

## PROBLEMS IN IMPLEMENTING NEWBORN SCREENING IN COSTA RICA

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In Costa Rica, concern for public education early in this century, together with specific public health actions in the early 1970's, have resulted in fairly good control of infectious and nutritional diseases. As a consequence, a disease profile of a developed country has emerged. Prematurity and other birth defects are the current leading causes of infant morbidity and mortality (1). Resources for the detection, diagnosis and treatment of these problems, including inborn errors of metabolism, are insufficient; in fact, the general and economical development of the country is well behind the partial improvements in public health.

To meet the formidable challenge that this "new pathology" represents, the installment of a nationwide neonatal screening program for inborn errors of metabolism seems imperative, not only as an efficient strategy to counteract the immediate impact of these diseases in terms of mental retardation and/or other disabilities, but also to serve as a basis for a broader program for the prevention of other genetic or partially genetic diseases.

The background for the initiation of the program (2), as well as the results of the pilot study (3), have been presented elsewhere. In summary, only PKU and MSUD have been included so far, using the Guthrie test. Congenital hypothyroidism and galactosemia have been considered, but not included yet because of lack of budget and relatively complicated screening tests. MSUD was included, because of an apparent high frequency of this disease in Costa Rica (3).

Specific problems detected through the pilot study include: early discharge that forced a low cutoff point for Phe, resulting in an intolerably high percentage of initial positives; impossibility to locate half of these initial positives, mainly because of false addresses given by mothers in order to be attended in one preferred hospital; the alleged saturation with work in the nurseries is an "excuse" for high percentage of inadequate samples, incompletely filled forms, and impossibility to expand the program (3). On the other side, promising results have been obtained through the pilot study when mothers were cited at 7 days postpartum, with a high rate of response, and the elimination of most of the problems found in the nursery.

The yield of this pilot program in terms of true positives is shown in Table 1.

In addition to the aforementioned specific problems, other general ones typical of underdeveloped countries are negatively affecting the implementation of screening. These major problems include: bureaucratic inefficiency, e.g. intolerable

TABLE 1  
NEONATAL SCREENING IN COSTA RICA  
Pilot Program: August 1983 - August 1986

Total screened:	29,853
Total true positives:	
Benign hyperphenylalaninemia:	1
MSUD*	3

\* Detected early on clinical grounds; screening program decisive in diagnosis and follow-up.

delays in clearing customs; economic situation, with a high foreign debt and scarcity of foreign currency for imports, all in competition with peremptory needs in - usually unplanned - medical care; political changes with the government in turn, actual or expected, that "freeze" the program in the meantime. Specific, short-term actions from the government, as well as long-term, more difficult to attain cultural changes mainly at the level of health professionals and bureaucrats, together with a strong support from developed nations, all are needed to overcome these major problems, as well as the derivative, specific ones.

The organization of the program is summarized in Figure 1.

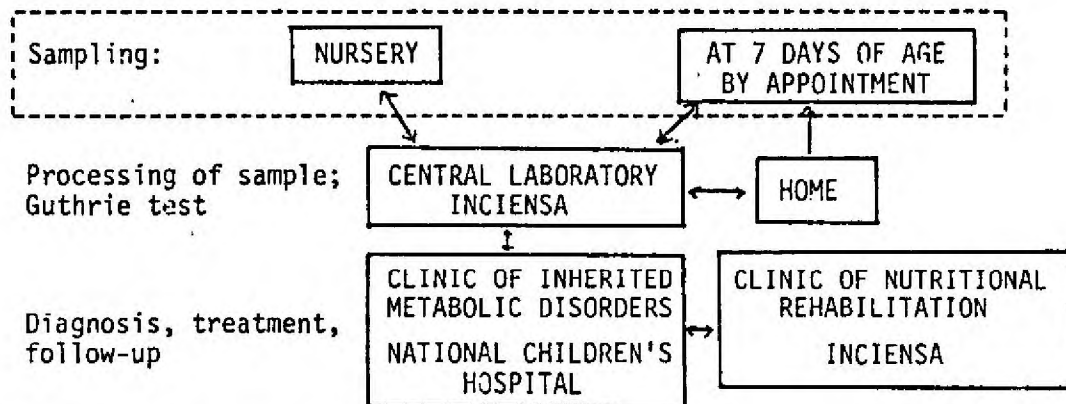


Figure 1. Current organization of newborn screening in Costa Rica. Dotted lined box shows the rate limiting step of the program, and two possibilities for sampling time. (INCIENSA = Costa Rican Institute for Research and Training in Nutrition and Health.)

#### REFERENCES

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