Birth Incidence, Age at Diagnosis, Mortality in Congenital Adrenal Hyperplasia in Korea: A Nationwide Population-Based Study

dongguk UNIVERSITY MEDICAL CENTER

Adrenals & HPA Axis 1
P1-P007

Department of Pediatrics[1], Dongguk University Ilsan Hospital, Goyang, Republic of Korea
Department of Otorhinolaryngology[2], College of Medicine, Konyang University, Daejon, Republic of Korea
Ji Hyun Kim[1], Jong Bin Lee[2]

INTRODUCTION

Congenital adrenal hyperplasia (CAH) is a disease caused by a decrease in the synthesis of cortisol and aldosterone, which results in adrenocortical dysfunction. The incidence of CAH has been reported mainly through neonatal screening tests which detect for 21-OHD. The worldwide incidence including all other enzyme deficiencies which is not detected by neonatal screening has rarely been reported. It has been reported that 21-OHD occurs in one in 15,000 people (6.6 per 100,000) per year through neonatal screening. CAH is a rare disease, studies on large populations are needed to know the actual incidence without using the screening test.

CAH is known to have high mortality and has been reported that adrenal crisis usually leads to death. In Korea, neonatal screening for 21-OHD was performed nationwide since 2006 and CAH has been designated as a rare intractable disease (RID) since 2008. The RID database is integrated into the national health insurance (NHI) system to record all healthcare information. We investigated the birth incidence, age at diagnosis, and mortality of CAH patients using this database.

OBSECTIVES & METHODS

This study used claims data from the National health insurance (NHI) database and registration data from the rare intractable disease (RID) database. The Korean government implemented a NHI program for all citizens, which covers more than 50 million Koreans. These data, which are recorded in the NHI database, contain information from the time of a patient's diagnosis and thereafter, including the diagnosis, demographics, prescription history, surgical records, and screening history. Patients' diagnostic information was recorded according to the International Classification of Diseases, 10th Revision (ICD-10).

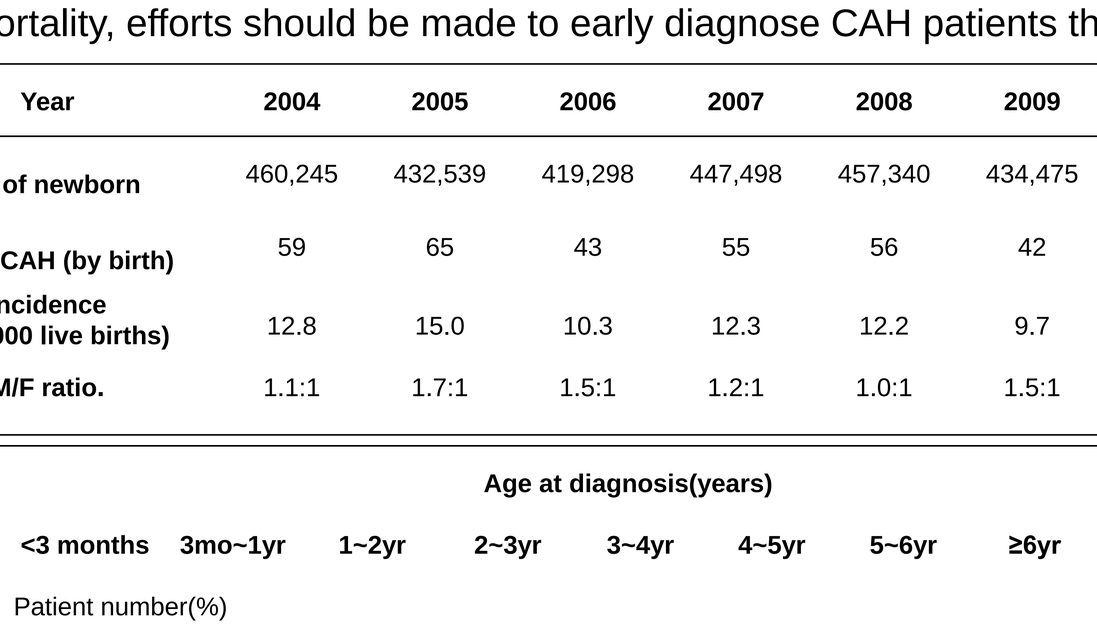
RESULTS & CONCLUSIONS

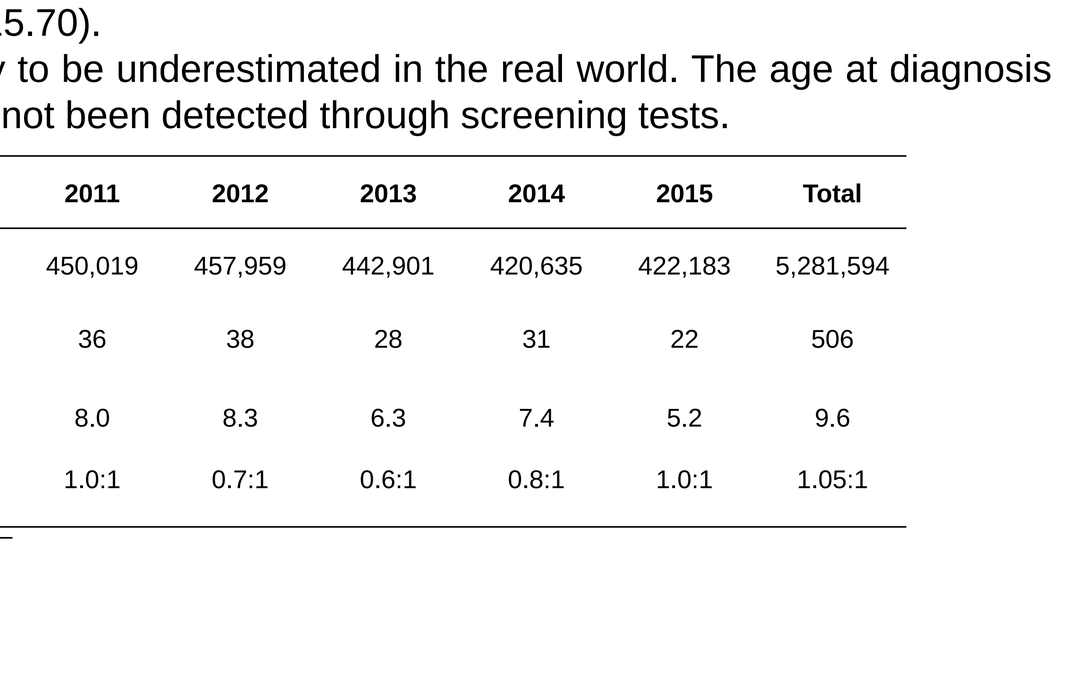
From 2004 to 2015, 506 CAH patients were born in Korea and CAH annual birth incidence was 9.6 / 100,000 live births. The age at diagnosis ranged from 0 to 8 years. Within 3 months of age, total 83.8% were diagnosed, about 15% of all patients were not diagnosed through the neonatal screening test. The number of confirmed deaths was 9, and this represents 1.8% of all CAH patients. The effect of the screening test on mortality was not statistically significant (HR 3.40, CI 0.42–27.68). Interestingly, the age at diagnosis was related to death, diagnosed after 3 months had a statistically significant 4-fold higher risk of death (HR 4.06, CI 1.07-15.70).

As our results show, CAH incidence data through neonatal screening test is likely to be underestimated in the real world. The age at diagnosis affects mortality, efforts should be made to early diagnose CAH patients that have not been detected through screening tests.

Year	2004	2005	2006	2007	2008	2009	2010	2011	2012	2013	2014	2015	Total
No. of newborn	460,245	432,539	419,298	447,498	457,340	434,475	436,502	450,019	457,959	442,901	420,635	422,183	5,281,594
No. of CAH (by birth)	59	65	43	55	56	42	31	36	38	28	31	22	506
Incidence (/100,000 live births)	12.8	15.0	10.3	12.3	12.2	9.7	7.1	8.0	8.3	6.3	7.4	5.2	9.6
M/F ratio.	1.1:1	1.7:1	1.5:1	1.2:1	1.0:1	1.5:1	1.2:1	1.0:1	0.7:1	0.6:1	0.8:1	1.0:1	1.05:1

	Age at diagnosis(years)								
	<3 months	3mo~1yr	1~2yr	2~3yr	3~4yr	4~5yr	5~6yr	≥6yr	Total
	Patient number(%)								
Male	235(88.0)	5(1.9)	8(3.0)	7(2.6)	3(1.1)	3(1.1)	2(0.8)	4(1.5)	267(100)
Female	189(79.1)	18(7.5)	12(5.0)	7(2.9)	3(1.3)	2(0.8)	3(1.3)	5(2.1)	239(100)
Total	424(83.8)	23(4.5)	20(3.9)	14(2.8)	6(1.2)	5(1.0)	5(1.0)	9(1.8)	506(100)





REFERENCES

1. Merke DP, Bornstein SR. Congenital adrenal hyperplasia. Lancet. 2005;365(9477):2125-36.
2. National Newborn Screening Information System [Internet]. 2009. Available from: http://www2.uthscsa.edu/nnsis.
3. Speiser PW, Azziz R, Baskin LS, Ghizzoni L, Hensle TW, Merke DP, et al. Congenital adrenal hyperplasia due to steroid 21-hydroxylase deficiency: an Endocrine Society clinical practice guideline. The Journal of clinical endocrinology and metabolism. 2010;95(9):4133-60.