



# Follow-up of Hemoglobinopathies Diagnosed Through Newborn Screening

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## Background

In 1987, the National Institutes of Health produced a consensus statement recommending sickle cell screening for all newborns based on the overwhelming benefits of early identification and treatment.<sup>1</sup> The state of Washington has followed these guidelines and currently screens all newborns for sickle cell anemia. However, the testing methods used also detect numerous other hemoglobinopathies and traits. Current practice in the Office of Newborn Screening is to report all detected hemoglobin abnormalities to the physician and they decide whether or not to pass the results on to the family of each affected newborn.

## Practicum Project

This practicum was done at the Office of Newborn Screening through the Washington State Department of Health. The goal was to gather data from follow-up surveys sent to physicians regarding their patients who were diagnosed as having either a hemoglobin trait (167 surveys) or hemoglobin Bart's (181 surveys). Data extracted from the forms was used to determine how well the information transfer from physician to patient was taking place and what follow-up services were being utilized.

1. NIH Consensus Statement. JAMA 1987; 258(9): 1205-9.

## Description of Survey

When newborn screening test results are sent to the physician, included are educational brochures and pamphlets and a survey form to be returned to the lab. Besides basic demographic information, the survey also includes checkboxes to answer the questions below and additional space for comments.

## Physician Survey Results

### Hemoglobin Traits (167 surveys returned)

Shared Results w/ Parent(s)	144	86%
Determine Hemoglobin Status of Parent(s)	62	37%
Refer for Genetic Counseling	28	17%

### Hemoglobin Bart's (181 surveys returned)

Shared Results w/ Parent(s)	122	67%
Determine Hemoglobin Status of Parent(s)	7	4%
Refer for Genetic Counseling	24	13%



*Saving lives with  
a simple blood spot*

## Conclusions

Because of the low response rate and the inconsistency with which the survey forms were filled out it is difficult to form any solid conclusions regarding the follow-up for newborns identified as having a hemoglobin abnormality. These results suggest, however, that although parents are being informed about their newborn's test results, they are not receiving follow-up testing for themselves or formal genetic counseling to explain the results and future reproductive risks.

A more thorough strategy for determining physicians actions taken after hemoglobinopathy results are received is needed. Based on these results, it may also be worthwhile to expend some resources to increase the numbers of families who receive follow-up testing and counseling.