

Congenital Adrenal Hyperplasia (CAH) Data Elements - Enrollment

| 1 | Measure | Category | Answer Type | Measure Answer | Units (for value box) |
|---|---|--------------|-------------|---|---|
| 2 | Unique registry ID | Demographics | Free Text | | (2digFIPS/2digBirthYr/ 1digCenter/3digAscession) |
| 3 | Was enrollment in registry refused? | Demographics | Yes No | | |
| 4 | Who consented enrollment in registry? | Demographics | Pick List | Both parents, mother only, father only, both guardians, female guardian, male guardian | |
| | Was assent obtained? | Demographics | Pick List | Missing/unknown data, no, yes | |
| 5 | Permission to contact about research | Demographics | Yes No | Permission to contact: I agree to be contacted with information on potential future research applicable to me and/or child's condition that becomes available. | |
| 6 | Permission for compensation | Demographics | Yes No | Permission for compensation: I agree that identifying information about me/my child may be used or disclosed as necessary to provide compensation if I and/or my child are eligible for compensation. | |
| 7 | Patient is enrolled in research studies | Demographics | Check boxes | Missing/unknown data, no, yes (if yes, please specify in next question) | |
| | Research studies patient is enrolled in | Demographics | Free Text | | |

Congenital Adrenal Hyperplasia (CAH) Data Elements - Enrollment

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| 8 | Follow up status | Demographics | Pick List | Active, Inactive - Deceased, Inactive - Lost to follow up, Inactive - Moved to another State participating in registry, Inactive - Moved to another State not participating in registry, Inactive -Refused follow up, Inactive - Treatment deemed not necessary; | |
| 9 | Has patient been followed by >1 center for this condition? | Demographics | Pick List | Missing/unknown data, no, yes - select state from pull down list, yes - select state from pull down list, yes - select state from pull down list | |
| 11 | If patient has been followed by >1 center, did patient grant permission to share data via registry between treating center? | Demographics | Pick List | Missing/unknown data, N/A, Yes, No | |
| 12 | If patient has been followed by >1 center for this condition, what type of provider was treating the patient? | Demographics | Check boxes | Missing/unknown data, adult endocrinologist, geneticist, family practice physician, gynecologist, internist, nurse practitioner, pediatric endocrinologist, pediatrician, reproductive endocrinologist, psychologist, psychiatrist, urologist | |
| 13 | Other health services received by patient | Demographics | Check boxes | Missing/unknown data, none, audiology, behavioral/developmental pediatrics, cardiology, dermatology, genetics, genetic counseling, hematology/oncology, home health care, nephrology, neurology, neuropsychology, OB-GYN, occupational therapy, ophthalmology, orthopedics, otolaryngology, pulmonology, physical therapy, psychiatry, | |
| 14 | Community resources received by patient | Demographics | Check boxes | Missing/unknown data, none, daycare, family support group related to this condition, family support - other, Medical Home, nutritional services (WIC/MAC), personal care attendant (PCA), preschool, Head Start, respite care, social services - county, social services - medical, social services - developmental disability, waived services (CAC/CADI waiver, other waivers) | |
| 15 | If deceased, date of death (if N/A enter 01/01/1901) | Demographics | Date Field | | |
| 16 | Ethnic category | Demographics | Pick List | Missing/unknown data, Hispanic or Latino, not Hispanic or Latino | |

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| 17 | Racial category | Demographics | Pick List | Missing/unknown data, American Indian/Alaska Native, Asian, Native Hawaiian or other Pacific Islander, Black or African American, Multi-racial, White | |
| 18 | Mother's maiden name at birth | Demographics | Free Text | | |
| 19 | Maternal education: highest level of education | Socioeconomic Status | Pick List | Missing/unknown data, 1-8 years, 9-12 years (no diploma), completed high school, training after high school, some college, college graduate, post-graduate | |
| 20 | Paternal education: highest level of education | Socioeconomic Status | Pick List | Missing/unknown data, 1-8 years, 9-12 years (no diploma), completed high school, training after high school, some college, college graduate, post-graduate | |
| 21 | Is primary caregiver proficient in written English? | Socioeconomic Status | Pick List | Missing/unknown data, yes, no | |
| 22 | Is primary caregiver proficient in spoken English? | Socioeconomic Status | Pick List | Missing/unknown data, yes, no | |
| 23 | Was written/web-based information on this condition provided in primary caregiver's primary language? | Socioeconomic Status | Pick List | Missing/unknown data, yes, no | |
| 24 | Primary insurance at time of enrollment | Socioeconomic Status | Pick List | Missing/unknown data, Private, Public, None, Self Pay | |
| 25 | Marital status of primary caregiver at time of enrollment | Socioeconomic Status | Pick List | Missing/unknown data, married, not married | |

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|----|--|----------------------|-------------|--|----------|
| 26 | Family members at time of enrollment (total including number of siblings, half-siblings, adopted, etc) | Socioeconomic Status | Check boxes | Missing/unknown data, # of siblings, # of half siblings, # of parents, # of grandparents, # of step siblings, # of adopted siblings | |
| | Family members currently living in the household at time of enrollment | Socioeconomic Status | Pick List | Missing/unknown data, # of siblings under 18, # of half siblings under 18, # of step siblings under 18, # of adopted siblings under 18, # of parents, # of grandparents, # of adults | |
| 27 | Mother's height self-report at time of patient enrollment in registry (or 88888 if unknown) | Family History | Decimal | NOTE: auto convert to cm. | (inches) |
| 28 | Father's height self-report at time of patient enrollment in registry (or 88888 if unknown) | Family History | Decimal | NOTE: auto convert to cm. | (inches) |
| 29 | Are parents affected with CAH? | Family History | Check boxes | Missing/unknown data, mother affected, father affected, mother not affected, father not affected | |
| 30 | Select family history | Family History | Check boxes | Missing/unknown data, acne, adrenalectomy, amenorrhea - primary, amenorrhea - secondary, azoospermia, cerebrovascular death, consanguinity - yes, consanguinity -no, diabetes - type 1, diabetes - type 2, endometriosis, hirsutism, hypertension, hypotension, hypoglycemia, hypothyroidism, hyperthyroidism, infant death - female, infant death - male, infertility - females, infertility - males, irregular menses, male pattern hair loss, miscarriages, obesity, oligospermia, osteoporosis, ovarian adrenal rests, ovarian tumors, pituitary | |
| 31 | Any infant/sibling deaths? | Family History | Pick list | Missing/unknown data, no, yes -1 death, yes - 2 deaths, yes - 3 deaths | |
| | Date and age of Infant/sibling death #1 (enter 88888 if unknown) | Family History | Free Text | _____ Month/Day/Year _____ Age | |

Congenital Adrenal Hyperplasia (CAH) Data Elements - Enrollment

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|----|---|----------------|-----------|--|--|
| | Cause(s) of Infant/sibling death #1 (enter 88888 if unknown) | Family History | Free Text | | |
| | Sex of infant/sibling #1 who died | Family History | Pick List | Missing/unknown, male, female | |
| | Date and age of Infant/sibling death #1 (enter 88888 if unknown) | Family History | Free Text | _____ Month/Day/Year _____ Age | |
| | Cause(s) of Infant/sibling death #1 (enter 88888 if unknown) | Family History | Free Text | | |
| | Sex of infant/sibling #1 who died | Family History | Pick List | Missing/unknown, male, female | |
| | Date and age of Infant/sibling death #1 (enter 88888 if unknown) | Family History | Free Text | _____ Month/Day/Year _____ Age | |
| | Cause(s) of Infant/sibling death #1 (enter 88888 if unknown) | Family History | Free Text | | |
| | Sex of infant/sibling #1 who died | Family History | Pick List | Missing/unknown, male, female | |
| 32 | Sibling #1 (oldest sibling) evaluated for this condition? | Family History | Pick List | Missing/unknown data, not tested, tested - affected (if affected and consented to registry participation, create new registry case and free text registry unique ID below), tested - unaffected, N/A | |

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|----|---|------------------|-------------|--|--|
| 33 | Sibling #2 (second oldest sibling) evaluated for this condition? | Family History | Pick List | Missing/unknown data, not tested, tested - affected (if affected and consented to registry participation, create new registry case and free text registry unique ID below), tested - unaffected, N/A | |
| 34 | Sibling #3 (third oldest sibling) evaluated for this condition? | Family History | Pick List | Missing/unknown data, not tested, tested - affected (if affected and consented to registry participation, create new registry case and free text registry unique ID below), tested - unaffected, N/A | |
| 35 | Sibling #4 (fourth oldest sibling) evaluated for this condition? | Family History | Pick List | Missing/unknown data, not tested, tested - affected (if affected and consented to registry participation, create new registry case and free text registry unique ID below), tested - unaffected, N/A | |
| 36 | Sibling #5 (fifth oldest sibling) evaluated for this condition? | Family History | Pick List | Missing/unknown data, not tested, tested - affected (if affected and consented to registry participation, create new registry case and free text registry unique ID below), tested - unaffected, N/A | |
| 37 | Sibling #6 (sixth oldest sibling) evaluated for this condition? | Family History | Pick List | Missing/unknown data, not tested, tested - affected (if affected and consented to registry participation, create new registry case and free text registry unique ID below), tested - unaffected, N/A | |
| 38 | If sibling(s) affected with this condition enter sibling # and sibling unique registry ID number(s) here (enter 99999 if N/A) | Family History | Pick List | Use numbers 1-9 with the option of "Other." | |
| 39 | Was prenatal testing for this condition done during this pregnancy? | Prenatal History | Pick List | Missing/unknown data, yes, no, offered but declined by parent | |
| 40 | Method(s) if prenatal testing for this condition done | Prenatal History | Check boxes | Missing/unknown data, N/A, amniocentesis (biochemical/enzyme), amniocentesis (DNA), chorionic villus (biochemical/enzyme), chorionic villus (DNA), fetal skin biopsy, periumbilical blood | |
| 41 | Was maternal steroid treatment received during this pregnancy? | Prenatal History | Check boxes | Missing/unknown data, no, yes - dexamethasone, yes - betamethasone, yes - prednisone, yes - other steroid | |

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|----|--|------------------|-------------|---|--|
| 42 | If maternal steroid treatment received during this pregnancy, reason for treatment? | Prenatal History | Check boxes | Missing/unknown data, N/A, offered but refused, premature labor, mother affected with CAH, prenatal treatment for suspected or known fetal CAH, other _____(free text) | |
| 43 | If maternal steroid treatment received during this pregnancy, what was the daily dose of steroid (enter 99999 if N/A or 88888 if unknown)? | Prenatal History | Decimal | (put in decimal point to minimize invalid entries) | |
| 44 | If prenatal steroid treatment received during this pregnancy, week of gestation at time of initiation of treatment (enter 99999 if N/A or 88888 if unknown)? | Prenatal History | Pick List | 4,5,6,7,8,9,10,11,12,13,14,15,16,17,18,19,20,21,22,23,24,25,26,27,28,29,30,31,32,33,34,35,36,37,38,39,40 | (weeks gestation) |
| 45 | If maternal steroid treatment received during this pregnancy, week of gestation at end of treatment (enter 99999 if N/A or 88888 if unknown)? | Prenatal History | Pick List | 4,5,6,7,8,9,10,11,12,13,14,15,16,17,18,19,20,21,22,23,24,25,26,27,28,29,30,31,32,33,34,35,36,37,38,39,40 | (weeks gestation) |
| 46 | If maternal steroid treatment received during this pregnancy, maternal steroid side effects | Prenatal History | Check boxes | Missing/unknown data, N/A - not prescribed, none, gestational diabetes, hyperglycemia, hypertension, mood swings, moon facies, striae, weight gain, other | |
| 47 | Enter any medications mother received during pregnancy | Prenatal History | Check boxes | Missing/unknown, none, anticonvulsants, antidepressants, antidiabetic medication, antihypertensives, antithyroid medication, anxiolytics, hormonal medications, narcotics, thyroid hormone, other Do we cover recreational drugs? For certain | |
| 48 | Pregnancy risk factors and/or complications | Prenatal History | Check boxes | Missing/unknown data, none, amniocentesis, chorionic villus sampling, premature labor, preeclampsia, bleeding, placenta previa, placental abruption, amniotic fluid infection, depression, diabetes, gestational diabetes, hypertension, hypothyroidism, hyperthyroidism, virilization. | Purpose is to cover general and endocrine issues that can influence outcomes |
| 49 | Did this pregnancy result from use of assisted reproductive technology? | Prenatal History | Check boxes | Missing/unknown data, no, ovarian hyperstimulation (e.g. clomiphene), cryopreservation of ovarian or testicular tissue, sperm donation, oocyte donation, embryo donation, in vitro fertilization, gamete intra-fallopian transfer (GIFT), zygote intra-fallopian transfer (ZIFT), intra-cytoplasmic sperm injection, pre-implantation genetic diagnosis, traditional surrogacy, gestational | |

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|----|--|------------------|-------------|---|---------------------------------|
| 50 | Weeks of gestation at birth (enter 88888 if unknown) | Neonatal History | Decimal | Unit for value box = weeks [set criteria for decimals e.g. 38.0] | weeks |
| 51 | Additional information about newborn period | Neonatal History | Check boxes | Missing/unknown data, none, antibiotics, breastfed, CNS bleeding, CNS infection, CNS injury, distress, hypoglycemia, hypotension, hypertension, hypoxia, jaundice, meconium stain amniotic fluid, nuchal cord, sepsis, TPN, transfused, ventilator | From national NBS data elements |
| 52 | Birth weight (enter 88888 if unknown) | Measurements | Decimal | set criteria for decimals QUESTION: Can we enter in lb oz and have automatic conversion to metric? Most parents will not report metric. | (kg) |
| 53 | Birth length (enter 88888 if unknown) | Measurements | Decimal | set criteria for decimals | (cm) |
| 54 | Birth head circumference (a.k.a occipitofrontal circumference-OFC) (enter 88888 if unknown) | Measurements | Decimal | set criteria for decimals | (cm) |
| | Additional information about the perinatal period. Please check any additional events that occurred or medications received in the prenatal period | Neonatal History | Check boxes | None, missing/unknown, fetal distress, intrauterine growth retardation, antibiotics, corticosteroids, other [specify] | |
| | Additional information about the delivery period. | Neonatal History | Check boxes | None, missing/unknown, fetal distress, hypoxia, meconium staining of amniotic fluid, nuchal cord | |
| | APGAR score | Neonatal History | Pick List | <3, 4,5,6,7,8,9,10 | |
| | Additional information about the postnatal period. | Neonatal History | Check boxes | Antibiotics, blood transfusion, central nervous system bleeding, central nervous system infection, central nervous system injury, hypertension, hypoglycemia, hypotension, hypoxia, jaundice requiring intervention, parenteral nutrition, seizure, sepsis, surgery | |

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|----|--|-------------------|-----------|---|---------|
| 55 | State newborn screen serial number (enter 88888 if unknown) | Newborn Screening | Integer | | |
| 56 | Age (days) at time primary care provider or endocrine provider was notified of FIRST abnormal newborn screen for this condition (enter 88888 if unknown) | Newborn Screening | Integer | This question is for State NBS programs to gauge how quickly post positive NBS are patients receiving care. The problem with having both primary and endo is that we don't which one provided care solely based on days. Also it can't be only days as SV and NC can come months, years later. Should we have separate questions? Another thing to consider is that we will not have the exact time that NBS notified the primary. We can guess | (days) |
| 57 | 17-hydroxyprogesterone (17-OHP) on FIRST newborn screen (enter 88888 if unknown) | Newborn Screening | Decimal | | (ng/mL) |
| 58 | 2nd tier 17-hydroxyprogesterone (17-OHP) results from FIRST newborn screen (enter 99999 if N/A or 88888 if unknown) | Newborn Screening | Decimal | | (ng/mL) |
| 59 | 2nd tier 17-hydroxyprogesterone (17-OHP) + androstenedione/cortisol ratio from FIRST newborn screen (enter 99999 if N/A or 88888 if unknown) | Newborn Screening | Decimal | | |
| 60 | 2nd tier results cortisol detected from FIRST newborn screen | Newborn Screening | Pick List | Missing/unknown data, N/A - 2nd tier screen not done, yes, no | |
| 61 | 2nd tier results 21-deoxycortisol from FIRST newborn screen (enter 99999 if N/A or 88888 if unknown) | Newborn Screening | Decimal | | (ng/mL) |
| 62 | 2nd tier results 11-deoxycortisol from FIRST newborn screen (enter 99999 if N/A or 88888 if unknown) | Newborn Screening | Decimal | | (ng/mL) |

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|----|---|-------------------|-----------|---|---------|
| 63 | 17-hydroxyprogesterone (17-OHP) on SECOND newborn screen (enter 99999 if N/A or 88888 if unknown) | Newborn Screening | Decimal | | (ng/mL) |
| 64 | 2nd tier 17-hydroxyprogesterone (17-OHP) results from SECOND newborn screen (enter 99999 if N/A or 88888 if unknown) | Newborn Screening | Decimal | | (ng/mL) |
| 65 | 2nd tier 17-hydroxyprogesterone (17-OHP) + androstenedione/cortisol ratio from SECOND newborn screen (enter 99999 if N/A or 88888 if unknown) | Newborn Screening | Decimal | | |
| 66 | 2nd tier cortisol results from SECOND newborn screen | Newborn Screening | Pick List | Missing/unknown data, N/A - 2nd tier screen not done, yes, no | |
| 67 | 2nd tier results 21-deoxycortisol from SECOND newborn screen (enter 99999 if N/A or 88888 if unknown) | Newborn Screening | Decimal | | (ng/mL) |
| 68 | 2nd tier results 11-deoxycortisol from SECOND newborn screen (enter 99999 if N/A or 88888 if unknown) | Newborn Screening | Decimal | | (ng/mL) |
| 69 | 17-hydroxyprogesterone (17-OHP) on THIRD newborn screen (enter 99999 if N/A or 88888 if unknown) | Newborn Screening | Decimal | | (ng/mL) |
| 70 | 2nd tier 17-hydroxyprogesterone (17-OHP) results from THIRD newborn screen (enter 99999 if N/A or 88888 if unknown) | Newborn Screening | Decimal | | (ng/mL) |
| 71 | 2nd tier 17-hydroxyprogesterone (17-OHP) + androstenedione/cortisol ratio from THIRD newborn screen (enter 99999 if N/A or 88888 if unknown) | Newborn Screening | Decimal | | |

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|----|--|--------------------|-----------|---|---------|
| 72 | 2nd tier cortisol results from THIRD newborn screen | Newborn Screening | Pick List | Missing/unknown data, N/A - 2nd tier screen not done, yes, no | |
| 73 | 2nd tier results 21-deoxycortisol from THIRD newborn screen (enter 99999 if N/A or 88888 if unknown) | Newborn Screening | Decimal | | (ng/mL) |
| 74 | 2nd tier results 11-deoxycortisol from THIRD newborn screen (enter 99999 if N/A or 88888 if unknown) | Newborn Screening | Decimal | | (ng/mL) |
| 75 | Baseline serum 17-hydroxyprogesterone (17-OHP) level (enter 99999 if NA or 88888 if unknown) | Diagnostic Testing | Decimal | | (ng/dL) |
| 76 | Baseline serum deoxycorticosterone (DOC) level (enter 99999 if N/A or 88888 if unknown) | Diagnostic Testing | Decimal | | (ng/dL) |
| 77 | Baseline serum 17-hydroxypregnenolone (17-OHPreg) level (enter 99999 if N/A or 88888 if unknown) | Diagnostic Testing | Decimal | | (ng/dL) |
| 78 | Baseline serum corticosterone (B) level (enter 99999 if N/A or 88888 if unknown) | Diagnostic Testing | Decimal | | (ng/dL) |
| 79 | Baseline serum cortisol (F) level (enter 99999 if N/A or 88888 if unknown) | Diagnostic Testing | Decimal | | (ug/dL) |
| 80 | Baseline serum testosterone (T) level (enter 99999 if N/A or 88888 if unknown) | Diagnostic Testing | Decimal | | ng/dl |

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|----|---|--------------------|-----------|---|------------|
| 81 | Baseline serum androstenedione (D4A) level (enter 99999 if N/A or 88888 if unknown) | Diagnostic Testing | Decimal | | ng/dl |
| 82 | Baseline serum dehydroxyepiandrosterone (DHEA) level (enter 99999 if N/A or 88888 if unknown) | Diagnostic Testing | Decimal | | ng/dl |
| 83 | Baseline serum dihydrotestosterone (DHT) level (enter 99999 if N/A or 88888 if unknown) | Diagnostic Testing | Decimal | | pg/ml |
| 84 | Baseline renin level (enter 99999 if N/A or 88888 if unknown) | Diagnostic Testing | Decimal | | ng/ml/hr |
| 85 | Was a cosyntropin (ACTH) stimulation test performed? | Diagnostic Testing | Pick List | Missing/unknown information, yes, no | |
| 86 | Dose of cosyntropin for high dose ACTH stimulation test | Diagnostic Testing | Pick List | Missing/unknown data, N/A - cosyntropin stimulation test not done, 125 mcg, 250 mcg, other dose | (mcg/dose) |
| 87 | ACTH stimulated serum 17-hydroxyprogesterone (17-OHP) level (enter 99999 if N/A or 88888 if unknown) | Diagnostic Testing | Decimal | | (ng/dL) |
| 88 | ACTH stimulated serum deoxycorticosterone (DOC) level (enter 99999 if N/A or 88888 if unknown) | Diagnostic Testing | Decimal | | (ng/dL) |
| 89 | ACTH stimulated serum 17-hydroxypregnenolone (17-OHPreg) level (enter 99999 if N/A or 88888 if unknown) | Diagnostic Testing | Decimal | | (ng/dL) |

Congenital Adrenal Hyperplasia (CAH) Data Elements - Enrollment

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|----|--|--------------------|-----------|--------------------------------------|---------|
| 90 | ACTH stimulated serum corticosterone (B) level (enter 99999 if N/A or 88888 if unknown) | Diagnostic Testing | Decimal | | (ng/dL) |
| 91 | ACTH stimulated serum cortisol (F) level (enter 99999 if N/A or 88888 if unknown) | Diagnostic Testing | Decimal | | (ug/dL) |
| 92 | ACTH stimulated serum testosterone (T) level (enter 99999 if N/A or 88888 if unknown) | Diagnostic Testing | Decimal | | (ng/dL) |
| 93 | ACTH stimulated serum androstenedione (D4A) level (enter 99999 if N/A or 88888 if unknown) | Diagnostic Testing | Decimal | | (ng/dL) |
| 94 | ACTH stimulated serum dehydroxyepiandrosterone (DHEA) level (enter 99999 if N/A or 88888 if unknown) | Diagnostic Testing | Decimal | | (ng/dL) |
| 95 | ACTH stimulated serum dehydroxyepiandrosterone sulfate (DHEA-S) level (enter 99999 if N/A or 88888 if unknown) | Diagnostic Testing | Decimal | | (ug/dL) |
| 96 | Was a β -hCG stimulation test performed? | Diagnostic Testing | Pick List | Missing/unknown information, yes, no | |
| 97 | Dose and frequency of β -hCG stimulation test? (enter 99999 if N/A or 88888 if unknown) | Diagnostic Testing | Free Text | | |
| 98 | If β -hCG stimulation test performed, at what age? (enter 99999 if N/A, or 88888 if unknown). | Diagnostic Testing | Integer | | years |

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| 99 | β-hCG stimulated serum testosterone (T) level (enter 99999 if N/A or 88888 if unknown) | Diagnostic Testing | Decimal | | (ng/dL) |
| 100 | β-hCG stimulated serum androstenedione (D4A) level (enter 99999 if N/A or 88888 if unknown) | Diagnostic Testing | Decimal | | (ng/dL) |
| 101 | β-hCG stimulated serum dihydrotestosterone (DHT) level (enter 99999 if N/A or 88888 if unknown) | Diagnostic Testing | Decimal | | (pg/mL) |
| 102 | Molecular testing: Common or targeted mutation panel | Diagnostic Testing | Pick List | Missing/unknown data, no, yes - abnormal: compound heterozygote, yes - abnormal: homozygote, yes - abnormal: simple heterozygote, yes - alteration(s) of unknown significance detected, yes but no mutations were detected | |
| 103 | If common or targeted mutation panel performed, please list laboratory | Diagnostic Testing | Free Text | | |
| 104 | Molecular testing: Full sequencing | Diagnostic Testing | Pick List | Missing/unknown data, no, yes - abnormal: compound heterozygote, yes - abnormal: presumed compound heterozygote 2nd mutation not identified, yes - abnormal: homozygote, yes - abnormal: simple heterozygote, yes - alteration(s) of unknown significance detected, yes but no | |
| 105 | Mutation description on patient: Allele 1 (format example 985A>G) (enter 99999 if N/A or 88888 if unknown) | Diagnostic Testing | Free Text | | |
| 106 | Mutation description on patient: Allele 2 (format example 985A>G) (enter 99999 if N/A or 88888 if unknown) | Diagnostic Testing | Free Text | | |
| 107 | Mutation description on biological mother: Allele 1 (format example 985A>G) (enter 99999 if N/A or 88888 if unknown) | Diagnostic Testing | Free Text | | |

Congenital Adrenal Hyperplasia (CAH) Data Elements - Enrollment

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| 108 | Mutation description on biological mother: Allele 2 (format example 985A>G) (enter 99999 if N/A or 88888 if unknown) | Diagnostic Testing | Free Text | | |
| 109 | Mutation description on biological father: Allele 1 (format example 985A>G) (enter 99999 if N/A or 88888 if unknown) | Diagnostic Testing | Free Text | | |
| 110 | Mutation description on biological father: Allele 2 (format example 985A>G) (enter 99999 if N/A or 88888 if unknown) | Diagnostic Testing | Free Text | | |
| 111 | Karyotype results | Diagnostic Testing | Pick List | Missing/unknown data, karyotype was not done, 46XX, 46XY, mosaicism or other (go to next question and explain) | |
| 112 | If karyotype is a mosaicism or other, please describe (enter 99999 if N/A or 88888 if unknown) | Diagnostic Testing | Free Text | | |
| 113 | Was genetic counseling for this condition provided? | Past Health History | Pick List | Missing/unknown data, yes, no | |
| 114 | If genetic counseling was provided, who provided genetic counseling? | Past Health History | Pick List | Missing/unknown data, endocrinologist - adult, endocrinologist - pediatric, geneticist, genetic counselor, family practice physician, internist, nurse practitioner, pediatrician | |
| 115 | Initial diagnosis of CAH found by: | Past Health History | Check boxes | Missing/unknown data, abnormal newborn screen, prenatal testing, post-mortem testing, by provider, by another provider – adult endocrinologist, by another provider – family practice physician, by another provider – geneticist, by another provider – internist, by another provider – nurse practitioner, by another provider – pediatric endocrinologist, by another provider – | |

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| 116 | Type of CAH | Past Health History | Pick List | 21-OHD – salt-wasting, 21-OHD – simple-virilizing, 21-OHD - non-classical, 3b-HSD-II – classical, 3b-HSD-II – non-classical, 11-OHD –classical, 11-OHD – non-classical, 17a-OHase/17,20-lyase combined complete, 17a-OHase/17,20-lyase combined partial, lipoid (StAR) – classical, lipoid (StAR) – non-classical, P450 oxidoreductase (POR) not yet determined | |
| 117 | Number of CAH related hospitalizations prior to enrollment in registry | Past Health History | Pick List | Missing/unknown data, 0,1,2,3,4,5,6,7,8,9,10, >10 | |
| 118 | Age at time of initial face to face intervention for this condition (enter 88888 if unknown) | Past Health History | Free Text | | (days, months, years) |
| 119 | Symptom(s) - manifestations at time of initial intervention for this condition | Past Health History | Check boxes | Missing/unknown data, none, acanthosis nigricans, acne, anovulation, dehydration, developmental delay, dysmorphic features, growth acceleration, gynecomastia, hirsutism, hypertonia, hypotonia, hypoglycemia, hyperpigmentation, hypertensive nephrosclerosis, infertility, insulin resistance, irregular menses, irritability, left ventricular hypertrophy, lethargy, male pattern hair loss, metabolic acidosis, osteopenia, osteoporosis, polycystic ovarian syndrome, poor feeding, precocious puberty, pubic hair development, retinopathy, seizures, skeletal malformations, teething, vaginal bleeding | |
| 120 | Other symptom(s) - manifestations at time of initial intervention for this condition | Past Health History | Free Text | | |
| 121 | Weight at time of initial intervention for this condition (enter 88888 if unknown or 77777 if not done) | Past Health History | Decimal | | (kg) |
| 122 | Height at time of initial intervention for this condition (enter 88888 if unknown or 77777 if not done) | Past Health History | Decimal | | (cm) |

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| 123 | Head circumference (OFC) at time of initial intervention for this condition (enter 88888 if unknown or 77777 if not done) | Past Health History | Decimal | | (cm) |
| 124 | Blood pressure at time of initial intervention for this condition | Past Health History | Pick List | Missing/unknown data, blood pressure not measured, hypertension, hypotension, normal blood pressure | |
| 125 | If patient has abnormal blood pressure, at what age was abnormal blood pressure originally observed (enter 99999 if N/A or 88888 if unknown) | Past Health History | Decimal | | (years, months) |
| 126 | Tanner Stage of pubic hair development at time of initial intervention for this condition | Past Health History | Pick List | Missing/unknown data, not done, I, II, III, IV, V | |
| 127 | Tanner Stage of breast development at time of initial intervention for this condition | Past Health History | Pick List | Missing/unknown data, N/A, not done, I, II, III, IV, V | |
| 128 | Menstrual history at time of initial intervention for this condition | Past Health History | Pick List | Missing/unknown data, N/A - patient is male, N/A - prepubertal female, primary amenorrhea, secondary amenorrhea (no menses for >6 months) oligomenorrhea, regular menses | |
| 129 | Lab tests collected at initial intervention for this condition | Past Health History | Check boxes | Missing/unknown data, none, ABG-WNL, ABG-high ABG-low, ALT-WNL, ALT-Abn, AST-WNL, AST-Abn, AlkPhos-WNL, AlkPhos-Abn, Anion Gap-WNL, Anion Gap-Abn, BUN-WNL, BUN-Abn, Ca+++WNL, Ca+++high, Ca+++low, Cr-WNL, Cr-Abn, Cl-WNL, Cl-high, Cl-low, CO2-WNL, CO2-high, CO2-low, FSH-WNL, FSH-high, FSH-low, GGT-WNL, GGT-Abn, Gluc-WNL, Gluc-low, Gluc-high, glucose (fasting) - WNL, glucose (fasting) - low, glucose (fasting) - high, insulin (fasting) - WNL, insulin (fasting) - Abn, K+-WNL, K+-high, K+-low, LH-WNL, LH-high, LH-low, Na+-WNL, Na+-high, Na+-low, Phosphorus - WNL, Phosphorus - high, Phosphorus - low, Total Cholesterol (fasting) - WNL, Total Cholesterol (fasting) - Abn, Total Cholesterol (random) - WNL, Total Cholesterol (random) - Abn, Urine (24 hours) for 17-ketosteroids - WNL, Urine (24 hours) for | |

Congenital Adrenal Hyperplasia (CAH) Data Elements - Enrollment

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| 130 | Lab tests other general (indicate type and if WNL or Abn) collected at initial intervention for this condition (enter 99999 if N/A or 88888 if unknown) | Past Health History | Free Text | | |
| 131 | If patient ever had an elevated fasting glucose level, at what age was the patient? (enter 99999 if N/A or 88888 if unknown) | Past Health History | Decimal | | (years, months) |
| 132 | If patient ever had an elevated fasting insulin level, at what age was the patient? (enter 99999 if N/A or 88888 if unknown) | Past Health History | Decimal | | (years, months) |
| 133 | Patient's age at menarche, if female (enter 99999 if N/A or 88888 if unknown) | Past Health History | Decimal | | (years, months) |
| 134 | Patient's age at puberty, if male (enter 99999 if N/A or 88888 if unknown) | Past Health History | Decimal | | (years, months) |
| 135 | If patient had atypical genitalia at time of initial intervention for this condition, what type of atypical finding was noted? (complete Urogenital Survey now) | Past Health History - Genital | Check boxes | Missing/unknown data, N/A - genitalia were normal, abdominal testes, abnormal anogenital distance, anorchia, bifid scrotum, chordee, clitoromegaly, complete labioscrotal fusion, cryptorchidism, glandular hypospadias, hyperpigmentation, inguinal testes, labial testes, proximal penile hypospadias, distal penile hypospadias, penoscrotal hypospadias, micropenis, penile enlargement, partial labioscrotal fusion, rugated labioscrotal folds, single perineal opening, vaginal pouch | |
| 136 | If patient had micropenis or penile enlargement at time of initial intervention for this condition, size of penis: enter as length, width in cm (enter 99999 if N/A or 88888 if unknown) | Past Health History - Genital | Decimal | | (cm) |
| 137 | If patient had clitoromegaly at time of initial intervention for this condition, size of clitoris: enter as length,width in cm (enter 99999 if N/A or 88888 if unknown) | Past Health History - Genital | Decimal | | (cm) |

Congenital Adrenal Hyperplasia (CAH) Data Elements - Enrollment

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| 138 | Was genital surgery performed before enrollment? | Past Health History - Surgery | Pick List | Missing/unknown data, N/A - no genital abnormalities indicating need for surgery, yes (complete Urogenital Survey now), no | |
| 139 | Testicular volume (enter volume as left testicle, right testicle) at time of initial intervention for this condition (enter 99999 if N/A or 88888 if unknown or | Past Health History - Genital | Decimal | | (mL) |
| 140 | At time of enrollment, did patient present with atypical genitalia? | Past Health History - Genital | Check boxes | Missing/unknown data, N/A - enrollment is initial intervention, N/A - genitalia were normal, abdominal testes, abnormal anogenital distance, anorchia, bifid scrotum, chordee, clitoromegaly, complete labioscrotal fusion, cryptorchidism, glandular hypospadias, hyperpigmentation, inguinal testes, labial testes, proximal penile hypospadias, distal penile hypospadias, penoscrotal hypospadias, micropenis, penile enlargement, partial labioscrotal fusion, rugated labioscrotal folds, single | |
| 141 | Pelvic/testicular/genitogram ultrasound results at initial intervention for this condition? | Past Health History - Imaging | Pick List | Missing/unknown data, not done, normal, abnormal pelvic ultrasound, abnormal testicular ultrasound, abnormal - gonads not visualized, non-diagnostic | |
| 142 | If abnormal pelvic ultrasound results, explain finding (enter 99999 if N/A or 88888 if unknown) | Past Health History - Imaging | Free Text | | |
| 143 | If abnormal testicular ultrasound results, explain finding (enter 99999 if N/A or 88888 if unknown) | Past Health History - Imaging | Free Text | | |
| 144 | If patient had a urogenital sinus, what was the confluence of the urogenital sinus with the vagina? | Past Health History - Imaging | Pick List | Missing/unknown data, N/A, low, high | |
| 145 | If patient had a urogenital sinus, enter size (enter 99999 if N/A or 88888 if unknown) | Past Health History - Imaging | Decimal | | (cm) |

Congenital Adrenal Hyperplasia (CAH) Data Elements - Enrollment

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| 146 | Results of bone age at initial intervention for this condition (enter 99999 if N/A or 88888 if unknown) | Past Health History - Imaging | Free Text | | (years, months) |
| 147 | Date of bone age at initial intervention for this condition (enter 99999 if N/A or 88888 if unknown) | Past Health History - Imaging | Date Field | | |
| 148 | Method of reading bone age | Past Health History - Imaging | Pick List | Missing/unknown data, N/A, bone age not done, Greulich Pyle, Tanner Whitehouse, other | |
| 149 | Has a dexa scan been performed prior to enrollment? | Past Health History - Imaging | Check boxes | Missing/unknown data, N/A, hip, heel, pelvis, spine, total body, wrist, other | |
| 150 | If a dexa scan was found to be abnormal, what was the patient's age at first abnormal dexa scan? | Past Health History - Imaging | Decimal | | (years of age) |
| 151 | If abnormal dexa scan (z-score \leq - 3 to - 2), specify site | Past Health History - Imaging | Check boxes | Missing/unknown, N/A, hip, heel, pelvis, spine, total body, wrist, other | |
| 152 | If abnormal dexa scan (z-score \leq - 4 to - 3), specify site | Past Health History - Imaging | Check boxes | Missing/unknown, N/A, hip, heel, pelvis, spine, total body, wrist, other | |
| 153 | If abnormal dexa scan (z-score \leq - 4), specify site | Past Health History - Imaging | Check boxes | Missing/unknown, N/A, hip, heel, pelvis, spine, total body, wrist, other | |
| 154 | Are oral steroids prescribed? | Past Health History - Pharmacotherapy | Pick list | Missing/unknown data, N/A, yes-liquid, yes-tablet, no | |

Congenital Adrenal Hyperplasia (CAH) Data Elements - Enrollment

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| 155 | If oral steroids are prescribed, name of product (enter N/A if not applicable) | Past Health History - Pharmacotherapy | Free Text | | |
| 156 | If on oral steroids, daily dose prescribed (enter 99999 if N/A or 88888 if unknown) | Past Health History - Pharmacotherapy | Decimal | | (mg/day) |
| 157 | If on oral steroids, frequency prescribed? | Past Health History - Pharmacotherapy | Pick list | Missing/unknown data, N/A - not prescribed, once/day, twice/day, three times/day, four times/day, only when ill, other | |
| 158 | At what age were oral steroids first initiated (enter 99999 if NA or 88888 if unknown) | Past Health History - Pharmacotherapy | Decimal | | (years, months) |
| 159 | Is fludrocortisone prescribed? | Past Health History - Pharmacotherapy | Pick list | Missing/unknown data, yes, no | |
| 160 | If on fludrocortisone, dose prescribed (enter 99999 if N/A or 88888 if unknown)? | Past Health History - Pharmacotherapy | Decimal | | (mg/day) |
| 161 | If on fludrocortisone, frequency prescribed? | Past Health History - Pharmacotherapy | Pick list | Missing/unknown data, N/A - not prescribed, once/day, twice/day, three times/day, four times/day, only when ill, other | |
| 162 | If on fludrocortisone, at what age was Florinef first initiated (enter 99999 if NA or 88888 if unknown)? | Past Health History - Pharmacotherapy | Decimal | | (years, months) |

Congenital Adrenal Hyperplasia (CAH) Data Elements - Enrollment

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| 163 | Other medications | Past Health History - Pharmacotherapy | Check boxes | Missing/unknown data, none, antianxiety, antidepressants, antihypertensives, antipsychotics, aromatase inhibitor - anastrozole, aromatase inhibitor - letrozole, calcitonin, calcium, contraceptives - oral, diuretics - calcium channel blocker, diuretics - loop diuretics, diuretics - potassium sparing, diuretics - thiazide, estrogen - oral, estrogen - patch, estrogen+progesterone-oral, estrogen+ progesterone-transdermal, estrogen+androgen-oral, estrogen-vaginal, GnRH analog, growth hormone, insulin, insulin sensitizers, iron, medroxyprogesterone - oral, testosterone - oral, sleeping | |
| 164 | If on growth hormone, at what age was growth hormone first initiated (enter 99999 if NA or 88888 if unknown)? | Past Health History - Pharmacotherapy | Decimal | | (years, months) |
| 165 | If on GnRH analog (Lupron), at what age was GnRH analog first initiated (enter 99999 if NA or 88888 if unknown)? | Past Health History - Pharmacotherapy | Decimal | | (years, months) |
| 166 | If on aromatase inhibitor, at what age was aromatase inhibitor first initiated (enter 99999 if NA or 88888 if unknown)? | Past Health History - Pharmacotherapy | Decimal | | (years, months) |
| 167 | Patient/primary caregiver was given the 24 hour on-call contact information for an endocrine provider? | Emergency Management | Pick List | Missing/unknown data, yes, no | |
| 168 | Patient/primary caregiver was given a written emergency medical letter specific to this condition? | Emergency Management | Pick List | Missing/unknown data, yes, no | |
| 169 | Patient/primary caregiver was given a sick day plan specific to this condition? | Emergency Management | Pick List | Missing/unknown data, yes, no | |
| 170 | Patient was enrolled in a web-based emergency alert program? | Emergency Management | Pick List | Missing/unknown data, yes - MEMSCIS, yes - other web-based program, no | |

Congenital Adrenal Hyperplasia (CAH) Data Elements - Enrollment

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| 171 | At time of enrollment, was patient provided instruction on how and when to administer an intramuscular hydrocortisone injection? | Emergency Management | Pick List | Missing/unknown data, N/A - non-classical form, no, yes | |
| 172 | Who provided the instruction on how and when to administer an intramuscular hydrocortisone injections? | Emergency Management | Pick List | Missing/unknown data, N/A - non-classical form, no, yes - provider, yes - nurse | |
| 173 | Was neuropsychological testing performed prior to enrollment? | Development | Pick List | Missing/unknown data, N/A - no neuropsych evaluation has been done on this patient, above average, average, below average | |
| 174 | Were behavioral concerns suspected prior to enrollment? | Development | Pick List | Missing/unknown data, yes (go to next question and explain), no | |
| 175 | If behavioral concerns were suspected prior to enrollment, explain | Development | Free Text | | |
| 176 | If behavioral concerns were suspected prior to enrollment, was patient referred for further evaluation at that time? | Development | Check boxes | Missing/unknown data, yes - to behavioral pediatrics, yes - to neuropsychologist, yes - to psychiatrist, yes - to psychologist, yes - to therapist/counselor, yes - to other | |
| 177 | Were there any psychological concerns related to this condition prior to enrollment? | Development | Pick List | Missing/unknown data, yes (go to next question and explain), no | |
| 178 | If psychological concerns were present prior to enrollment, explain | Development | Free Text | | |
| 179 | Was patient referred for Special Education evaluation prior to enrollment? | Education | Pick List | Missing/unknown data, yes, no | |

Congenital Adrenal Hyperplasia (CAH) Data Elements - Enrollment

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| 180 | Were Special Educational services received by this patient prior to enrollment? | Education | Pick List | Missing/unknown data, yes -IEP/IFSP, yes - 504 plan, yes - other, no | |
| 181 | If Special Educational services were received prior to enrollment: age (in years) child qualified for services? | Education | Pick List | Missing/unknown data, N/A - no Special Ed services were received, N/A - patient is >21 years old, <1, >1-2, >2-3, >3-4, >4-5, 6-10, 11-17, 18+ | |
| 182 | Reason Special Educational services were received prior to enrollment? | Education | Check boxes | Missing/unknown data, N/A (no Special Ed services are currently received), cognitive disability, fine motor disability, gross motor disability, learning disability, social-emotional disability, speech-language disability, other health impaired (OHI), other | |
| 183 | At time of enrollment, were educational materials relating to this condition provided to the parent/guardian and/or patient? | Education | Pick List | Missing/unknown data, no, yes | |
| 184 | If educational materials relating to this condition were provided, what were the materials? (enter 99999 if N/A, 88888 if unknown) | Education | Free Text | | |