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

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, 1 – SV form, no false-negative cases were found.

- 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).

CONCLUSIONS

- During NBS for CAH, the positive predictive value was 11%. The specificity 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.

REFERENCES


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Figures:

Figure 1. Descriptive scheme of cases.

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