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ORIGINAL ARTICLE

Neonatal screening program for hemoglobinopathies in the city of São Carlos, state of São Paulo, Brazil: analysis of a series of cases

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KEYWORDS
Hemoglobinopathies;
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Abstract
Objective: To analyze the neonatal screening program for hemoglobinopathies in São Carlos, Southeast Brazil, by investigating a series of cases in which the screening test was abnormal. More specifically, it was aimed to know the information regarding the neonatal screening received by mothers at the hospital and at primary health care, in addition to information related to genetic counseling.
Methods: A descriptive study that enrolled 119 mothers, accounting for 73% of all children born between 2010 and 2011 with abnormal results of neonatal screening for hemoglobinopathies. The mothers completed a questionnaire that assessed the information received at hospital and primary health care, and issues related to genetic counseling. Descriptive statistics was performed.
Results: Of the 119 participating mothers, 69 (58%) had children with sickle cell trait, 22 (18.5%) with hemoglobin *C trait*, 18 (15.1%) with alpha thalassemia trait and, in 10 cases (8.4%), the result was inconclusive. At the hospital, 118 mothers (99.2%) received information about where to go to collect the test and 115 (96.6%) were informed about the correct time to collect the test. Only 4 mothers (3.4%) were informed about which diseases are investigated and the risks of not performing the screening. Seventeen mothers (14.3%) recognized the difference between trait and disease, and 42 (35.3%) considered that a positive screening test could have implications for future pregnancies. In 70 cases (58.8%), the child's physician was not informed about the screening test results.

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Conclusions: The neonatal screening program needs further improvement. In both scenarios investigated, health professionals demonstrated a lack of training in providing information to mothers and families.

PALAVRAS-CHAVE
Hemoglobinopatias;
Triagem neonatal;
Assistência à saúde

Triagem neonatal de hemoglobinopatias no município de São Carlos, São Paulo, Brasil: análise de uma série de casos

Resumo

Objetivo: Realizar uma análise do programa de triagem neonatal de hemoglobinopatias no município de São Carlos, São Paulo, Brasil, por meio da investigação de série de casos cujo resultado do teste de rastreio foi alterado. Objetivou-se conhecer as informações a respeito da triagem neonatal recebidas pelas mães na maternidade e na atenção primária à saúde, além das informações relacionadas à orientação genética.
Métodos: Estudo descritivo, no qual participaram 119 mães cujos filhos apresentaram teste de triagem de hemoglobinopatia alterado, correspondendo a 73% das crianças nascidas entre 2010 e 2011 com resultado de triagem neonatal para hemoglobinopatia anormal. As mães responderam a um questionário que avaliou informações recebidas na maternidade e na atenção primária à saúde, além aspectos relacionados à orientação genética. Foi realizada estatística descritiva dos dados.
Resultados: Das 119 mães participantes, 69 (58%) possuíam filhos com traço falciforme, 22 (18,5%) traço C, 18 (15,1%) traço alfa talassêmico e, em 10 (8,4%) casos, o resultado foi inconclusivo. Na maternidade, 118 mães (99,2%) receberam informação sobre onde ir e 115 (96,6%) foram orientadas sobre o momento correto para coleta do teste. Somente 4 mães (3,4%) foram informadas sobre quais doenças seriam investigadas e os riscos de não realizar o rastreio. Das 119 mães participantes, 17 (14,3%) reconheceram a diferença entre traço e doença e 42 (35,3%) consideraram que um teste alterado poderia ter implicações para futuras gestações. Em 70 casos (58,8%), o médico da criança não foi informado sobre o resultado da triagem.
Conclusões: O programa de triagem neonatal necessita de aperfeiçoamento. Nos dois cenários investigados, os profissionais de saúde carecem de treinamento para orientar mães e famílias.

Introduction

Neonatal screening allows the early identification of several congenital diseases that have no symptoms at birth, in order to intervene in its natural course, thus attenuating the clinical consequences. The criteria used for the inclusion of a disease in a neonatal screening program in general follow those proposed by James Wilson and Gunnar Jungner in 1968: the condition being screened must be an important health problem; the natural history of the disease needs to be well known; there must be an identifiable early stage; early treatment should bring greater benefits than at later stages; an adequate test should be developed for the early stage; the test should be acceptable to the population; retesting intervals should be determined; health care services need to be adequate for the extra clinical work resulting from screening; risks, both physical and psychological, should be fewer than the benefits.[1]

In Brazil, in 2001, the Ministry of Health established the National Neonatal Screening Program (PNTN) with the objective of expanding the existing screening program at the time (restricted to phenylketonuria and congenital hypothyroidism), including the identification of other congenital diseases, such as hemoglobinopathies and cystic fibrosis.[2] Moreover, the PNTN established a comprehensive approach of the subject, involving early detection, increased population coverage, active search for patients, diagnostic confirmation, follow-up and treatment, and the creation of an information system to register patients.[3]

PNTN was designed as a five-step system, usually organized and conducted by the public health system, which has the necessary conditions and authority to carry out universal screening, in which the pediatrician plays an important role.[4] The first step comprises the screening test itself and it aims at the universal coverage of screening, i.e., that all newborns be tested. Obstetricians and pediatricians are essential at this stage. Parents need to know about the existence of neonatal screening and be counseled beforehand about which diseases will be screened and benefits of early detection, the risks for the newborn who is not submitted to the test, the appropriate age for its performance, the need for subsequent confirmatory