

Inherited Genes and Family Myths

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While family stories may not be genetic, they certainly can be inherited from one generation to the next. We've all heard them. Many are harmless "old wives' tales," while others can perpetuate misunderstandings. Genetic counselors deal with family myths every day and are often in a position to unravel and replace these yarns with accurate information.

John and Mary are scheduled to attend a genetic counseling session at the suggestion of Mary's obstetrician. They are expecting their first baby this summer but are concerned because John has "beta-thalassemia," or so he has been told all of his life by several family members. The counselor notices John is an active duty Marine. This is interesting because under usual circumstances someone with thalassemia is excluded from military service. The counselor asked John to bring his medical records to the appointment so she and the physician can review his blood results to clarify his diagnosis.

During the appointment, John reports he takes iron tablets, which have not been prescribed by a doctor. John believes, "The extra iron will help keep me from having anemia, like my grandfather who had thalassemia." Mary remarks, "Maybe if John's grandfather had taken iron tablets, he wouldn't have needed so many blood transfusions for his thalassemia!" John says he has never had symptoms of thalassemia nor ever needed a blood transfusion, unlike his grandfather.

Beta-thalassemia is a genetic disorder of the red blood cells which affects the size and oxygen carrying ability of these cells. Beta-thalassemia is inherited as an autosomal recessive disorder, meaning both parents have to carry an altered gene in order to have a child affected with the disorder. An individual who carries beta-thalassemia usually has few or no clinical problems. Symptoms in affected individuals can include severe anemia in infancy resulting in a shortened life span, if left untreated. Moreover, iron supplementation can actually cause an iron overload in individuals who are affected with or are carriers for thalassemia.

As it turns out, John's medical records show his blood cell tests are within the normal range. These findings suggest John is not affected with beta-thalassemia and probably isn't a carrier. When discussing this with the counselor, John says, "But my grandfather had it, and my mom got it from him, and my mom always said I have it too." When asked if he ever mentioned this during his military screening, John states, "No. I didn't think I should or else they might not let me be a Marine!"

So then, how did this couple end up in a genetic counselor's office? When Mary discovered the pregnancy, she discussed her concerns with her doctor because of all the family stories about John's thalassemia. John, his sister, mother and maternal grandfather were all believed to have the disorder. While meeting with the counselor, the couple received written materials about thalassemia and began reading about the disorder. They discuss their concern with the counselor that they both may carry an altered hemoglobin gene. Tests were offered to determine their risk. The genetic tests show neither of them carries an altered hemoglobin gene. Based on this information, their children will not be at increased risk for inheriting thalassemia.

John and Mary are very relieved. John thanks the counselor and remarks, "You know, in all my life no one has ever explained any of this to me in a way I could understand. This has been so helpful...and to think, I almost didn't come along with Mary today because I figured I wouldn't learn anything."

Today, many people attempt to find information on their own before, or in some cases instead of, ever seeing their doctor. Anyone can go on the internet and look for information about self-treatment, but the medical information on the internet can be confusing, incomplete or sometimes just wrong. Genetic counselors can fully research and explain complicated medical issues in terms that are easy to understand. For many patients, these appointments can provide much needed information and give participants the opportunity to ask many of the questions they've been afraid to ask. Counselors know how commonplace it is for family myths and "old wives' tales" to be interpreted as factual. Counselors are able to clarify these stories by replacing fiction with helpful information.

Hemoglobin disorders can be very puzzling, even for healthcare providers. Individuals who are concerned about their risk of carrying beta-thalassemia or other genetic disorders are encouraged to discuss testing with their physicians and genetic counselors. GeneCare Medical Genetics Center, in Chapel Hill, offers genetic evaluation, counseling, and laboratory services. For more information: (919) 942-0021 or 1-800-277-4363 or visit www.genecare.com.