Delayed Diagnosis of Classic Congenital Adrenal Hyperplasia Secondary to False-Negative Newborn Screening Results

Bedour Jafar, MB, BCh, BAO, Marissa J. Kilberg, MD, MSEd, and Maria G. Vogiatzi, MD

False-negative newborn screening data for congenital adrenal hyperplasia are limited and have not included Pennsylvania and New Jersey. Within our program, we identified 16 missed cases of 96 patients born in Pennsylvania and New Jersey between 1998 and 2024, highlighting the need for continued evaluation in clinically suspicious circumstances despite normal screening. *(J Pediatr 2025;283:114597).*

Classical congenital adrenal hyperplasia (CAH) attributable to 21-hydroxylase deficiency is characterized by adrenal insufficiency. Severe aldosterone deficiency resulting in electrolyte abnormalities and dehydration is seen in most patients, ie, salt-wasting (SW).[1] The newborn screening (NBS) for CAH was established to prevent a potentially life-threatening adrenal crisis in the newborn.[2] The screening is determined by the measurement of 17 hydroxyprogesterone (17OHP) concentrations, the biomarker for 21-hydroxylase deficiency, by fluoroimmunoassay on filter paper blood spots.[2] Currently, all NBS programs in the US screen for CAH.[3] Considerable challenges of the screening are the frequent false-positive results, seen primarily babies with low birth weight and prematurity.[2] To address this concern, programs have developed various protocols, such as weight and/or gestational age-adjusted 17OHP criteria.[2,4] Several programs have implemented additional procedures, such as a second-tier screening using measurement of 17OHP and/or other steroids with other methodologies (eg, liquid chromatography–tandem mass spectrometry).[5] A small number of programs also include a second screen, usually at 8-14 days of life.[6-9] Overall, there is no standardized approach across states, and specific 17OHP cutoffs vary considerably.[2,4]

Although NBS efforts have focused on reducing the number of false-positive cases, the sensitivity for detecting SW CAH is generally reported as 100%.[9,10] A case series from Minnesota, however, identified a 22.5% false-negative rate for classical disease.[11] The missed cases included children with genital atypia and SW. High false-negatives rates for classical CAH also have been reported from Wisconsin and Indiana.[12,13] All of these states screen infants once, at 24-48 hours of life. Some states, like Texas and Colorado, apply a second screening during the second week of life and observed that approximately 15%-30% of cases of classical CAH were missed with the first screening but identified with the second.[6-9] Although the exact false-negative rate for these states is uncertain, their data support that one-time screening shortly after birth is associated with high rates of missed cases.

In this report, we assessed missed cases for classical CAH within our program. Our patient population is primarily from Pennsylvania and New Jersey, and therefore, our findings reflect the Pennsylvania and New Jersey NBS program results. In both Pennsylvania and New Jersey, blood spot testing for NBS is obtained at 24-48 hours of life. In the state of New Jersey, however, additional screening occurs if there is prolonged hospital admission. False-negative results from these states have yet to be reported.

Methods

We performed a retrospective chart review of 204 medical records of children diagnosed with CAH at our institution from 1998 to 2024. Patients with classical CAH secondary to other enzymatic deficiencies or nonclassical CAH were excluded. One hundred thirteen patients with classical CAH secondary to 21-hydroxylase deficiency were identified (**Figure**). Of 113 patients, 96 patients were born in either Pennsylvania (n = 68) or New Jersey (n = 28). The remaining 17 patients were born in other states and were excluded because of low sample size and risk of biased sampling from those states. Data collected included age of presentation, genotype, phenotype, laboratory evaluations at presentation, and NBS results. Both Pennsylvania and New Jersey used the same fluoroimmunoassay for 17OHP measurement in filter paper blood spot samples (DELFIA TRF [time-resolved fluorescence] assay; Perkin Elmer) using a Revvity GSP analyzer. Samples are collected between 24 and 48 hours of life. Cutoff values are set according to birth weight, albeit they vary between the 2 states. In Pennsylvania, if the initial assessment indicates

17OHP	17 hydroxyprogesterone
CAH	Congenital adrenal hyperplasia
NBS	Newborn screening
SW	Salt wasting

From the Department of Endocrinology and Diabetes, Children's Hospital of Philadelphia, Philadelphia, PA

0022-3476/$ - see front matter. © 2025 Elsevier Inc. All rights are reserved, including those for text and data mining, AI training, and similar technologies.
https://doi.org/10.1016/j.jpeds.2025.114597

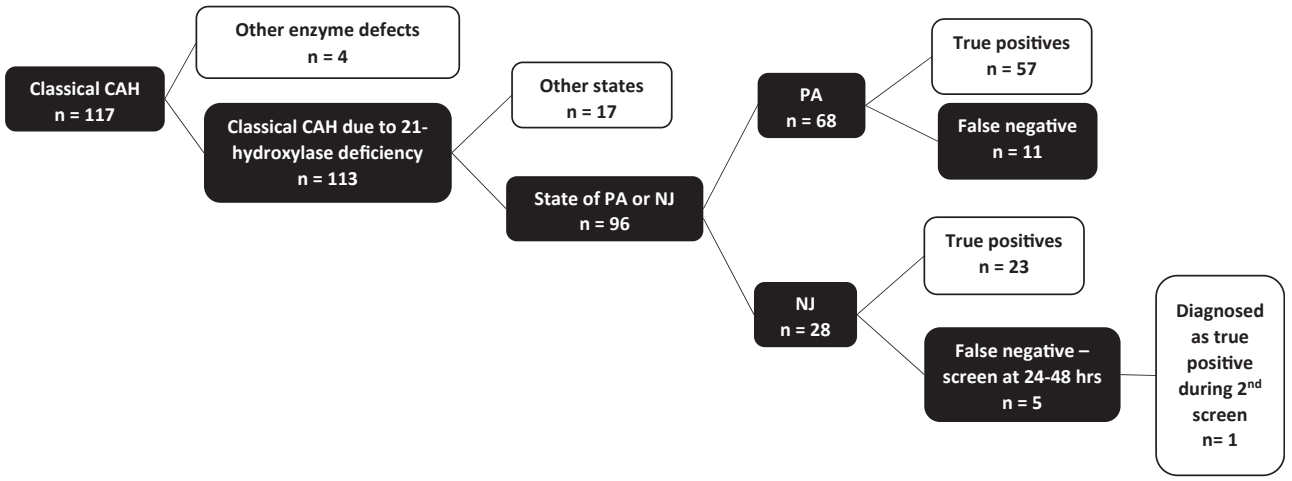

Figure. Flow diagram of patients with CAH followed by our program demonstrating patient identification, inclusion, and exclusion. *NJ*, New Jersey; *PA*, Pennsylvania.

an elevated 17 OHP value, a second-tier screening is undertaken with repeat 17OHP measurement of the original sample after extraction using again an immunoassay, which separates structurally similar steroids to distinguish true values from false-positive results. The results are reported according to a specific cutoff concentration (eg, values lower than 19 ng/mL or 57.5 nmol/L are reported as normal). There is no second screen. In contrast, in New Jersey, all hospitalized newborns have a second screening at 7, 14, 21 days, or at time of discharge regardless of initial screening result, which is performed at 24-48 hours of life. Results of the New Jersey screening are reported on the basis of on the birth weight. False-negative results were confirmed with the state NBS laboratories. The study was approved by the institutional review board for a retrospective chart review with a Health Insurance Portability and Accountability Act (HIPAA) waiver, for all patients with differences of sexual development, including CAH.

Results

Among our 96 patients with classical CAH, we identified 16 children (11 Pennsylvania, 5 New Jersey) who were missed by NBS (**Figure**, **Table**). Therefore, the rate of false-negative NBS results was 16.7% in our program. Subjects were mostly male (10/16 [62.5%]) and included children with SW (5/16 [31%]) and genital atypia (5/16[31%]). Age at presentation ranged from birth to 8.4 years. Female children presented early in life with genital atypia (2 weeks-2 years). Male children often presented later, with premature adrenarche with associated tall stature, growth acceleration, and significant bone age advancement (bone age minus chronologic age ranged from 1.6 to 8 years). However, there was 1 male patient who presented with SW. There was an overlap in 17OHP baseline or stimulated values between subjects who presented with electrolyte disturbance and those who did not (**Table**). One patient was diagnosed prenatally through genetic testing as the result of family history, in which mother was treated with prenatal glucocorticoids from 6 to 23 weeks of pregnancy (case 15 in the **Table**). No evidence of prenatal glucocorticoid use was found for the other 15 patients. One female patient, who was screened negative for CAH by New Jersey at 24-48 hours of life, remained hospitalized for evaluation of genital atypia. A second screen was obtained before discharge at 8 days of life, as per requirements in New Jersey for hospitalized babies, and returned as positive (case 16 in the **Table**). These 2 cases were included in our review because, in the absence of prenatal testing or genital atypia (ie, if baby was male with normal genital development), these cases would have been missed.

Discussion

In this report, we observed that 16.7% of our patients with classical CAH were missed by NBS. Our data describe a unique case series of false-negative NBS results for the states of Pennsylvania and New Jersey, reflective of a single institution's experience. Our institution is a quaternary referral center for CAH, which may have introduced a referral bias. Overall false-negative rates for the states of Pennsylvania and New Jersey remain uncertain, because there is no consistent way of reporting missed cases back to the states. False-negative rates also may have been underestimated, because some babies may not survive an undiagnosed adrenal crisis.

Our cases highlight that babies with SW disease can escape detection, thereby missing a primary aim of NBS. Consistent with previous reports, most subjects had no history of electrolyte abnormalities, and thus, presented later in life, indicating a milder phenotype than those with SW.[9] Regardless, detection of all babies with classical disease remains

Table. Description of case presentations including rationale for further testing (clinical presentation), laboratory assessment resulting in diagnosis, genetic testing (where available) and CAH clinical diagnosis

Cases (no.)	NBS state	Sex	Gestational age (wk + d)	Birth weight, kg	NBS 17OHP, ng/mL, (normal range)*	NBS collected at hours of life	At diagnosis				
							Clinical presentation	Age	BA, y	17OHP, ng/dL Baseline/ post-ACTH†	Cortisol, μg/dL Baseline/→ post ACTH†	Genotype (allele 1/allele 2)
1	PA	F	37 + 4	2.96	14.7 (<30)	34	Clitoromegaly, posterior labial fusion	2 wk	NA	6632/24 115	6.1/6.2	Deletion/In2G, P30 L
2	PA	F	41 + 2	3.5	12 (<25)	34	Clitoromegaly, posterior labial fusion, UG sinus. Electrolyte abnormalities.	1 m	NA	9400	5.2	NA
3	PA	F	36	2.87	9.5 extracted (<19)	45	Clitoromegaly, posterior labial fusion, urogenital sinus. Electrolyte abnormalities	5 wk	NA	9460	1.23	In2G/R356 W
4	PA	M	39	4.29	18.1 (<25)	28	Positive family history	13 mo	NA	12 920	7.6	In2G/p.I172 N
5	PA	F	40	3.6	14.7 extracted (<19)	35	Clitoromegaly	2 y	NA	3932/32 925	NA	In2G/p.I172 N
6	PA	M	39	3.99	23.2 (<30)	24	Premature adrenarche, rapid growth/tall stature advanced BA	3.5 y	11.5	36 268	12.5	In2G/p.172 N
7	PA	M	39	3.18	Normal (<25)	24	Premature adrenarche, rapid growth/tall stature, advanced BA, large phallus	4.2 y	10	16 500/24 100	2.7/3.2	p.I172 N/p.I172 N
8	PA	M	40	3.11	28.1 (<25) 3.4 extracted (<19)	48	Premature adrenarche, rapid growth/tall stature advanced BA	5 y	12	2860	4.1	p.I72 N/R356 W
9	PA	M	39 + 6	4.08	39.4 (<25) 8.6 extracted (<19)	48	Premature adrenarche, rapid growth/tall stature, advanced BA	5 y	11	19 081	4.2	NA
10	PA	M	40	3.62	23 (<25)	36	Premature adrenarche, rapid growth, advanced BA, large phallus	5 y	11	13 034	5.7	NA
11	PA	M	39 + 3	3.15	32 (<25) 5.9 extracted (<19)	24	Premature adrenarche, rapid growth/tall stature, advanced BA	6.5 y	13	13 348/24 182	3.2/3.2	In2G/p.I172 N
12	NJ	M	40 + 3	4.1	Normal (<35)	42	Premature adrenarche, rapid growth/tall stature, advanced BA	4 y	9	13 446	NA	p.I172 N/p.I172 N
13	NJ	M	39	3.97	7 (<35)	24	Premature adrenarche, rapid growth, advanced BA	8.4 y	10	123/16 755	1.6/7	NA
14	NJ	M	30	1.47	29 (<100) Second screen: 93 (<100) at dol# 7	48	SW crisis	5 wk	NA	10 900	NA	Deletion/p.172 N
15‡	NJ	F	39	3.571	Normal (<35)	39	Positive family history-amniocentesis	Birth	NA	2158	NA	In2G/p.I172 N
16§	NJ	F	40	4.165	Normal (<35) Second screen: 76 (<54) at dol# 8	47	Clitoromegaly, posterior labial fusion, urogenital sinus	2 wk	NA	12 100	5.43	p.I172 N/p.I172 N

BA, bone age; *dol*, day of life; *F*, female; *M*, male; *NA*, not available; *NJ*, New Jersey; *PA*, Pennsylvania.

*PA NBS 17OHP (ng/mL) cut-off values according to birth weight (g): ≤130 for 1001-1500 g; ≤60 for 1501-2000 g; ≤ 47 for 2001-2250 g; ≤45 for 2251-2500 g; ≤30 for 2501-3000 g; ≤25 for >3000 g. If values are greater than these cutoffs, an extracted 17OHP is measured with cut-off 19 ng/mL for all birth weights. NJ NBS 17OHP (ng/mL) cut-off values according to birth weight (g): <100 for <1500 g; <60 for 1500-2499; <40 for 2500-2999; <35 for ≥3000.

†If only one cortisol or 17OHP value is available, adrenocorticotropic hormone stimulation test was not performed.

‡Mother received dexamethasone as prenatal treatment during 6-23 weeks of gestation.

§The baby presented with genital atypia at birth and remained hospitalized for 8 days for laboratory assessment and management. Although the first NBS was negative, the second NBS before discharge was positive. The state of NJ requires a one-time screen, unless the baby remains hospitalized, in which case repeat testing is obtained.

important because these children have adrenal insufficiency and may experience a life-threatening adrenal crisis during physiologic stress. As our data indicate, they may also face a late diagnosis and other potential harms, eg, precocious puberty, markedly advanced bone age, and stunted adult height. Our results are similar to those observed in other states, such as Minnesota and Indiana.[11,13]

Concentrations of 17OHP in the newborn are affected by birth weight, gestational age, concomitant illness, or time of blood sample collection.[14] In healthy babies, 17OHP levels are typically greater at birth and decline to stable concentrations over the course of weeks.[15] In some newborns with classic CAH, 17-OHP concentrations may be within the accepted cutoff values at the time of birth and increase over time, resulting in false-negative NBS for samples collected around 24-48 hours of life. The reason behind the delayed increase in 17OHP values in CAH not fully understood. It is postulated that these babies have increased glucocorticoid sensitivity to maternal cortisol,[2] such as seen with certain glucocorticoid receptor haplotypes.[16] Maternal cortisol, crossing into fetal circulation toward the end of gestation,[17,18] may suppress the fetal hypothalamic-pituitary-adrenal axis. Similarly, prenatal exposure to certain exogenous glucocorticoids such as betamethasone and dexamethasone can also suppress 17OP values in the newborn and lead to false-negative NBS results. Antenatal maternal treatment with glucocorticoids has been shown to lower 17-OHP by close to 30%.[19] In our series, case 15 may be explained with prenatal treatment with dexamethasone, although one may have anticipated a recovery of 17OHP values by the time of birth, because treatment was stopped at 23 weeks of gestation. No other baby in our series had an identifiable history of glucocorticoid exposure. In an interesting case report of a false-negative NBS result, as an example, maternal treatment with triamcinolone as nasal spray throughout pregnancy was found to result in detectable circulating triamcinolone levels in the newborn.[20] As glucocorticoid use has become widespread, its impact on NBS results may be hard to assess.

To reduce false-negative rates, all NBS programs recommend sample collection after 24 hours of life.[2] They then implement screening algorithms on the basis of gestational age and/or birth weight, although protocols and 17OHP cut-offs vary.[2,4] Given the delayed increase in 17OHP concentrations in some newborns with classic CAH, a second screening around the second week of life is employed by some states.[7] However, false-negative results continue to occur, although at rates lower than those with a one-time screen.[7] In contrast to a study in Texas, in which researchers observed a diminished false-positive rate when adjusting weight-based cut-offs, a change in cut-off values in our cohort would not drastically reduce false-negative rates, because of the patients having 17OHP values well within the normal NBS range.[9] Furthermore, it has been argued that a second screen is not cost-effective, with estimates in the range of $25 000 to $250 000/case with a second screen.[21-23] Alternative approaches adopted by some states is a second-tier screening using liquid chromatography–tandem mass spectrometry for 17OHP and additional steroids, such as androstenedione and cortisol.[24] Such screening has not been shown to diminish false-negative rates in Minnesota.[24] Application of second tier screening with measurement of 21-deoxycortisol, a biomarker specific for 21-hydroxylase deficiency, has been proposed to improve the diagnostic accuracy of NBS.[25,26] Although the experience is limited, data from Netherlands are encouraging.[26] A recent report using molecular testing identified its own unique challenges as the differential between carrier status and disease without parental testing can be difficult in some cases.[27] These efforts highlight the difficulties around the diagnostic accuracy of NBS and the need for further research.

Our study has several limitations. Given the retrospective design, there is risk of missing or inaccurate data. We do not have specific state data to calculate false-negative rates for the Pennsylvania and New Jersey NBS programs but rather are extrapolating on the basis of the findings at our quaternary care clinic. Nonetheless, our study's findings are important, as they indicate that a significant number of patients were missed during NBS. Given the variability among NBS programs in both 17OHP cutoffs and additional procedures, providers need to be aware about the specific metrics of the state in which practice and the limitations of the testing obtained. It is critical that the pediatrician maintain a keen index of suspicion as clinically indicated with thorough family history and physical examination to identify subtle ambiguity and prompt further workup despite normal screening results.

In conclusion, our findings support that high false-negative NBS rates can occur in many regions of the country, particularly among states that apply a single screening at 24-48 hours of life. This report serves as a reminder to practitioners that classic CAH can be missed on NBS and that they need to maintain a high index of suspicion for the disease when faced with clinical evidence of androgen excess or adrenal insufficiency. This study also highlights a need to continue to assess opportunities at the laboratory level to minimize false-negative rate with options such as routine second screening or second tier adrenal biomarkers, where further data are needed. ■

CRediT authorship contribution statement

Bedour Jafar: Writing – review & editing, Writing – original draft, Methodology, Investigation, Data curation, Conceptualization. **Marissa J. Kilberg:** Writing – review & editing, Writing – original draft, Supervision, Project administration, Methodology, Investigation, Conceptualization. **Maria G. Vogiatzi:** Writing – review & editing, Writing – original draft, Supervision, Methodology, Conceptualization.

Declaration of competing interest

M.V. reports research support from Neurocrine Biosciences, Spruce Biosciences, and Adrenas Therapeutics; and

consulting for Crinetics and Eton Pharmaceuticals. There are no other conflicts of interest.

Submitted for publication Dec 27, 2024; last revision received Apr 10, 2025; accepted Apr 14, 2025.

Reprint requests: Marissa Kilberg, MD, MSEd, Department of Endocrinology and Diabetes, Children's Hospital of Philadelphia, 3500 Civic Blvd, Philadelphia, PA 19104. E-mail: kilbergm@chop.edu

References

1. Claahsen-van der Grinten HL, Speiser PW, Ahmed SF, Arlt W, Auchus RJ, Falhammar H, et al. Congenital adrenal hyperplasia—current insights in pathophysiology, diagnostics, and management. Endocr Rev 2022;43:91-159. https://doi.org/10.1210/endrev/bnab016
2. Held PK, Bird IM, Heather NL. Newborn screening for congenital adrenal hyperplasia: review of factors affecting screening accuracy. Int J Neonatal Screen 2020;6:67.
3. Edelman S, Desai H, Pigg T, Yusuf C, Ojodu J. Landscape of congenital adrenal hyperplasia newborn screening in the United States. Int J Neonatal Screen 2020;6:64.
4. Speiser PW, Chawla R, Chen M, Diaz-Thomas A, Finlayson C, Rutter MM, et al. Newborn screening protocols and positive predictive value for congenital adrenal hyperplasia vary across the United States. Int J Neonatal Screen 2020;6:37. https://doi.org/10.3390/ijns6020037
5. Sarafoglou K, Gaviglio A, Hietala A, Frogner G, Banks K, McCann M, et al. Comparison of newborn screening protocols for congenital adrenal hyperplasia in preterm infants. J Pediatr 2014;164:1136-40. https://doi.org/10.1016/j.jpeds.2014.01.038
6. Chan CL, McFann K, Taylor L, Wright D, Zeitler PS, Barker JM. Congenital adrenal hyperplasia and the second newborn screen. J Pediatr 2013;163:109-13.e1. https://doi.org/10.1016/j.jpeds.2013.01.002. Epub 2013 Feb 12. Erratum in: J Pediatr. 2013 Jul;163(1):308. PMID: 23414665.
7. Eshragh N, Doan LV, Connelly KJ, Denniston S, Willis S, LaFranchi SH. Outcome of newborn screening for congenital adrenal hyperplasia at two time points. Horm Res Paediatr 2020;93:128-36. https://doi.org/10.1159/000508075
8. Held PK, Shapira SK, Hinton CF, Jones E, Hannon WH, Ojodu J. Congenital adrenal hyperplasia cases identified by newborn screening in one- and two-screen states. Mol Genet Metab 2015;116:133-8. https://doi.org/10.1016/j.ymgme.2015.08.004
9. Therrell BL, Berenbaum SA, Manter-Kapanke V, Simmank J, Korman K, Prentice L, et al. Results of screening 1.9 million Texas newborns for 21-hydroxylase-deficient congenital adrenal hyperplasia. Pediatrics 1998;101:583-90. https://doi.org/10.1542/peds.101.4.583
10. Votava F, Török D, Kovács J, Mösslinger D, Baumgartner-Parzer SM, et al., Middle European Society for Paediatric Endocrinology—Congenital Adrenal Hyperplasia (MESPE-CAH) Study Group. Estimation of the false-negative rate in newborn screening for congenital adrenal hyperplasia. Eur J Endocrinol 2005;152:869-74. https://doi.org/10.1530/eje.1.01929
11. Sarafoglou K, Banks K, Kyllo J, Pittock S, Thomas W. Cases of congenital adrenal hyperplasia missed by newborn screening in Minnesota. JAMA 2012;307:2371-4. https://doi.org/10.1001/jama.2012.5281
12. Varness TS, Allen DB, Hoffman GL. Newborn screening for congenital adrenal hyperplasia has reduced sensitivity in girls. J Pediatr 2005;147:493-8. https://doi.org/10.1016/j.jpeds.2005.04.035
13. Saroufim R, Nebesio TD, Eugster EA. Characteristics of patients with classic congenital adrenal hyperplasia missed on the newborn screen. Horm Res Paediatr 2024;97:470-6. https://doi.org/10.1159/000535405
14. Anandi VS, Shaila B. Evaluation of factors associated with elevated newborn 17-hydroxyprogesterone levels. J Pediatr Endocrinol Metab 2017;30:677-81. https://doi.org/10.1515/jpem-2016-0459
15. Gruñeiro DPL, Prieto L, Chiesa A, Bengolea S, Bergadá C. Congenital adrenal hyperplasia and early newborn screening: 17α-hydroxyprogesterone (17α-OHP) during the first days of life. J Med Screen 1998;5:24-6. https://doi.org/10.1136/jms.5.1.24
16. Lockett J, Inder WJ, Clifton VL. The glucocorticoid receptor: isoforms, functions, and contribution to glucocorticoid sensitivity. Endocr Rev 2024;45:593-624. https://doi.org/10.1210/endrev/bnae008
17. Howland MA, Sandman CA, Glynn LM. Developmental origins of the human hypothalamic-pituitary-adrenal axis. Expert Rev Endocrinol Metab 2017;12:321-39. https://doi.org/10.1080/17446651.2017.1356222
18. Zhu P, Wang W, Zuo R, Sun K. Mechanisms for establishment of the placental glucocorticoid barrier, a guard for life. Cell Mol Life Sci 2019;76:13-26. https://doi.org/10.1007/s00018-018-2918-5
19. Gatelais F, Berthelot J, Beringue F, Descamps P, Bonneau D, Limal JM, et al. Effect of single and multiple courses of prenatal corticosteroids on 17-hydroxyprogesterone levels: implication for neonatal screening of congenital adrenal hyperplasia. Pediatr Res 2004;56:701-5. https://doi.org/10.1203/01.PDR.0000142733.50918.6E
20. Houang M, Nguyen-Khoa T, Eguether T, Ribault B, Brabant S, Polak M, et al. Analysis of a pitfall in congenital adrenal hyperplasia newborn screening: evidence of maternal use of corticoids detected on dried blood spot. Endocr Connect 2022;11:e220101. https://doi.org/10.1530/EC-22-0101
21. Brosnan CA, Brosnan P, Therrell BL, Slater CH, Swint JM, Annegers JF, et al. A comparative cost analysis of newborn screening for classic congenital adrenal hyperplasia in Texas. Public Health Rep 1998;113:170-8.
22. Carroll AE, Downs SM. Comprehensive cost-utility analysis of newborn screening strategies. Pediatrics 2006;117:S287-95. https://doi.org/10.1542/peds.2005-2633H
23. Yoo BK, Grosse SD. The cost effectiveness of screening newborns for congenital adrenal hyperplasia. Public Health Genomics 2009;12:67-72. https://doi.org/10.1159/000156115. Epub 2008 Sep 14. Erratum in: Public Health Genomics. 2018;21(1-2):100. doi: 10.1159/000495265. PMID: 19039250; PMCID: PMC6706242.
24. Sarafoglou K, Banks K, Gaviglio A, Hietala A, McCann M, Thomas W. Comparison of one-tier and two-tier newborn screening metrics for congenital adrenal hyperplasia. Pediatrics 2012;130:e1261-8. https://doi.org/10.1542/peds.2012-1219
25. Miller WL. Congenital adrenal hyperplasia: time to replace 17OHP with 21-deoxycortisol. Horm Res Paediatr 2019;91:416-20. https://doi.org/10.1159/000501396
26. Stroek K, Ruiter A, van der Linde A, Ackermans M, Bouva MJ, Engel H, et al. Second-tier testing for 21-hydroxylase deficiency in The Netherlands; a newborn screening pilot study. J Clin Endocrinol Metabol 2021;106:e4487-996. https://doi.org/10.1210/clinem/dgab464
27. Sarafoglou K, Gaviglio A, Wolf C, Lorentz CP, Lteif A, Kyllo J, et al. Can incorporating molecular testing improve the accuracy of newborn screening for congenital adrenal hyperplasia? J Clin Endocrinol Metab 2024;110:e1194-12030.