State of California - Health and Human Services Agency

Confidential Case Report of a Birth Defect

In a Fetus or Infant Less than One Year of Age

California Department of Public Health
Genetic Disease Screening Program

INSTRUCTIONS

- In accordance with State of California law (*California Code of Regulations, Title 17, Sections 6531 & 6532*), report neural tube defects (NTDs) and/or chromosomal abnormalities found in fetuses or infants less than one year of age to the California Genetic Disease Screening Program (GDSP) within 30 days of initial diagnosis.
- Reportable neural tube defects (NTDs) are outlined by ***ICD-10-CM Codes Q00.0-Q01.9, Q05.0-Q05.9, Q07.0-Q07.03***.
- Reportable chromosomal abnormalities are outlined by ***ICD-10-CM Codes Q90.0-Q99.9, excluding Q97.3, Q98.3, Q99.1, and Q99.2***.
- Submit a separate form for each individual specimen and for each fetus or infant in a multiple gestation.
- Report the simultaneous occurrence of a neural tube defect and a chromosomal abnormality for the same patient on one single form.
- Print clearly in ink or type using UPPER CASE.
- Fill bubbles completely when marking.

Genetic Disease Screening Program
850 Marina Bay Parkway
Room F-175, Mailstop 8200
Richmond, CA 94804

(510) 412-1560 (FAX)

INFORMATION ABOUT THE MOTHER

1. LAST NAME
2. FIRST NAME
3. MIDDLE INITIAL
4. MAIDEN NAME / AKA / OTHER NAMES USED FOR MOTHER
5. PRENATAL ACCESSION NUMBER *(if mother participated in the California Prenatal Screening Program)*
6. DATE OF BIRTH *(MM/DD/YYYY)*
7. APPROXIMATE AGE AT EDD *(if date of birth is unknown)*
8. SOCIAL SECURITY NUMBER
9. STREET ADDRESS *(include apartment number)*
10. CITY
11. STATE
12. ZIP CODE
13. MOTHER'S RACE / ETHNICITY *(mark all that apply)*

- O Asian Indian
- O Black
- O Cambodian
- O Chinese
- O Filipino
- O Guamanian
- O Hawaiian
- O Hispanic
- O Japanese
- O Korean
- O Laotian
- O Middle Eastern
- O Native American
- O Samoan
- O Vietnamese
- O White
- O Unknown
- O Other Southeast Asian
- O Other *(specify)*

INFORMATION ABOUT THE INFANT

14. LAST NAME
15. FIRST NAME
16. OTHER NAMES USED FOR INFANT *(including father's last name, if known)*
17. DATE OF BIRTH *(MM/DD/YYYY)*
18. GENDER
19. FETUS LETTER CODE *(A, B, C, etc.)*
20. BIRTHWEGHT OF INFANT — grams
21. IF DECEASED, DATE OF EXPIRATION *(MM/DD/YYYY)*

INFORMATION ABOUT THE REPORTING SOURCE

22. LAST NAME *(of person completing this form)*
23. FIRST NAME *(of person completing this form)*
24. DATE FORM COMPLETED *(MM/DD/YYYY)*
25. TELEPHONE NUMBER *(including extension)* — ext
26. FACILITY TYPE

- O Cytogenetic Laboratory
- O Hospital
- O MD
- O PDC *(please provide PDC code)*
- O Other *(specify)*

27. FACILITY NAME AND ADDRESS

INFORMATION ABOUT THE PREGNANCY

28. LMP / LAST MENSTRUAL PERIOD *(MM/DD/YYYY)*
29. EDD / ESTIMATED DATE OF DELIVERY *(MM/DD/YYYY)*
30. # OF FETUSES IN PREGNANCY *(including fetal demises)*
31. # OF FETUSES IN PREGNANCY WITH A BIRTH DEFECT
32. PREGNANCY STATUS

- O Continuing Pregnancy
- O Elective Termination
- O Fetal Demise / SAB / Stillbirth / Missed Abortion
- O Pregnancy Completed with a Livebirth
- O Selective Reduction
- O Unknown / Lost to Follow-Up
- O Other *(specify)*

33. DATE OF PREGNANCY STATUS *(MM/DD/YYYY)*
34. GESTATIONAL AGE AT TIME OF STATUS — weeks/days
35. METHOD USED TO DETERMINE GESTATIONAL AGE

- O LMP
- O Physical Exam
- O Ultrasound

PRIVACY STATEMENT: The Information Practices Act of 1977 (Civil Code 1798 et. seq.) requires that the following details be provided when a form is used to obtain information from individuals. The data requested in this form are required by the Genetic Disease Screening Program (GDSP) of the California Department of Public Health and are mandated by California Code of Regulations, Title 17, Section 6532. These data are used to provide information to subjects on the prevention of birth defects, to determine the prevalence of neural tube defects and chromosomal abnormalities, and to monitor trends of occurrence. These data will also be used to determine the effectiveness of the California Expanded Alpha Fetoprotein (AFP) Screening Program. It is mandatory that health professionals completing this form provide complete and accurate information. The records maintained by the GDSP are confidential, as defined in Civil Code 1798.34, and are exempt from access by any individual, except licensed medical personnel designated by the subject. The information may also be used in special studies, as defined in Health and Safety Code 100330. The furnishing of such information to the Department or its authorized representative or any other cooperating individual, agency, or organization in any such special study shall not subject any person, hospital, or other organization furnishing such information to any actions or damages.

Please list the patient's name in case of page separation:

INFORMATION ABOUT THE HOSPITAL

36. NAME OF BIRTH HOSPITAL

38. MOTHER'S MEDICAL RECORD NUMBER

37. TELEPHONE NUMBER OF BIRTH HOSPITAL

39. INFANT'S MEDICAL RECORD NUMBER

INFORMATION ABOUT THE PHYSICIAN

40. NAME AND ADDRESS OF MOTHER'S PHYSICIAN

42. NAME AND ADDRESS OF INFANT'S PHYSICIAN

41. TELEPHONE NUMBER OF MOTHER'S PHYSICIAN

43. TELEPHONE NUMBER OF INFANT'S PHYSICIAN

BIRTH DEFECT DIAGNOSIS – CHROMOSOMAL ABNORMALITIES

44. CYTOGENETIC DIAGNOSIS (ISCN Short Form) – *Copy and paste or write diagnosis clearly using UPPER CASE below. Include Human Genome Build in nomenclature when applicable.*

Do not report a) Heterochromatin Variants; b) Satellite / Stalk Variants of Chromosomes 13, 14, 15, 21, or 22; c) Inv(2)(p11;q13); d) Inv(9)(p11;q12 or q13) e) Familial Y Variants; f) Pseudomosaics; g) Known Benign Variants; or h) Regions of Homozygosity

45. CYTOGENETIC ANALYSIS TYPE
- O Karyotype
- O FISH
- O Microarray

46. SPECIMEN PREPARATION
- O Cultured
- O Direct

47. CYTOGENETIC LABORATORY SPECIMEN NUMBER

48. NAME OF CYTOGENETIC LABORATORY

49. CYTOGENETIC SPECIMEN TYPE
- O Abortus Specimen
- O Amniotic Fluid
- O Chorionic Villus (CVS)
- O Liveborn Blood
- O Liveborn Bone Marrow
- O Liveborn Cord Blood
- O Liveborn Tissue
- O Umbilical Blood (PUBS)
- O Stillborn Tissue / Blood

50. CLINICAL SIGNIFICANCE
- O Known Clinical Significance
- O Unknown Clinical Significance

51. INHERITANCE OF REARRANGEMENTS
- O De Novo
- O Paternal
- O Maternal
- O Unknown

52. REASON FOR SAMPLE
- O Confirmation of Prenatal Diagnosis
- O Congenital Anomalies
- O Dysmorphic Features
- O Maternal Age
- O Other *(specify)*

53. IS DIAGNOSIS PART OF A SYNDROME?
- O Yes *(specify)*
- O No
- O Unknown

54. SAMPLING DATE *(MM/DD/YYYY)*

55. GESTATIONAL AGE (GA) AT TIME OF SAMPLING
weeks/ days

56. METHOD USED TO DETERMINE GA
- O LMP
- O Physical Exam
- O Ultrasound

BIRTH DEFECT DIAGNOSIS – NEURAL TUBE DEFECTS

57. NEURAL TUBE DEFECT DIAGNOSIS
Spina Bifida includes Lipomeningocele, Meningocele, Meningomyelocele, and Myelomeningocele
- O Acrania
- O Anencephaly
- O Craniorachischisis
- O Encephalocele
- O Exencephaly
- O Iniencephaly
- O Meckel Gruber
- O Rachischisis
- O Spina Bifida / Myelomeningocele - Open
- O Spina Bifida / Myelomeningocele - Closed
- O Other *(specify)*

58. IS HYDROCEPHALY PRESENT?
- O Yes
- O No
- O Unknown

59. IS NTD PART OF A SYNDROME?
- O Yes *(specify)*
- O No
- O Unknown

60. ARE OTHER ABNORMALITIES PRESENT?
- O Yes *(specify)*
- O No
- O Unknown

61. ULTRASOUND INFORMATION:
NAME AND ADDRESS OF FACILITY OR PDC CODE

GESTATIONAL AGE AT TIME OF PROCEDURE
weeks/days

DATE OF PROCEDURE *(MM/DD/YYYY)*

DID ULTRASOUND PROCEDURE DETECT NTD?
- O Yes
- O No

62. AMNIOCENTESIS INFORMATION:
NAME AND ADDRESS OF FACILITY OR PDC CODE

GESTATIONAL AGE AT TIME OF PROCEDURE
weeks/days

DATE OF PROCEDURE *(MM/DD/YYYY)*

DID AMNIOCENTESIS PROCEDURE DETECT NTD?
- O Yes
- O No

63. AF-AFP LEVEL *(in M.o.M.)*

64. AF-AChE RESULT
- O Positive
- O Negative
- O Not Performed

65. IF POSTNATALLY DIAGNOSED, WHEN WAS NTD DIAGNOSED?
- O At Time of Livebirth
- O At Time of Stillbirth
- O During Physical Examination
- O Other *(specify)*

66. DATE OF POSTNATAL DIAGNOSIS *(MM/DD/YYYY)*

67. WAS THE FETAL ABNORMALITY POSTNATALLY CONFIRMED?
- O Yes
- O No
- O Pending

68. SOURCE(S) OF CONFIRMATION *(mark all that apply)*
- O Autopsy / Pathology Report
- O Clinician Notes
- O Delivery Room Report
- O Outcome of Pregnancy
- O Ultrasound Report
- O Other *(specify)*