abnormal – dysmorphic (6 or more features)

List Features:

5) **NEW Miles ALGORITHM RESULTS**

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.

