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DISCLOSURE STATEMENT

The authors declare that there is no potential conflict of interest.

NEONATAL SCREENING TESTS IN PREMATURE NEWBORNS IN SOUTHERN BRASIL

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Introduction and Aims

Neonatal screening tests are used for the screening of genetic, endocrine and metabolic diseases. Preterm newborns have a higher false-positive and false-negative results in neonatal screening. The objective of this study was to estimate the prevalence of false-positive and false-negative results in the neonatal screening tests for phenylketonuria (PKU), congenital hypothyroidism (CH), biotinidase deficiency and congenital adrenal hyperplasia (CAH) in preterm newborns in Curitiba, (Southern Brazil), to analyze other factors that influence the results and to evaluate the adherence to the newborn screening guidelines for preterm newborns.

METHODS

A cross-sectional study with prospective data collection was carried out in 11 hospitals from March to December 2015. The results were compared with the results of the screening tests performed on the term newborns. For premature infants, the first collection is recommended at the time of hospital discharge, after 48h of life and a second collection after 15 days of life. For CAH screening, cut-off values based on birth weight are used.

RESULTS

- ✓ A total of 1,753 preterm newborns and 18,028 term newborns were included.
- ✓ Only 486 (28%) of the preterm newborn performed the second dried blood spot, according to guidelines.
- ✓ Prevalence of false-positive in preterm newborn was: for PKU 1:150; CH 1:133; biotinidase deficiency 1:447 and for CAH 1:5,6. Classification of screening tests for PKU and CH are shown in figure 1 and 2 respectively.
- ✓ Early dried blood spots (collected before 48 hours) showed a higher risk of false-positive results for CH and CAH (26.8 and 16.4 fold, respectively).
- ✓ Early sample collection did not influence the results of the screening tests for PKU and biotinidase deficiency.
- ✓ Figure 3 shows median 17-hidroxiprogesterone (17-OHP) values in relation to gestacional age.
- ✓ Only one patient had delayed TSH rise (figure 4).
- ✓ There were no reports of false-negative results during the study period.

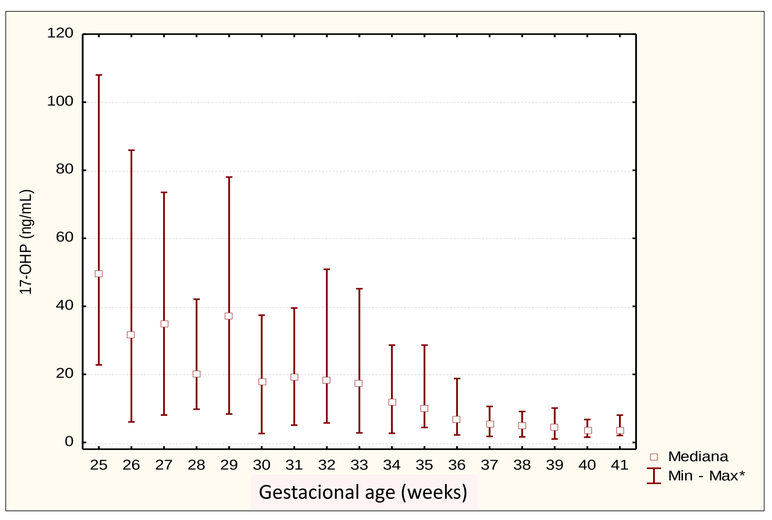

Figure 3. Median 17-OHP values for premature infantes in relation to gestacional age.

	Premature	%	Term	%	Total	%
True positive	0	0	1	0.006	1	0.005
True -negative	447	99.33	17.924	99.99	18.371	99.98
False-positive	**3**	**0.67**	**0**	**0**	**3**	**0.02**
False-negative	0	0	0	0	0	0
Total	450	100	17.925	100	18.375	100

Figure 1. Classification of screening tests for Phenylketonuria.

	Premature	%	Term	%	Total	%
True-positive	2	0.4	6	0.034	8	0.04
True-negative	435	96.7	17.898	99.85	18.333	99.77
False-positive	**13**	**2.9**	**21**	**0.12**	**34**	**0.19**
False-negative	0	0	0	0	0	0
Total	450	100	17.925	100	18.375	100

Figure 2. Classification of screening tests for Congenital Hypothyroidism

	First test	Second test	First visit
Age	48,1 h	25 days	35 days
TSH (µU/mL)	1.5	27.4	69.5
Weight (g)	1065	1390	-

Figure 4. Delayed TSH rise in one preterm newborn.

CONCLUSIONS

- ✓ Prematurity and early collection are risk factors for higher frequency of false-positive in CAH and CH neonatal screening tests.
- ✓ A second dried blood spot for screening test is recommended to diagnose cases of delayed TSH rise.
- ✓ Efforts should be made to improve the appropriate collection of tests in preterm infants to avoid false-negative results.

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