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.

OBJECTIVES & 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.

REFERENCES