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The Case For Identity Testing In Newborn Screening

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Newborn screening provides for early identification of infants at risk for congenital disorders. Follow-up, an integral component of the system, is a labor intensive activity with the goal of ensuring that infants who screen positive are evaluated and diagnosed for the condition in question. Follow-up becomes more complicated when an infant is referred for evaluation and the clinical presentation and subsequent work-up do not support the original screening result. This may suggest that there are two different infants involved. The New York State Newborn Screening Program began to use identity testing as a tool to help resolve this concern in 1997. A retrospective review of case histories was conducted to classify the circumstances for which the identity of the newborn came into question in which identity testing was done. Identity testing results were reviewed to evaluate how this additional information contributed to resolution of these issues. Preliminary data of hemoglobin results suggest that misidentification of specimens may be a greater issue than previously thought for specimens submitted for screening. The case can be made for the use of identity testing, normally associated with use in forensics or paternity testing, in newborn screening when there is a question regarding the origin of the blood submitted for testing.