First Three Years of the Congenital Adrenal Hyperplasia Neonatal Screening Program of the State of Parana, Southern Brazil

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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.
- Salt-losing form, if not treated, develops acute adrenal insufficiency and death in the first weeks of life.
- Neonatal Screening for CAH (CAH-NS) of the State of Parana: commenced in 2013.

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.

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

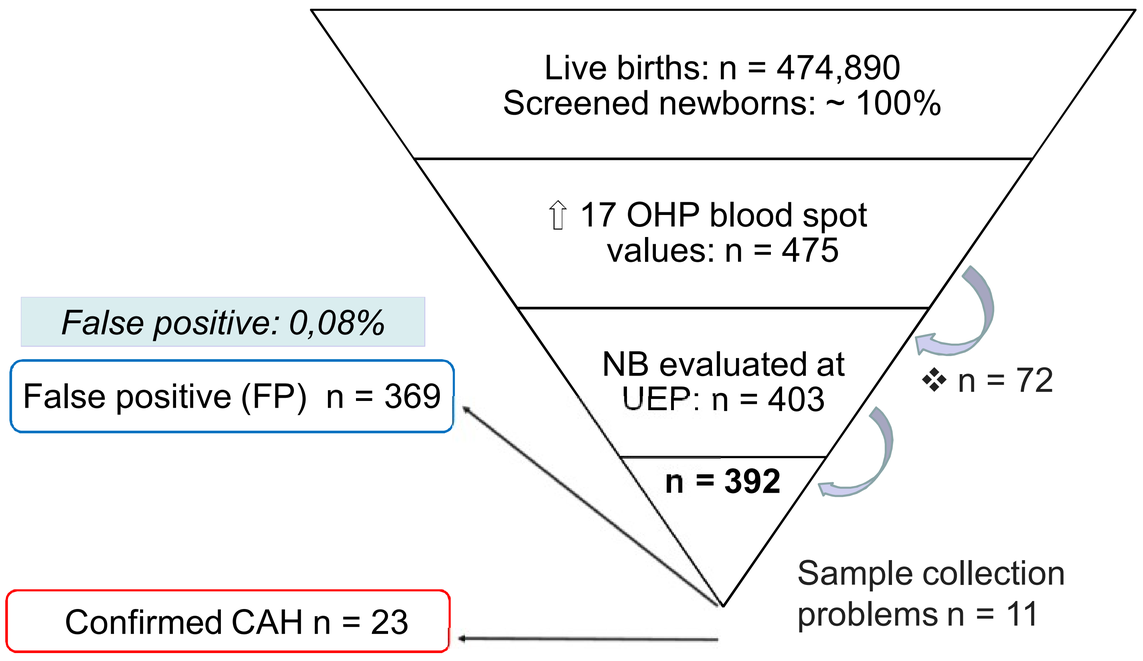
Table 1. 17 OHP cut-off values (ng/mL) for CAH-NS

Weight (g)	17 - OHP ≤ 99ºP NORMAL	17 - OHP > 99ºP to 2X 99ºP SUSPECT	17 - OHP ≥ 2X 99ºP HIGHLY SUSPECT
< 1500	110	110,1 a 220	220,1
1501 – 2000	43	43,1 a 86	86,1
2001 – 2500	28	28,1 a 56	56,1
>2500	15	15,1 a 30	30,1

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: 1.4:1.0.
- 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%.

Figure 1. Diagram of the study population



❖ Hospitalized NB. Phone contact, orientation to neonatologists, normal repeated 17 OHP

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

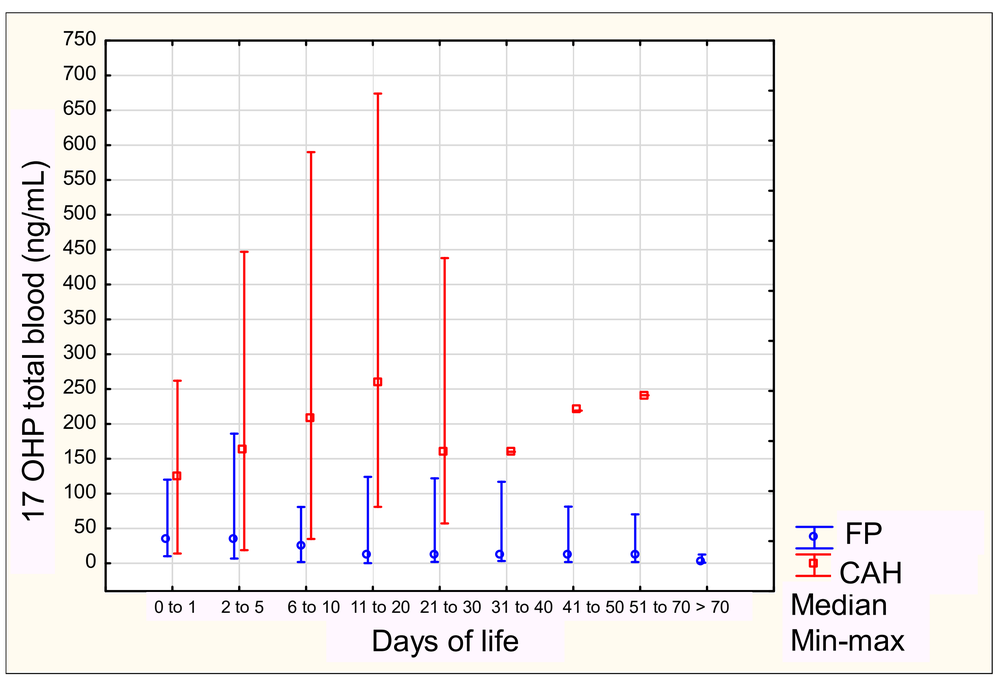


Figure 3. Prevalence of CAH in Brazil and Parana

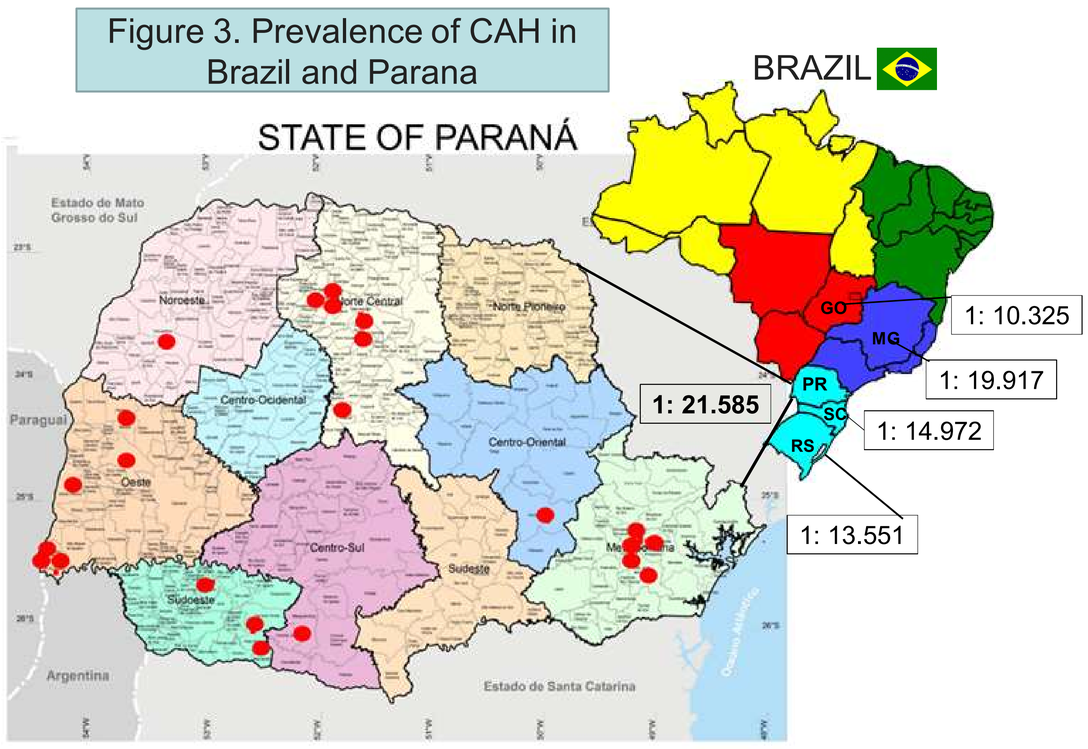
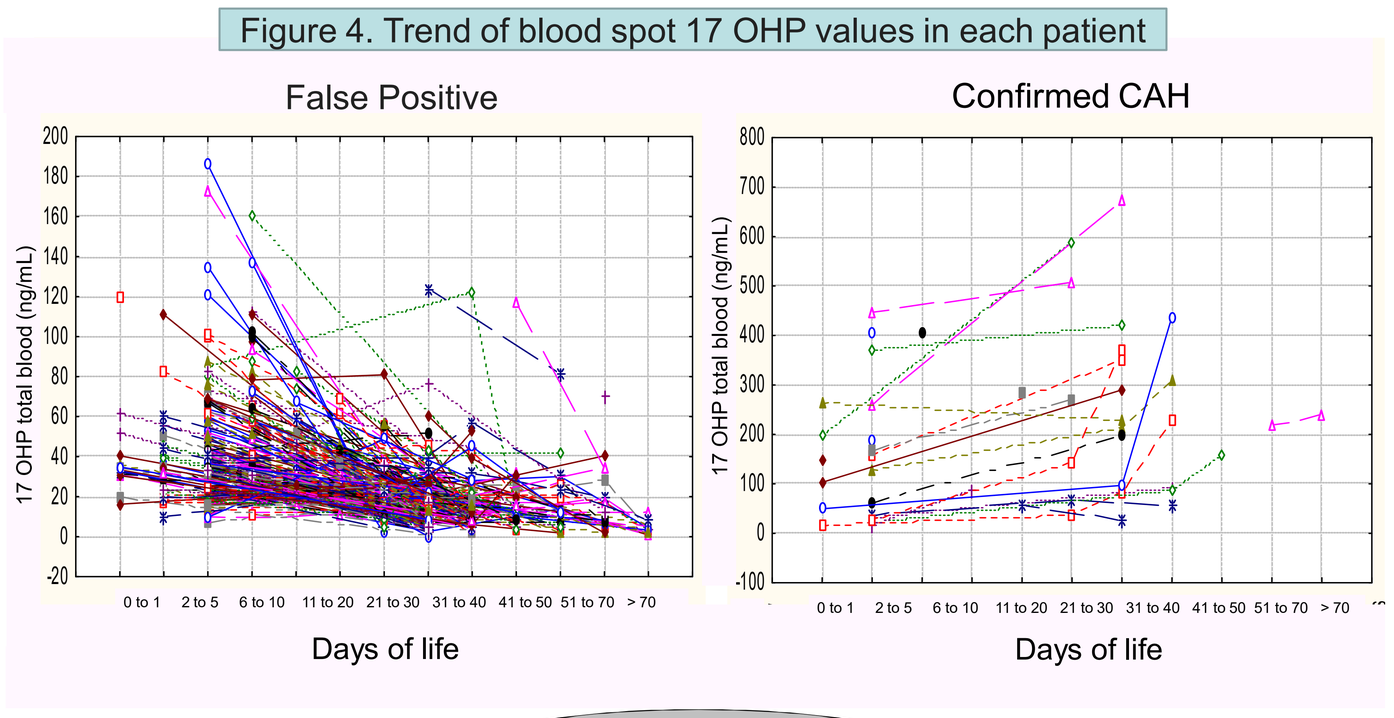


Figure 4. Trend of blood spot 17 OHP values in each patient



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.

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