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ORIGINAL ARTICLE

Evaluation of a neonatal screening program for sickle-cell disease☆,☆☆

Rodrigo Eller[a,*], **Denise Bousfield da Silva**[b]

[a] *Medical School, Universidade Federal de Santa Catarina (UFSC), Florianópolis, SC, Brazil*
[b] *Department of Pediatrics, Universidade Federal de Santa Catarina (UFSC), Florianópolis, SC, Brazil*

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KEYWORDS
Sickle cell disease;
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Abstract
Objective: Evaluate the Neonatal Screening Program of the Health Secretariat of the State of Santa Catarina for sickle-cell disease, from January 2003 to December 2012, regarding program coverage and disease frequency.
Methods: Descriptive, observational, cross-sectional study with retrospective data collection. The variables analyzed were: number of live births in the State of Santa Catarina; number of screened children; number of children diagnosed with sickle-cell trait and sickle-cell disease; type of sickle-cell disease diagnosed; age at the time of sample collection, ethnicity/skin color, gender, and origin of children with sickle-cell disease. Descriptive measures and frequency tables were used for data analysis.
Results: During the study period, there were 848,833 live births and 730,412 samples were screened by the program, resulting in a coverage of 86.0%. There were 6173 samples positive for sickle-cell trait and 39 for sickle-cell disease. Among children with sickle-cell disease, the median age at the time of sample collection was 6 days. Regarding the ethnicity/skin color, 25 (64.1%) children were white, seven were black, and seven others were not specified. The Midwest and the Highland (Planalto Serrano) of Santa Catarina were the regions with the highest incidence of sickle-cell disease.
Conclusion: Coverage by the Neonatal Screening Program of Santa Catarina is good, but did not demonstrate an improvement trend over the years. The frequency of sickle-cell disease is low and lower than in the North, Northeast, and Midwest regions. The median age in days at the time of collection is older than the age recommended by the Ministry of Health.

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* Corresponding author.
E-mail: rodrigoellersrl@gmail.com (R. Eller).

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PALAVRAS-CHAVE
Doença falciforme;
Triagem neonatal;
Estado de Santa Catarina

Avaliação de um programa de triagem neonatal para doença falciforme

Resumo
Objetivo: avaliar o Programa de Triagem Neonatal da Secretaria de Saúde do Estado de Santa Catarina (PTN-SES/SC) para doença falciforme no período de janeiro de 2003 a dezembro de 2012, em relação à sua cobertura e à frequência da doença.
Métodos: estudo descritivo, observacional e transversal com coleta retrospectiva dos dados. As variáveis analisadas foram: número de nascidos-vivos no Estado de Santa Catarina; número de crianças triadas; número de crianças diagnosticadas com traço e doença falciforme (DF); tipo de DF diagnosticada; idade da coleta, cor/raça, sexo e procedência das crianças com DF. Foram utilizadas as medidas descritivas e as tabelas de frequência para análise dos dados.
Resultados: no período estudado, houve 848.833 nascidos-vivos e 730.412 amostras triadas pelo programa, gerando cobertura de 86,0%. Das amostras triadas, foram encontradas 6.173 crianças com traço falciforme e 39 com DF. Entre as crianças com DF, a mediana da idade em dias na data da coleta foi de 6. Das 39 crianças doentes, 25 (64,1%) eram da cor/raça branca, 7 da negra e 7 de outra cor/raça. As regiões do planalto serrano e do meio-oeste de Santa Catarina foram as regiões com maior incidência de DF.
Conclusões: a cobertura do PTN-SES/SC é boa, contudo não apresentou tendência de melhora ao longo dos anos. A frequência da DF é baixa e menor que nas regiões Norte, Nordeste e Centro-oeste. A mediana da idade em dias no momento da coleta está acima do preconizado pelo Ministério da Saúde.

Introduction

Sickle-cell disease (SCD) represents a group of autosomal-recessive inherited hematological diseases, which includes several genotypes, with a prevalence of hemoglobin S (HbS).[1–3] The presence of this abnormal hemoglobin is responsible for the main clinical manifestations of the disease, which originate from vaso-occlusive phenomena and chronic ischemic disorders.[4]

The distribution of SCD is a heterogeneous one and is more common among those of African ascendancy. According to 2009 data from the Brazilian Institute of Geography and Statistics (Instituto Brasileiro de Geografia e Estatística – IBGE), in Brazil, black and brown individuals represented 6.9% and 44.2% of the population, respectively, reflecting the heterogeneity of SCD in the country.[5]

In June 2001, through Ministry of Health Decree No. 822, several diseases were added to the list of those screened through the existing National Neonatal Screening Program (NSP) (phenylketonuria and congenital hypothyroidism), to include the detection of SCD and other hemoglobinopathies, as well as cystic fibrosis.[6] The diagnosis of SCD is performed by a laboratory through the detection of HbS and its association with quantitative fractions of other hemoglobins.

The NSP, together with multidisciplinary care, has allowed a significantly reduction in morbidity and mortality from the disease, as shown by studies in other countries.[6]

The aim of this study was to evaluate the NSP of the Health Secretariat of the State of Santa Catarina (Programa de Triagem Neonatal da Secretaria da Saúde do Estado de Santa Catarina [NSP-SES/SC]) for SCD and other hemoglobinopathies in relation to their coverage and the incidence of SCD and sickle-cell trait from January 2003 to December 2012.

Methods

This was a descriptive, observational, cross-sectional study approved by the Ethics Committee of Hospital Infantil Joana de Gusmão under opinion No. 029/2013.

The assessed variables were obtained retrospectively from a database at the Central Laboratory (Laboratório Central [LACEN]) of the Health Secretariat of the State of Santa Catarina (Secretaria da Saúde do Estado de Santa Catarina [SES/SC]) and the Live Birth Information System (Sistema de Informações sobre Nascidos Vivos [SINASC]). The collected data comprise the period from January 2003 to December 2012.

The variables included: number of live births in the State of Santa Catarina; number of children screened by the NSP-SES/SC for SCD and other hemoglobinopathies; number of children diagnosed with SCD and sickle-cell trait by the NSP-SES/SC; type of SCD diagnosed by the aforementioned program; final diagnosis of children whose first samples were inconclusive; age at collection, ethnicity/skin color, gender, and origin of children with SCD according to the macro-regions of Santa Catarina.[7]

Regarding the variable ethnicity/skin color, it used the criterion established by the Brazilian Institute of Geography and Statistics (Instituto Brasileiro de Geografia e Estatística [IBGE]).[5] The information on this variable was obtained from the file completed by the nursing staff at the time of blood sample collection.

Blood sample collection was carried out by the nursing staff in hospitals, maternity hospitals, or basic health units