Background-Aim
Congenital adrenal hyperplasia (CAH) is the most common form of primary adrenal insufficiency in children. 21-hydroxylase enzyme deficiency (21-OHD) occurs in 90 to 95% of all cases of CAH. Despite it being a treatable condition, if unrecognized, CAH may present with life-threatening cardiovascular collapse. Mortality in the first years is reported to be higher than in the general population. Neonatal screening for CAH is effective in detecting the salt-wasting form and thereby reducing mortality. This study describes the incidence of CAH in Turkey and analyses the results obtained from a pilot study of public CAH screening program of Turkish Directorate of Public Health comprising four cities of Turkey.

Method
A pilot newborn CAH screening study was carried out under the authority of Turkish Directorate of Public Health. Newborn babies ≥32 gestational weeks and ≥1500 gram birth weight from four cities between March 27-September 15, 2017 were included. Screening protocol included one sample two-tier testing. In the first step, 17α-hydroxyprogesterone (17-OHP) was measured by fluorimunoassay in dried blood spots obtained at 3-5th days of life. The cases with positive initial screening were tested by steroid profiling in dried blood spots using liquid chromatography–tandem mass spectrometry method to measure 17-OHP, 21-deoxycortisol, cortisol, 11-deoxycortisol and androstenedione as a second-tier test. The babies with steroid ratio of (21-deoxycortisol+17-OHP)/cortisol>0.5 were referred to pediatric endocrinology clinics for diagnostic assessment (Figure 1).

Results
38,935 infants were tested, 2265 (5.82%) had second-tier testing, and 212 (0.54%) were referred for clinical assessment, 6 of whom were diagnosed with CAH (four males, two females). Four cases were identified as salt-wasting 21-hydroxylase deficiency (21-OHD) (2 males,2 females), one male baby had simple virilizing 21-OHD, one male baby had 11-OHD CAH. The incidence of classical 21-OHD in the screened population was 1:7,787. Detailed analyses of screening data is given in Table 1-5.

Conclusion
The incidence of CAH due to classical 21-OHD is higher in Turkey in comparison to previous reports. Thus, it is suggested to add CAH to newborn screening panel in Turkey. The use of steroid profiling as a second-tier test improves the efficacy of the screening and reduces false-positives.