

Unidentified Hemoglobin Variant

- Newborn screening detects thousands of infants each year in the United States with unidentified hemoglobin (Hb U). There are over 600 structural hemoglobin variations and the majority detected by screening cannot be definitively identified. **99% of these unidentified variants are clinically benign.**
- Mutations in the alpha, beta, or gamma hemoglobin genes may cause these unidentified variants. Some of the gamma chain variants may disappear in early infancy, when fetal hemoglobin is no longer produced.
- Most of these infants have **Unidentified Hemoglobin Variant Traits**. They are heterozygotes with the percentage of adult hemoglobin (Hb A) equal to or greater than the percentage of unidentified hemoglobin variant (Hb U). The percentage of both Hb A and Hb U is less than the percentage of fetal hemoglobin (Hb F) on the newborn screen. Instead of the typical newborn screening result for hemoglobin (“FA”), these babies have the result “FAU”.
- Most infants with the newborn screening hemoglobin result **FAU** have no clinical or hematological consequences. A few unidentified variants may show altered oxygen affinity and the individual may have erythrocytosis. A few unidentified variants may be chemically unstable and the individual may have hemolysis or anemia. Some unidentified hemoglobin variants can cause sickle cell disease when co-inherited with hemoglobin S, but most unidentified hemoglobin variants have no significant genetic implications.
- If the infant has the newborn screening hemoglobin result **FUA**, then the percentage of unidentified hemoglobin (Hb U) is greater than the percentage of adult hemoglobin (Hb A). **A complete blood count is recommended for babies with FUA hemoglobin at 9 months of age.**
- **If you have specific concerns for a given family, please contact Dr. Brian Abbott in Great Falls at 406-530-9788 for further assistance.** Dr. Abbott, a board-certified pediatric hematologist, has been contracted to provide follow-up for Montana’s Newborn Screening Program.

Adapted with permission from the Utah Department of Health <http://health.utah.gov/newbornscreening>

Newborn Metabolic Screening, Montana Department of Public Health and Human Services
W.F. Cogswell Building, 1400 Broadway, PO Box 4369, Helena MT 59604-4369,
Phone: (406) 444-0984 or (800) 821-7284, Fax 406-444-1802 www.newborn.hhs.mt.gov