

SHORT COMMUNICATION

NEONATAL SCREENING FOR AMINO ACIDOPATHIES AND CONGENITAL HYPOTHYROIDISM: A PILOT PROGRAM IN SOUTHERN BRAZIL

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ABSTRACT

A pilot study on neonatal screening for aminoacidopathies and congenital hypothyroidism was carried out at the Hospital de Clínicas de Porto Alegre. Although preliminary, the program has detected a case of classical phenylketonuria, a patient with congenital hypothyroidism and two newborns with neonatal tyrosinaemia. The utility of such programs in developing countries is discussed.

INTRODUCTION

Neonatal screening for various inherited metabolic disorders has been routinely performed during the last 25 years in most western industrialized countries (Wilcken and Webster, 1991). Whereas practically all newborns of these countries are examined for phenylketonuria and congenital hypothyroidism (CH) other disturbances such as galactosaemia, homocystinuria, cystic fibrosis, biotinidase deficiency, maple syrup urine disease are currently screened in only a few programs (Bickel, 1987; American Academy of Pediatrics, 1989).

In 1990 a law was passed in Brazil making neonatal screening for "metabolic disorders" mandatory in all private and public maternities. In 1992, a pilot study on neonatal screening for amino acidopathies and CH was set up in Porto Alegre, capital of Rio Grande do Sul, the southern most state of Brazil.

MATERIAL AND METHODS

The study, sponsored by the National Health Service, was designed to test all children born at the Clinical Hospital of Porto Alegre (about 300 each month). Blood samples, collected at the hospital outpatient clinic in filter paper from the 5th till the 90th day of life, were tested for increased TSH, using the Delfia hTSH kit-anti-hTSH microtitration strips, and for elevations of amino acids, using one-dimensional thin layer chromatography on cellulose Merck precoated chromatoplates.

RESULTS AND DISCUSSION

So far we have tested 4,330 individuals and the results are as follows. In the first year of the program, 70% of the target population was screened. Among them, 64% had their blood taken between the 5th and the 14th day of life, as originally planned. The rest were screened at an older age.

Twenty cases with TSH above the established cut off level (30 mIU/L) were retested by serum TSH and T4 assays. All abnormal samples from the first collection had TSH values between 30 and 50 mIU/L, except for one whose value was 470 mIU/L. This case was later confirmed

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as CH. However, treatment began only at the age of two months, when symptoms were already apparent.

An abnormal result in the amino acid chromatographic pattern was verified in 12 newborns (eight elevations of tyrosine, one of phenylalanine, two of glycine and one of valine) who were recalled and retested. Amino acid quantification in second samples of these children was normal, except for three infants. We detected one case of classical phenylketonuria and two cases of neonatal transitory tyrosinaemia, one labelled as moderate (25 mg/dL) and the other as marked (42 mg/dL). The phenylketonuric patient was put on a low phenylalanine diet (Lofenalac) at 14 days of age and the individuals with neonatal tyrosinaemia on a low protein diet supplemented by ascorbic acid. The prevalence of neonatal tyrosinaemia (1:2,165) was similar to that found previously in a private voluntary neonatal screening program performed in our region (Camargo Neto *et al.*, 1991).

Our study indicates that such programs can be of benefit for target populations of developing countries. Nevertheless, scarce available resources may be better applied to other programs such as control of infectious diseases by immunization. On the other hand, amino acid thin layer chromatography seems to be preferable to other methods which identify single amino acids (such as Guthrie tests for phenylalanine, methionine and leucine) because the cost of a single Guthrie test is comparable to chromatography (Brauer *et al.*, 1991) and the latter screens for most amino acidopathies (Giugliani *et al.*, 1989).

Government sponsored neonatal mass screening programs for inherited metabolic disorders in developing countries like Brazil, with a huge geographical area and under economic crisis, are very difficult to conduct. We think that pilot studies like ours (similar programs were since set up in two other government sponsored hospitals of Porto Alegre) are important for learning how to set up screening programs in developing countries and in training human resources as well, in addition to providing valuable data on the prevalence of common inborn errors of metabolism.

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RESUMO

Um estudo piloto sobre triagem neonatal para aminoacidopatias e hipotireoidismo congênito foi realizado no Hospital de Clínicas de Porto Alegre. Embora preliminar, o programa detectou até agora um caso de fenilcetonúria clássica, um paciente com hipotireoidismo congênito e dois recém-nascidos com tirosinemia neonatal. A utilidade de tais programas em países em desenvolvimento é discutida.

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