

Dysmorphology Core Manual for Pediatricians

The neonatologist will be contacted by the study coordinator to inform him/her that a child, either subject or control, who is in the study, has been born. The status of the child, either subject or control, will not be provided by the study coordinator and the neonatologist must not determine if the child is a case or control until after completion of the physical examination, completion of the Dysmorphology Core Physical Examination form. This includes having checked in question 63 “YES” (the child has FAS) or “NO” (the child does not have FAS) or “Deferred”. The neonatologist can then determine from the study coordinator if the child is a case or control.

Each child in the study will be examined on the third day after birth or any anytime up to 1 month if the child is kept in the nursery until that age.

Every question on the Dysmorphology Core Physical Examination Form must be completed.

1. Identification number – This will be provided by the study coordinator and will be the same number as that given to the child’s mother.
2. A check must be made to indicate if the examination has been completed by the Pediatrician/Neonatologist or by the expert (Ken Jones, Luther Robinson, Gene Hoyme or Miguel del Campo).
3. The last name of the examiner must be provided.
4. The exact date of the examination must be indicated in the format dd/mm/yy.
5. The patient’s first and last name must be provided.
6. The patient’s gender (male or female) must be indicated by a check mark.
7. The child’s birth date must be indicated (dd/mm/yy).
8. Number of completed weeks of gestation must be provided.
9. If less than 1 year of age, a check mark must be made to indicate if the child was born prior to 37 weeks of gestation. That means if the child was born at 36 weeks 6 days or before you need to check to check “YES”. If the child was born at 37 weeks or greater, you check “NO”.
10. If the answer to question 9 is “NO”, the child’s current age should be indicated in completed months and weeks. For example, if the child was born after 37 weeks and is 3 days old, his age will be 0 months 0 weeks, move on to question 14.
11. If the answer to question 9 is “YES”, it must be determined if the child’s age at the time of the examination is prior to the expected date of confinement (EDC = 40 weeks of gestation) or if the examination takes place after the EDC (EDC = 40 weeks of gestation). For example: If the child was born at 32 weeks gestation and the examination takes place 2 weeks after birth, the examination would take place prior to the EDC. If, on the other hand, the child was born at 36 weeks gestation and examined 6 weeks after birth, the examination would take place after the EDC. If the child is examined prior to

the EDC, the age should be given in gestational weeks. For example, if the child was born at 32 weeks gestation and the examination takes place 2 weeks after delivery, the completed gestational weeks at exam would be 34 weeks (32 weeks plus 2 weeks).

12. If the child is examined after the EDC, and was born prior to 37 week gestation, an adjusted age should be calculated and given in completed months and weeks. For example, if the child was born at 36 weeks gestation and is examined at 8 weeks after birth, the adjusted age would be 1 month 0 weeks. In order to calculate this, you subtract the age from 40 weeks (in this case 36 weeks from 40 weeks) and get 4 weeks. You then subtract that from the child's chronologic age (the age since birth). This gives you the child's adjusted age. Move on to question 14.
13. If the child is equal to or greater than 1 year of age, indicate the child's age in completed years and months.
14. Height should be indicated in centimeters.

For percentiles of all anthropometric measurements:

Please specify EXACT PERCENTILES (no ranges, "<" or ">" signs, since such entries are not valid for Access database and statistical analysis). If child's percentile is above the highest or below the lowest percentile indicated on the chart, please specify the next whole number. For instance: if child is above 95th percentile for a certain measurement, enter "96", while if he/she is below 5th percentile enter "4". If somebody is above 90th percentile or below 10th (the highest and lowest specified percentiles on premature charts for some measurements), then enter "91" and "9", respectively.

15. Ht% required determining the child's height on a gender specific, age specific chart. For full-term children from birth to < 24 months old use **CHART 1** (Boys) or **CHART 2** (Girls). If children are ≥ 2 years old, use **CHART 3** (Boys) or **CHART 4** (Girls). Charts are provided for premature babies (**CHART 5**) and should be used if the baby is born prior to 37 weeks gestation. For babies born ≥ 37 weeks gestation, the newborn should be plotted on the birth to 36 months chart (**CHART 1 or 2**) NOT on the premature Chart. Adjusted age should be used to plot the Ht% if the child was born prior to 37 weeks gestation and was examined after the EDC. Gestational weeks of examination should be used to plot the Ht% if the child was born prior to 37 weeks gestation and was examined prior to the EDC. In that case the HT% should be calculated using the premature chart (**CHART 5**).
16. Whether the Ht is $\leq 10^{\text{th}}$ percentile should be indicated by checking "YES" or "NO".
17. Weight should be indicated in kilograms to one decimal point. For example, 3.2kg.
18. Wt% requires determining the child's weight on a gender specific, age specific chart. For full-term children from birth to < 24 months old use **CHART 1** (Boys) or **CHART 2** (Girls). If children are ≥ 2 years old, use **CHART 3** (Boys) or **CHART 4** (Girls). Charts are provided for premature babies (**CHART 6**) and should be used if the baby was born prior to 37 weeks gestation. For babies born ≥ 37 weeks gestation, the newborn should be plotted on the birth to 36 months chart (CHART 1 or 2) NOT on the Premature chart. Adjusted age should be used to plot the Wt% if the child was born prior to 37 weeks gestation and was examined after EDC. Gestational weeks at examination should be used to plot the Wt% if the child was born prior to 37 weeks

- gestation and was examined prior to the EDC. In that case the WT% should be calculated using the premature chart (**CHART 6**).
19. Whether the Wt is $\leq 10^{\text{th}}$ percentile should be indicated by checking “YES” or “NO”.
 20. Occipito-Frontal-Circumfrance (OFC) should be designated in centimeters (cm).
 21. OFC% requires determining the child’s OFC on a gender specific age specific chart. For full-term children (born ≥ 37 weeks gestation) use **CHART 7** (boys) or **CHART 8** (girls). Adjusted age should be used to plot the OFC% if the child was born prior to 37 weeks gestation and was examined after the EDC (use **CHART 7 or 8**). Gestational weeks at examination should be used to plot the OFC% if the child was born prior to 37 weeks gestation and was examined prior to the EDC. In that case the OFC% should be calculated using the premature chart (**CHART 9**).
 22. Whether the OFC is $\leq 10^{\text{th}}$ percentile should be indicated by checking “YES” or “NO”.
 23. Inner Canthal Distance (ICD) should be recorded in centimeters (cm). For example: 2.2cm.
 24. ICD% requires determining the child’s ICD on an age specific chart. One chart is provided for both sexes for infants born 27-41 weeks gestation and another one for children from birth to 14 years. The premature/newborn chart (**CHART 11**) should be used only if the baby was born prior to 37 weeks gestation. For babies born ≥ 37 weeks gestation, the child should be plotted on the Birth to 14 year chart (**CHART 10**), NOT on the chart for premature babies. Adjusted age should be used to plot the ICD% if the child was born prior to 37 weeks gestation and was examined after the EDC (**CHART 10**). Gestational weeks at examination should be used to plot the ICD% if the child was born prior to 37 weeks gestation and was examined prior to the EDC. In that case the ICD% should be calculated using the premature/newborn chart (**CHART 11**).
 25. Whether the ICD is $\leq 25^{\text{th}}$ percentile or ≤ -2 s.d. in a premature baby should be indicated by checking “YES” or “NO”.
 26. The left palpebral fissure length (PFL) should be measured and recorded in centimeters (cm). For example: 1.8cm.
 27. PFL% requires determining the child’s PFL on an age specific chart. Charts are provided for both sexes from 27 weeks gestation and from 30 weeks gestation to 14 years. The premature chart (**CHART 12**) should be used only if the baby was born prior to 37 weeks gestation. For children born ≥ 37 weeks gestation, the child should be plotted on the 30 week gestation to 14 years chart (**CHART 13**). Adjusted age should be used to plot the PFL% if the child was born prior to 37 weeks gestation and was examined after the EDC. Gestational weeks at examination should be used to plot PFL% if the child was born prior to 37 weeks gestation and was examined prior to the EDC. In that case the PFL% should be calculated using the premature chart (**CHART 12**).
 28. Whether the PFL is $\leq 10^{\text{th}}$ percentile or ≤ -2 s.d. in a premature baby should be indicated by checking “YES” or “NO”.
 29. Indicate maxillary arc in centimeters
 30. A hypoplastic midface is determined by looking at the child from the side to determine if the maxillae are underdeveloped. Indicate by checking “YES” or “NO”.

31. Indicate mandibular arc in centimeters.
32. Railroad Track configuration of the ears is depicted in the **CHART 14C**. Whether present or not must be indicated by checking “YES” or “NO”.
33. The presence of strabismus must be indicated by checking “YES” or “NO”.
34. If strabismus is present, indicated if it is unilateral or bilateral.
35. The present of ptosis must be indicated by checking “YES” or “NO” (**CHART 14A**).
36. If ptosis is present, indicate if it is unilateral or bilateral.
37. Epicanthal folds of the eyes are depicted in the **CHART 14B**. The presence of epicanthal folds must be indicated by checking “YES” or “NO”.
38. If epicanthal folds are present, indicated if they are unilateral or bilateral.
39. Anteverted nares refers to an upturned nose. One way to judge whether the nares are upturned is to determine if you can see in the baby’s nares as you are looking straight at him/her. Anteverted nares are depicted in the **CHART 15B**. Indicate whether anteverted nares are present by checking “YES” or “NO”.
40. The philtrum is measured from the base of the nasal septum to the upper margin of the vermilion border in the midline. It should be recorded in centimeters.
41. Philtrum % requires determining the child’s philtral length on the **CHART 16**. There is only one chart available for premature, full term babies, and older children. Adjusted age should be used to plot the Philtrum % if the child was born prior to 37 weeks gestation and was examined after the EDC. If the child was born prior to 37 weeks gestation and was examined prior to the EDC, the child should be plotted on the chart as Birth.
42. If the child’s philtral % is $\geq 90^{\text{th}}$ percentile, check “YES” for long philtrum. If the philtral % is $< 90^{\text{th}}$ percentile, check “NO”.
43. Indicate the smoothness of the philtrum by comparing the child’s philtrum to the 5 pictures on the Philtrum Likert Scale (**CHART 17**) and checking the number that is most similar to the child’s philtrum.
44. If the child’s philtrum is most similar to a 4 or 5, a check should be made next to the “YES”. If the philtrum is most similar to a 1, 2 or 3, a check should be made next to the “NO”.
45. Indicate the thinness and smoothness of the vermilion border by comparing the child’s Vermilion border to the 5 pictures on the Upper Lip Likert Scale (**CHART 17**), and checking the number that is most similar to the child’s Vermilion border (upper lip).
46. If the child’s Vermilion border (upper lip) is most similar to a 4 or 5, a check should be made next to the “YES”. If the Vermilion border (upper lip) is most similar to 1, 2 or 3, a check should be made next to the “NO”.
47. Clinodactyly of the 5th finger is depicted on the **CHART 15C**. If it is present a check should be made next to the “YES”. If it is not present a check should be made next to the “NO”.
48. If Clinodactyly of the 5th finger is present, indicate if it is unilateral or bilateral.

49. Camptodactyly refers to incomplete ability to completely extend the fingers. If present check "YES". If camptodactyly is not present, check "NO".
50. If camptodactyly is present indicate whether it is unilateral (on at least one finger of one hand) or bilateral (on at least one finger of both hands).
51. When the arm is flexed at 90 degrees at the elbow, is it possible to freely pronate/supinate the forearm? If there is any difficulty, check "YES", if no difficulty check NO.
52. If there are contractures at any other joints, check "YES". If there are no contractures of any other joints, check "NO".
53. Identify the particular joint which is contracted.
54. Hockey stick crease is depicted in the **CHART 14D**. If present, indicate by checking "YES". If no hockey stick crease is present, check "NO".
55. Whether unilateral or bilateral should be indicated with a check.
56. If other alterations of the palmar creases are present, it should be indicated by checking "YES". The normal crease pattern is depicted on the **CHART 15A**. If no alterations of the palmar creases are present, check "NO".
57. If other altered palmar creases are present, it should be indicated if they are unilateral or bilateral.
58. If it is a single palmar crease, a hypoplastic thenar crease or other, it should be specified. If other, the alteration should be indicated.
59. If a heart murmur is present, indicate by checking "YES". If no heart murmur is present check "NO".
60. If a heart defect has been documented by cardiac ultrasound or cardiac catheterization, it should be indicated by checking "YES". If no cardiac catheterization or cardiac ultrasound has been performed to document a cardiac defect, the answer to question 60 must be "NO".
61. If a cardiac defect has been documented by cardiac catheterization or ultrasound, specify the cardiac defect.
62. If any neurologic problem is present, check "YES". If none is present check "NO".
63. If "YES" is checked in 62, specify by checking the appropriate defect.
64. Other comments. If any other significant abnormalities are present, please indicate.
65. A check should be placed to indicate if the child is growth deficient (as specified), microcephalic (as specified), structurally abnormal (as specified) or has none of these.
66. A check should be placed to indicate "YES" if the child has FAS (as specified), "NO" if the child does not have FAS (as specified) or is deferred (as specified).
67. A check should be placed to indicate if pictures of the child have been taken.
68. Any other diagnoses should be indicated. For example, Down Syndrome or Perinatal Anoxia.