

CAH due to 21-OHD Interval Survey: Infancy Through Puberty

New Measure	New Category	Answer type	Measure Answer	Units (for value box)
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	
Is patient followed by >1 endocrine center?	Demographics	Pick List	Missing/unknown data, yes, no	
If patient is followed by >1 endocrine center note which endocrine centers in which States	Demographics	Free Text		
If patient is followed by >1 endocrine center, did patient grant permission to share data via registry between treating endocrine centers?	Demographics	Pick List	Missing/unknown data, N/A, Yes, No	
If deceased, date of death (if N/A enter 01/01/1901)	Demographics	Date Field		
Current parental marital status	Socioeconomic Status	Check boxes	Missing/unknown data, married, not married	
Current insurance status	Socioeconomic Status	Check boxes	Missing/unknown data, commercial/private insurance, military insurance, none (self-pay), State program (newborn screening funds, special needs program, MCH block grant), State/Federal insurance (Medicaid/Medicare), Other	

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Has there been a change in family constellation since last visit?	Socioeconomic Status	Check boxes	Missing/unknown data, no change, yes-sibling, yes-adoption, yes-sibling death, yes-parent death	
Weight at this visit (enter 88888 if unknown or 77777 if not done)	Measurements	Decimal		(kg)
Height at this visit (enter 88888 if unknown or 77777 if not done)	Measurements	Decimal		(cm)
Head circumference (OFC) at this visit (enter 88888 if unknown or 77777 if not done)	Measurements	Decimal		(cm)
Blood pressure at this visit	Measurements	Pick List	Missing/unknown data, blood pressure not measured, hypertension, hypotension, normal blood pressure	
Date of last outpatient endocrine visit (if unknown enter 01/01/1901)	Interval Health History	Date Field		
Tanner Stage of pubic hair development	Interval Health History	Pick List	Missing/unknown data, not done, I, II, III, IV, V	
Tanner Stage of breast development	Interval Health History	Pick List	Missing/unknown data, N/A, not done, I, II, III, IV, V	

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Testicular volume (enter volume as left testicle, right testicle) (enter 99999 if N/A or 88888 if unknown or 77777 if not done)	Interval Health History	Decimal		(mL)
Has parent reported any change in patient's external genitalia?	Interval Health History	Pick List	Missing/unknown data, yes, no	
If patient has had a change in the size of the clitoris, enter as length, width in cm (enter 99999 if N/A or 88888 if unknown or 77777 if not done)	Interval Health History	Decimal		(cm)
If patient has penile enlargement, enter size of penis as length,width in cm (enter 99999 if N/A or 88888 if unknown or 77777 if not done)	Interval Health History	Decimal		(cm)
Clinical symptoms - manifestations presenting during this visit	Interval Health History	Check boxes	Missing/unknown data, none, acanthosis nigricans, acne, amenorrhea, anovulation, dehydration, developmental delay, growth acceleration, hirsutism, hypoglycemia, hypertonia, hypotonia, hyperpigmentation, insulin resistance, irregular menses, irritability, lethargy, male pattern hair loss, metabolic acidosis, osteopenia, osteoporosis, poor feeding, precocious puberty, pubic hair development, seizures, tachypnea, vaginal bleeding, vomiting, other (go to next question and explain)	
Other symptom(s) - manifestations presenting during this visit	Past Health History	Free Text		
Was genital surgery performed since the last outpatient endocrine visit?	Interval Health History - Surgical	Pick List	Missing/unknown data, N/A - no genital abnormalities indicating need for surgery, yes (complete Urogenital Survey now), no	

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Patient/primary caregiver knows how to reach the 24 hour on-call contact information for an endocrine provider?	Emergency Management	Pick List	Missing/unknown data, yes, no	
Patient/primary caregiver currently has a written emergency medical letter specific to this disorder?	Emergency Management	Pick List	Missing/unknown data, yes, no	
Patient/primary caregiver currently has a sick day plan specific to this disorder?	Emergency Management	Pick List	Missing/unknown data, yes, no	
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	
Has patient accessed their web-based emergency plan since the last outpatient endocrine visit?	Emergency Management	Pick List	Missing/unknown data, N/A - patient does not have a web-based emergency plan, yes - MEMSCIS, yes - other web-based emergency plan, no	
Number of emergency room visits since last outpatient	Emergency Management	Pick List	Missing/unknown data, 0,1,2,3,4,5,6,7,8,9,10, >10	
Number of hospital admissions (total) since last endocrine visit	Emergency Management	Pick List	Missing/unknown data, 0,1,2,3,4,5,6,7,8,9,10, >10	
Other health services received currently	Care Coordination	Check boxes	Missing/unknown data, none, audiology, behavioral/developmental pediatrics, cardiology, dermatology, endocrinology, hematology/oncology, home health care, nephrology, neurology, neuropsychology, occupational therapy, ophthalmology, orthopedics, otolaryngology, pulmonology, physical therapy, psychology, public health nursing, respiratory therapy, speech-language therapy, other	

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Community resources received currently	Care Coordination	Check boxes	Missing/unknown data, none, daycare, family support group related to this disorder, 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)	
Providers seen at this outpatient endocrine visit	Care Coordination	Check boxes	Missing/Unknown data, child and family life specialist, genetic counselor, general surgeon, geneticist, neuropsychologist, nurse, nurse practitioner, OB/GYN, physician assistant, psychologist, social worker, urology surgeon, other (go to next question and explain)	
Other providers seen at this outpatient endocrine visit (enter N/A if not applicable)	Care Coordination	Free Text		
Developmental screening occurred at this visit?	Development	Pick List	Missing/unknown data, yes, no	
Developmental screening tool used at this visit (if screening was done)	Development	Check boxes	Missing/unknown data, N/A - no developmental screening occurred at this visit, Ages and Stages Questionnaire, Ages and Stages - Social/Emotional Questionnaire, Denver (DDST-II), PEDS Questionnaire, provider history, other standardized screening tool	
Developmental milestones achieved at this time?	Development	Pick List	Missing/unknown data, N/A - no developmental screening occurred at this visit, yes, no	
If developmental milestone(s) not achieved, which one(s) were not achieved?	Development	Check boxes	Missing/unknown data, cognitive, fine motor, gross motor, social-emotional, speech-language, N/A - no developmental screening occurred at this visit, N/A - developmental screening occurred at this visit and all developmental milestones were achieved	

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If developmental milestones were not achieved, was patient referred for further developmental evaluation?	Development	Pick List	Missing/unknown data, N/A - no developmental screening occurred at this visit, yes, no	
Was neuropsychological evaluation done since last outpatient endocrine visit?	Development	Pick List	Missing/unknown data, yes (enter available data under separate Neuropsych Test Results Survey), no	
Overall neuropsychological testing impression (from most recent neuropsych evaluation)	Development	Pick List	Missing/unknown data, N/A - no neuropsych evaluation has been done on this patient, above average, average, below average	
Are behavioral concerns suspected at this time?	Development	Pick List	Missing/unknown data, yes (go to next question and explain), no	
If behavioral concerns are suspected at this time, explain	Development	Free Text		
If behavioral concerns are suspected at this time, was patient referred for further evaluation?	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	
Are there any psychological concerns related to this disorder?	Development	Pick List	Missing/unknown data, yes (go to next question and explain), no	
If psychological concerns are present, please explain	Development	Free Text		

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Was patient referred for Special Education evaluation at this time?	Education	Pick List	Missing/unknown data, yes, no	
Are Special Educational services received by this patient currently?	Education	Pick List	Missing/unknown data, yes -IEP/IFSP, yes - 504 plan, yes - other, no	
Special Educational services are received currently: age (in years) child qualified for services?	Education	Pick List	Missing/unknown data, N/A - no Special Ed services are currently received, N/A - patient is >21 years old, <1, >1-2, >2-3, >3-4, >4-5, 6-10, 11-17, 18+	
Reason Special Educational services are received currently?	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	
Lab tests collected at this visit	Laboratory Studies	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, 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, 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 17-ketosteroids - high, Urine (24 hours) for 17-ketosteroids - low, Urine (24 hours) for 17-hydroxycorticosteroids - WNL, Urine (24 hours) for 17-hydroxycorticosteroids - high, Urine (24 hours) for 17-hydroxycorticosteroids - low, Other	

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Other general lab tests (indicate type and if WNL or Abn) collected at this visit (enter 99999 if not applicable or 88888 if unknown)	Laboratory Studies	Free Text		
Serum 17-hydroxyprogesterone (17-OHP) level at this outpatient visit (enter 99999 if N/A or 88888 if unknown or 77777 if not done)	Laboratory Studies	Decimal		(ng/dL)
Serum testosterone (T) level at this outpatient visit (enter 99999 if N/A or 88888 if unknown or 77777 if not done)	Laboratory Studies	Decimal		(ng/dL)
Serum androstenedione (D4A) level at this outpatient visit (enter 99999 if N/A or 88888 if unknown or 77777 if not done)	Laboratory Studies	Decimal		(ng/dL)
Serum dehydroxyepiandrosterone (DHEA) level at this outpatient visit (enter 99999 if N/A or 88888 if unknown or 77777 if not done)	Laboratory Studies	Decimal		(ng/dL)
Serum dehydroxyepiandrosterone sulfate (DHEA-S) level at this outpatient visit (enter 99999 if N/A or 88888 if unknown or 77777 if not done)	Laboratory Studies	Decimal		(ng/dL)
Serum leutinizing hormone (LH) level at this outpatient visit (enter 99999 if N/A or 88888 if unknown or 77777 if not done)	Laboratory Studies	Decimal		(MIU/mL)

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Serum follicle stimulating hormone (FSH) level at this outpatient visit (enter 99999 if N/A or 88888 if unknown or 77777 if not done)	Laboratory Studies	Decimal		(IU/L)
Serum adrenocorticotrophic hormone (ACTH) level at this outpatient visit (enter 99999 if N/A or 88888 if unknown or 77777 if not done)	Laboratory Studies	Decimal		(pg/mL)
Renin level at this outpatient visit (enter 99999 if N/A or 88888 if unknown or 77777 if not done)	Laboratory Studies	Decimal		(ng/mL/hr)
Current biochemical evidence of osteopenia	Laboratory Studies	Check boxes	Missing/unknown data, N/A - no markers checked, serum osteocalcin-WNL, serum osteocalcin-high, serum osteocalcin-low, serum bone-specific alkaline phosphatase-WNL, serum bone-specific alkaline phosphatase-high, serum bone-specific alkaline phosphatase-low, serum total alkaline phosphatase-WNL, serum total alkaline phosphatase-high, serum total alkaline phosphatase-low, serum procollagen type 1 C-terminal peptide (PICP)-WNL, serum procollagen type 1 C-terminal peptide (PICP)-high, serum procollagen type 1 C-terminal peptide (PICP)-low, serum procollagen type 1 N-terminal peptide (PINP)-WNL, serum procollagen type 1 N-terminal peptide (PINP)-high, serum procollagen type 1 N-terminal peptide (PINP)-low, urinary CTX-telopeptide-WNL, urinary CTX-telopeptide-high, urinary CTX-telopeptide-low, urinary NTX-telopeptide-WNL, urinary NTX-telopeptide-high, urinary NTX-telopeptide-low, urinary deoxypyridinoline-WNL, urinary pyridinoline-high, urinary deoxypyridinoline-low, urinary pyridinoline-WNL, urinary deoxypyridinoline-high, urinary pyridinoline-low, bone sialoprotein (BSP)-WNL, bone sialoprotein (BSP)-high, bone sialoprotein (BSP)-low	

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Molecular testing: Common or targeted mutation panel done at this visit?	Laboratory Studies	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	
If common or targeted mutation panel performed, please list laboratory	Laboratory Studies	Free Text		
Molecular testing: Full sequencing done at this visit?	Laboratory Studies	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 mutations were detected	
Mutation description of patient: Allele 1 (format example 985A>G) (enter 99999 if N/A or 88888 if unknown)	Laboratory Studies	Free Text		
Mutation description of patient: Allele 2 (format example 985A>G) (enter 99999 if N/A or 88888 if unknown)	Laboratory Studies	Free Text		
Mutation description on biological mother: Allele 1 (format example 985A>G) (enter 99999 if N/A or 88888 if unknown)	Laboratory Studies	Free Text		
Mutation description on biological mother: Allele 2 (format example 985A>G) (enter 99999 if N/A or 88888 if unknown)	Laboratory Studies	Free Text		

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Mutation description on biological tather: Allele 1 (format example 985A>G) (enter 99999 if N/A or 88888 if unknown)	Laboratory Studies	Free Text		
Mutation description on biological father: Allele 2 (format example 985A>G) (enter 99999 if N/A or 88888 if unknown)	Laboratory Studies	Free Text		
Was dexa scan performed at this visit or since previous endocrine visit?	Imaging Studies	Check boxes	Missing/unknown data, N/A, hip, heel, pelvis, spine, total body, wrist, other	
If abnormal dexa scan (z-score \leq - 3 to - 2), specify site	Imaging Studies	Check boxes	Missing/unknown data, N/A, hip, heel, pelvis, spine, total body, wrist, other	
If abnormal dexa scan (z-score \leq - 4 to - 3), specify site	Imaging Studies	Check boxes	Missing/unknown data, N/A, hip, heel, pelvis, spine, total body, wrist, other	
If abnormal dexa scan (z-score \leq - 4), specify site	Imaging Studies	Check boxes	Missing/unknown data, N/A, hip, heel, pelvis, spine, total body, wrist, other	
Results of bone age done at this endocrine visit (enter N/A if not applicable)	Imaging Studies	Free Text		(years, months)
Method of reading bone age	Imaging Studies	Pick List	Missing/unknown data, N/A, bone age not done, Greulich Pyle, Tanner Whitehouse	

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Pelvic/testicular/genitogram performed at this visit or since last outpatient endocrine visit?	Imaging Studies	Pick list	Missing/unknown data, not done, normal, abnormal pelvic ultrasound, abnormal testicular ultrasound, abnormal - gonads not visualized, non-diagnostic	
If abnormal pelvic ultrasound results, explain finding (enter 99999 if N/A or 88888 if unknown)	Imaging Studies	Free Text		
If abnormal testicular ultrasound results, explain finding (enter 99999 if N/A or 88888 if unknown)	Imaging Studies	Free Text		
Are oral steroids prescribed?	Pharmacotherapy	Pick list	Missing/unknown data, N/A, yes-liquid, yes-tablet, no	
If oral steroids are prescribed, name of product (enter 99999 if N/A)	Pharmacotherapy	Free Text		
If on oral steroids, daily dose prescribed (enter 99999 if N/A)	Pharmacotherapy	Decimal		(mg/day)
If on oral steroids, frequency prescribed?	Pharmacotherapy	Pick list	Missing/unknown data, N/A - not prescribed, once/day, twice/day, three times/day, four times/day, only when ill, other	
Patient/primary caregiver reports oral steroids are taken as prescribed?	Pharmacotherapy	Pick list	Missing/unknown data, N/A - not prescribed, yes - 4-7 days/week, yes - 1-3 days/week, no (0 days/week)	

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If oral steroids are not taken as prescribed, reason given (enter 99999 if N/A)	Pharmacotherapy	Free text		
Number of episodes during which oral steroid stress doses were given since last outpatient endocrine visit (enter 99999 if N/A or 88888 if unknown)	Pharmacotherapy	Integer		
Number of episodes during which IM Soluortef were given since last outpatient endocrine visit (enter 99999 if N/A or 88888 if unknown)	Pharmacotherapy	Integer		
Is fludrocortisone prescribed?	Pharmacotherapy	Pick list	Missing/unknown data, yes, no	
If on fludrocortisone , dose prescribed (enter 99999 if N/A or 88888 if unknown)	Pharmacotherapy	Decimal		(mg/day)
If on fludrocortisone , frequency prescribed?	Pharmacotherapy	Pick list	Missing/unknown data, N/A - not prescribed, once/day, twice/day, three times/day, four times/day, only when ill, other	
Patient/primary caregiver reports fludrocortisone is taken as prescribed?	Pharmacotherapy	Pick list	Missing/unknown data, N/A - not prescribed, yes - 4-7 days/week, yes - 1-3 days/week, no (0 days/week)	
If fludrocortisone is not taken as prescribed, reason given (enter 99999 if N/A)	Pharmacotherapy	Free text		

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Other medications (see optional DocSite detailed medication survey also)	Pharmacotherapy	Check boxes	Missing/unknown data, none, antianxiety, antidepressants, antihypertensives, antipsychotics, aromatase inhibitor - anastrozole, aromatase inhibitor - letrozole, calcitonin, calcium, diuretics - calcium channel blocker, diuretics - loop diuretics, diuretics - potassium sparing, diuretics - thiazide, GnRH analog, growth hormone, insulin, insulin sensitizers, iron, sleeping medications, vitamin D, vitamins – daily, other	
If on growth hormone, weekly dose prescribed (enter 99999 if N/A, enter 66666 if discontinued)	Pharmacotherapy	Decimal		(mg/day)
If on growth hormone, frequency prescribed?	Pharmacotherapy	Pick list	Missing/unknown data, N/A - not prescribed, daily, every other day, twice a week, other	
If on GnRH analog, dose prescribed (enter 99999 if N/A, enter 66666 if discontinued)	Pharmacotherapy	Decimal		(mg/day)
If on GnRH analog, frequency prescribed?	Pharmacotherapy	Pick list	Missing/unknown data, N/A - not prescribed, every 4 weeks, every 3 months, annually, other	
If aromatase inhibitor prescribed, which brand or generic (enter 99999 if N/A, enter 66666 if discontinued)?	Pharmacotherapy	Free Text		
Method of payment for prescribed medications	Pharmacotherapy	Check boxes	Missing/unknown data, N/A - no medication prescribed, commercial/private insurance, military insurance, none - can't afford to fill medication prescription, self-pay, State program (newborn screening funds, special needs program, MCH block grant), State/Federal insurance (Medicaid/Medicare), Other	

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If any current prescribed medications used in treating this patient are not covered by some form of insurance, please list them (enter 99999 if N/A or 88888 if unknown)	Pharmacotherapy	Free Text		
Is patient on a special diet or regimen?	Nutrition	Pick List	Missing/unknown data, no, yes - go to next question.	
What is the special diet or regimen?	Nutrition	Free Text		