# THE EVALUATION OF ACCURACY AND EFFECTIVENESS OF NEWBORN SCREENING FOR CONGENITAL ADRENAL HYPERPLASIA IN LITHUANIA

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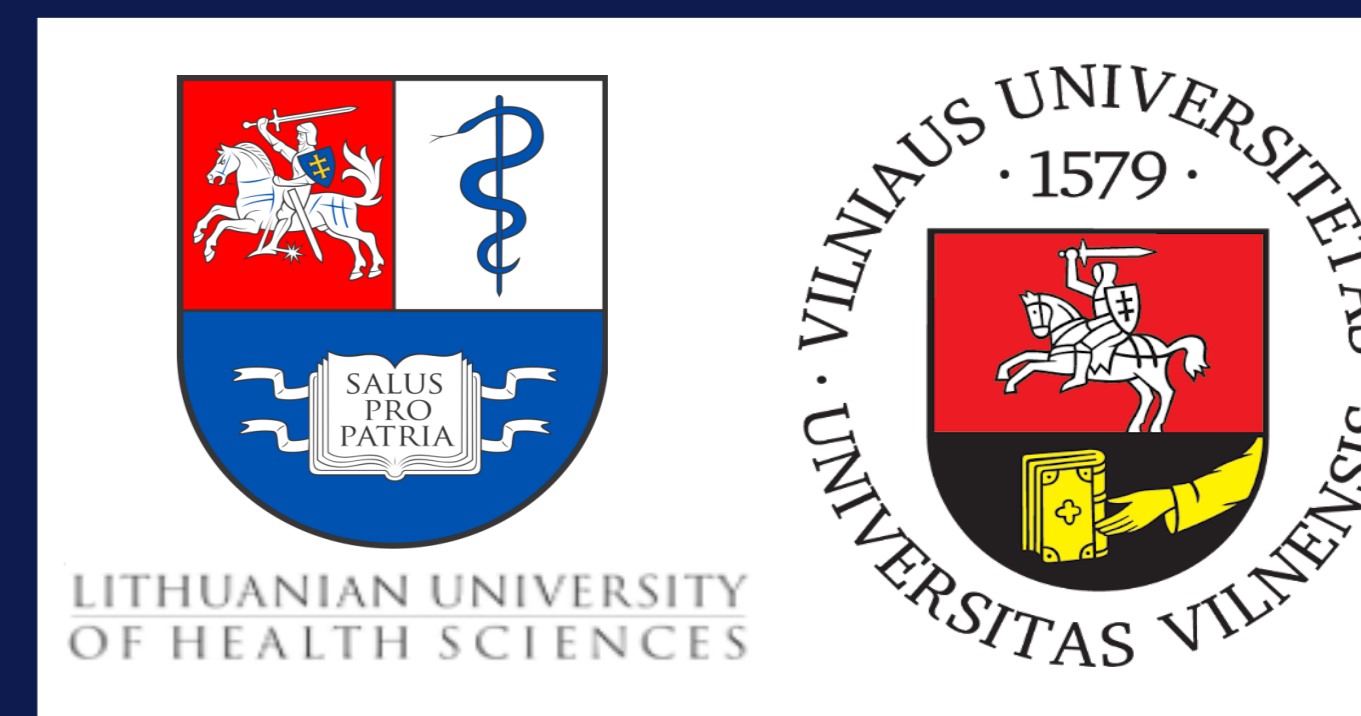
INTRODUCTION

The main goal of Newborn screening (NBS) for congenital adrenal hyperplasia (CAH) is to prevent adrenal insufficiency that can lead to life-threatening conditions. However, screening programmes are not always sensitive and effective enough to detect the disease.

AIM

We aimed to evaluate the specificity, sensitivity and efficiency of the national NBS program for CAH in Lithuania.

METHODS

Retrospective study was performed on the data of 88 patients with CAH born from 1989 to 2020:

- Patients with confirmed CAH were divided into two groups: 1) 75 patients diagnosed before NBS: 52 cases with salt-wasting (SW), 21 with simple virilising (SV) and 2 with non-classical (NC) form; 2) 13 patients diagnosed with NBS: 12 cases with SW and 1 case with SV form.

- Data on gestational age, birth weight, weight, symptoms, and laboratory tests (serum potassium and sodium levels) on the day of diagnosis were analyzed.

- For the evaluation of NBS effectiveness, data of male infants with SW CAH were analysed separately (25 unscreened, and 9 screened).

RESULTS

15,8482 neonates were screened for CAH from 2015 to 2020 in Lithuania.

After the establishment of NBS since 2015, CAH was confirmed in 13 patients (12 SW, 1 – SV form), no false negative cases were found.
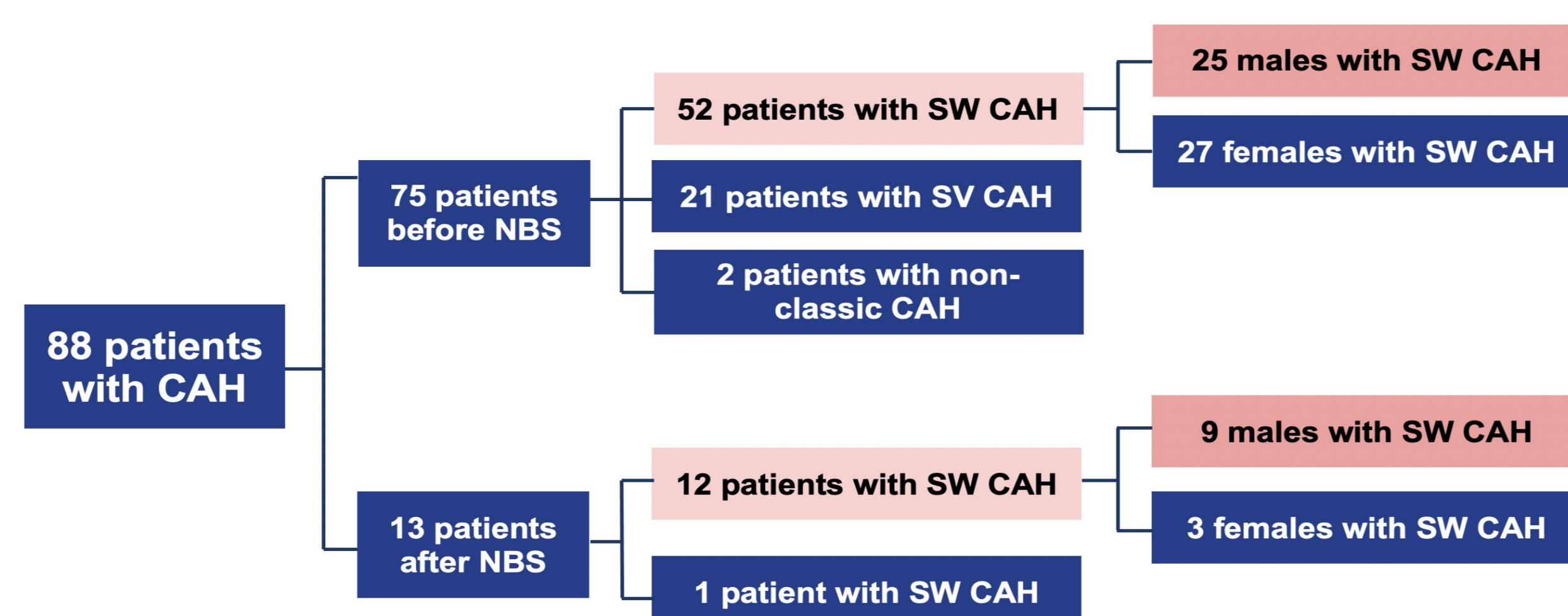

Figure 1. Descriptive scheme of cases.

- The predictive value of a positive test was 11%. The sensitivity was 100% as no false-negative cases were found and the specificity was >99.9%. (Table 1) Calculated incidence of CAH was 1:12190.

Table 1. Results of CAH screening from 2015 to 2020.

	Numbers of newborns
Screened newborns (2015-2020)	158,486
Positive tests	118
False positive tests	105
True positive tests	13
False negative tests	0

- Significant differences were found in weight at diagnosis between the groups (-1.67±1.12 vs. 0.046±1.01 SDS of unscreened and screened patients, respectively, p=0.001).

- There were no significant differences between unscreened and screened male infants groups in terms of age at diagnosis (19.13±7.15 vs. 15.44±7.79 days, p=0.189), serum potassium (7.7±1.5 vs. 6.89±1.5 mmol/l, p=0.180) and serum sodium (124.5±9.7 vs. 126.31±8.99 mmol/l, p=0.64) levels.

- Clinical signs and symptoms distribution in unscreened and screened male infants groups are shown in Figure 2.
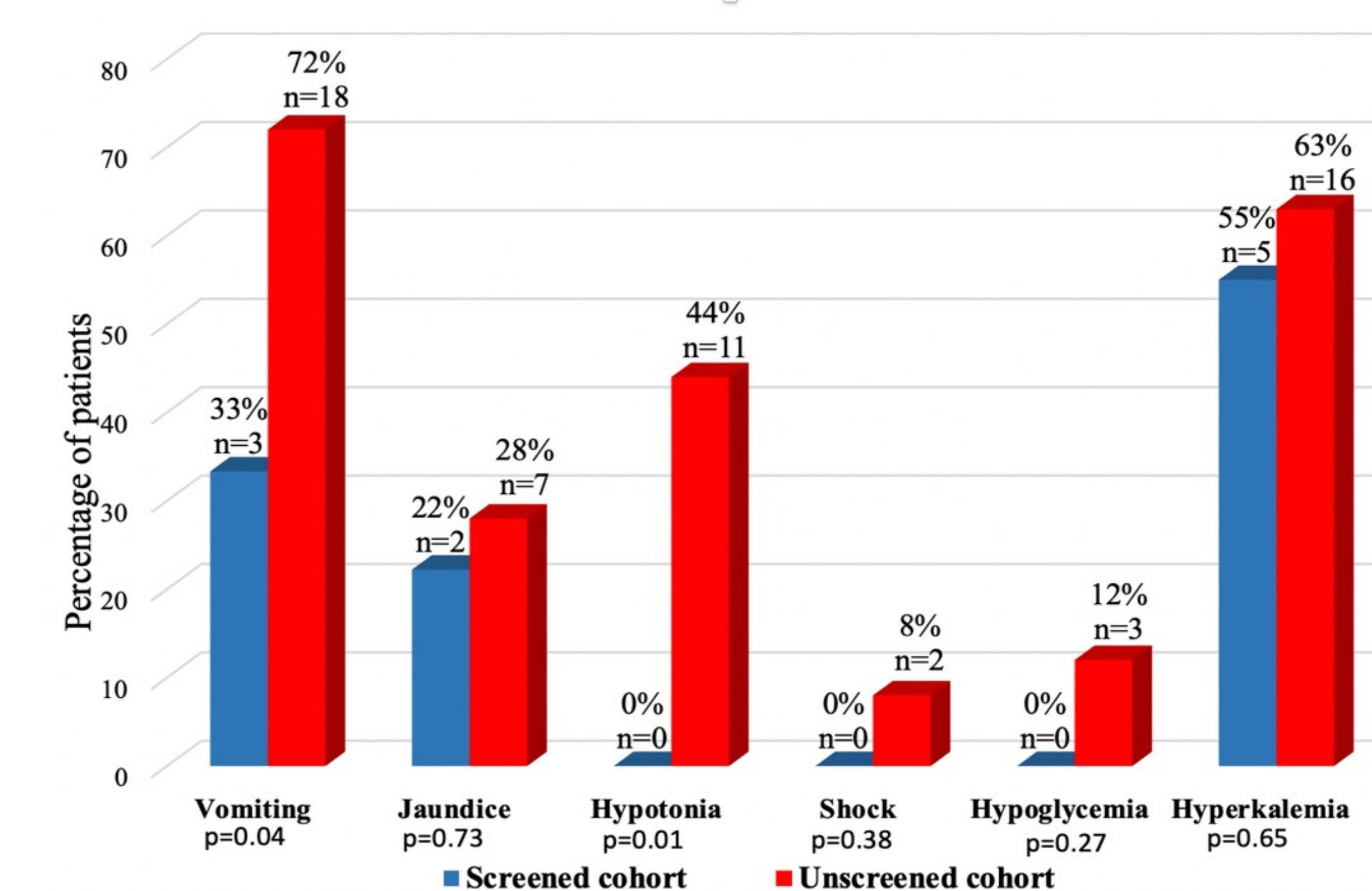

Figure 2. The clinical signs and symptoms in unscreened and screened males at the day of diagnosis.

- Eight (32%) of 25 unscreened patients and 2 (22.2%) of 9 screened patients were treated in the Neonatal intensive care unit (p=0.58).

CONCLUSIONS

- During NBS for CAH, the positive predictive value was 11%. The sensitivity was 100% as no false-negative cases were found and the specificity was >99.9%.

- Weight loss was significantly lower and the weight SDS at diagnosis was significantly higher in the group of screened patients.

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