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Hematologist discovers, names the 'Toms River' blood mutation in N.J. family

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A newborn described as a "happy blue baby" because of her bluish skin color but healthy appearance made a small mark in medical history when one of her physicians discovered something new in her genes—the hemoglobin Toms River mutation.

Scientists have identified hundreds of mutations in genes that carry instructions for producing hemoglobin—the four-part protein that carries oxygen in everyone's red blood cells. By tradition, whoever discovers a mutation in hemoglobin genes names it after the hometown of the patient, said pediatric hematologist Mitchell J. Weiss, M.D., Ph.D., of The Children's Hospital of Philadelphia.

Weiss and colleagues published a brief report on the mutation in the May 12 issue of the *New England Journal of Medicine.* He collaborated with biochemist John S. Olson, Ph.D., of Rice University.

Fortunately, this particular mutation, which causes blue coloration (cyanosis) and anemia, has only mild, transient effects, and disappeared within a few months of birth, as the baby's hemoglobin made a normal transition from fetal hemoglobin to a different form of hemoglobin found in older children and adults.

Often, the researchers at Children's Hospital follow a "bench to bedside" path—working to translate scientific findings into treatments of benefit to children's health. "This was sort of a bedside-to-bench event, in which exploration of a patient's condition led us to better knowledge of

biology," said Weiss, who has investigated hemoglobin disorders for much of his career in medicine.

Weiss added, "We started with an unusual case. The baby had a blue color, but otherwise appeared healthy. We were able to rule out more common, serious causes, such as heart or lung disease. Then we learned some family history when the grandmother mentioned that the child's father also had experienced temporary cyanosis as a newborn."

Weiss had a clinical DNA diagnostics lab at Children's Hospital, directed by Catherine A. Stolle, Ph.D., perform DNA sequencing on the infant's and the father's blood. The analysis revealed that a gene carrying the code for a subunit of hemoglobin had a rare mutation, not previously identified, which the study team named after Toms River, N.J., where the patient lives.

Further biochemical analysis provided new scientific knowledge of blood disorders.The Toms River mutation that leads to a different amino acid than that found in normal fetal hemoglobin fortunately had limited, temporary effects in the infant. When a similar chemical substitution occurs in the hemoglobin subunit gene that is expressed in adults, the health effects can be more sustained and serious.

"While this finding will not broadly change the way we do medicine, it helped us care for the child and counsel this family, added Weiss. "Perhaps this report may remind clinicians who encounter a healthy blue baby to consider explanations that don't involve a serious underlying disorder. In an unusual but mild case like this, an accurate diagnosis can spare babies and families unnecessary concern and inappropriate medical testing."

Provided by Children's Hospital of Philadelphia

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