

Sickle Selections

a quarterly newsletter from the University of Rochester Sickle Cell Program

April 1998

Birth Statistics for Newborns in HSA Region II of NY State¹

The New York State Newborn Screening Program requires a blood specimen from every baby born in the state. Among the 7 disorders tested for by the state on these specimens are hemoglobinopathies. A list of the hemoglobinopathies identified in newborns in HSA region II over the past 3 years is listed below.

	1997	1996	1995
Disease			
S/C	4	2	6
S no A ²	3		4
S/S	2	1	
S/ β -Thal	1		
E/E	1		1
C no A ²		1	
Total	11	4	11
Trait			
A/S	233	192	215
A/Fast	60	56	56
A/C	57	57	61
A/E or O	19	9	19
A/D or G	5	13	4
A/Fetal	4	7	20
A/O Arab	3	1	1
A/Variant		1	
Total	381	336	376

¹ Chemung, Livingston, Monroe, Ontario, Schuyler, Seneca, Stueben, Wayne, Yates counties

² An abnormal hemoglobin and no hemoglobin A can indicate either a homozygote for the abnormal hemoglobin or a compound heterozygote for the abnormal hemoglobin and β^0 -thalassemia. The best way to distinguish these is to test both parents.

A Fast-Moving Hemoglobin at Birth

A newborn screening report of "A/Fast" means that hemoglobin A and a band that migrates faster than hemoglobin A was observed on cellulose acetate electrophoresis. The fast-moving hemoglobin can represent:

- hemoglobin Barts (γ_4), indicative of α -thalassemia
- a hemoglobin variant such as J or N or
- an artifact of the filter paper blood specimen

In such cases we recommend:

if the newborn is Asian or African-American, the fast-moving hemoglobin is probably Barts, indicating α -thalassemia. The most common cause is the deletion of one or two alpha genes. Usually the condition is asymptomatic and the fast-moving hemoglobin will be gone on retesting at a year of age. If it persists into adulthood the child may have hemoglobin H disease (a 3-gene deletion type α -thalassemia) producing moderately severe anemia.

if the newborn is Caucasian, repeat the hemoglobin electrophoresis at one year of age. If it is gone it was a fetal hemoglobin mutation. If it is still present it is an adult hemoglobin mutation. Only if anemia is present at that time is further investigation needed, viz. checking for an unstable hemoglobin.

Note:

Blood samples may be sent to our laboratory for testing for a fast-moving hemoglobin. Such blood samples should be kept at room temperature and electrophoresed promptly since a fast-moving hemoglobin may precipitate at 4^o C.

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MEDICAL CENTER
Strong Sickle Program
Offers Help

Regarding treatment contact:

Dr. Norma Lerner or Pat Lamarche R.N., P.N.P. Department of Pediatrics 275-2981

Regarding laboratory diagnosis, newborn screening and genetic counseling, contact:

Dr. Peter Rowley, Sandra LaBella or Starlene Loader, Division of Genetics 275-4602

Visit our website: www.urmc.rochester.edu/genetics