INTRODUCTION

- Diagnosis of Congenital Adrenal Hyperplasia (CAH) is a challenge for the pediatrician due to the complexity of its pathophysiology and the variety of its clinical manifestations.
- Female newborns (NB) with classical forms of CAH present virilization of the external genitalia while in boys it is usually normal.
- Prematurity and neonatal stress were related to false positive screening values.
- Female: male ratio: 1.4:1.0.
- Prevalence of the classical form of CAH in the state of Parana was 1:21,596 live births, with higher incidence in the west and southwest regions of the state.
- To characterize interfering factors in the prevalence of false positive (FP) tests in CAH-NS
- To evaluate the prevalence of CAH in the State of Parana
- To determine sensitivity, specificity, accuracy, positive and negative predictive values and FP rate of the employed 17-OHP method.

AIMS

- To evaluate the prevalence of CAH in the State of Parana
- To characterize interfering factors in the prevalence of false positive (FP) tests in CAH-NS
- To determine sensitivity, specificity, accuracy, positive and negative predictive values and FP rate of the employed 17-OHP method.

RESULTS

- Prevalence of the classical form of CAH in the state of Parana was 1:21,596 live births, with higher incidence in the west and southwest regions of the state.
- Female: male ratio: 1:1.1:0.
- Prematurity and neonatal stress were related to false positive screening values.
- 17-OHP method in blood spot sample sensitivity: 100%; specificity and accuracy: 99.9%; predictive positive value: 5.1%; predictive negative value: 100%; recall rate: 0.99% and false positive rate: 0.08%.

PATIENTS AND METHODS

- Study carried out in the Pediatric Endocrinology Unit (UEP) of the Clinics Hospital of the Federal University of Parana.
- Period: August 2013 through July 2016.
- Inclusion criteria: NB with 17-OHP suspect values in NBS
- Exclusion criteria:
  - NB not evaluated in the Pediatric Endocrinology Unit (UEP)
  - NB with inappropriate sample collection

<table>
<thead>
<tr>
<th>Weight (g)</th>
<th>17-OHP ≤ 99P NORMAL</th>
<th>17-OHP &gt; 99P to 2X 99P SUSPECT</th>
<th>17-OHP ≥ 2X 99P HIGHLY SUSPECT</th>
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<td>&lt; 1500</td>
<td>110</td>
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<td>43</td>
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<td>2001 – 2500</td>
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<tr>
<td>&gt;2500</td>
<td>15</td>
<td>15,1 a 30</td>
<td>30,1</td>
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Figure 1. Diagram of the study population

- Live births: n = 474,890
- Screened newborns: ~ 100%
- 17 OHP blood spot values: n = 475
- NB evaluated at UEP: n = 403
- False positive (FP) n = 392
- Sample collection problems n = 11
- Confirmed CAH n = 23
- Hospitalized NB. Phone contact, orientation to neonatologists, normal repeated 17 OHP

Figure 2. 17 OHP blood spot values according to age at collection

Figure 3. Prevalence of CAH in Brazil and Parana

Figure 4. Trend of blood spot 17 OHP values in each patient

CONCLUSIONS

- Challenges of CAH screening program: to minimize errors in the collection of the blood spots; standardization of the method of serum 17OHP determination and, to afford adequate treatment to all affected children.
- Clinical evaluation is fundamental for proper diagnose in order to avoid overtreatment.

References

- PORTAL DA SAÚDE. NASCIDOS VIVOS – PARANÁ.
- WHITE, P. C. Nat Rev Endocrinol, v. 5, n. 9, p. 490-8, Sep 2009
- FEPE, Fundação Ecumênica de Proteção ao Excepcional