

Giving Birth to Someone Else's Children? A Case of Disputed Maternity

by

Jessica Hutchison
Biological Sciences
Cameron University, Lawton, OK

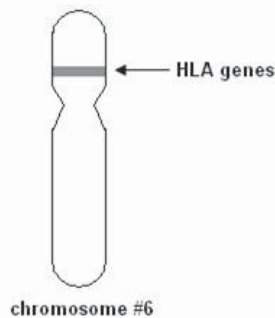
Part I—Haplotypes

"You know, Karen, something very unusual has happened here. We've tested your sons because they were possible donors. Your sons' blood does not match your blood and that's an impossibility, so they couldn't be your children...these could not be your children."

Karen's hand shook as she listened to the nurse on the telephone tell her that two of her three sons could not possibly be hers. Karen, who was 52 years old, was in need of a kidney transplant. She and her sons had undergone blood tests to determine if they were suitable donors.

The tests determined their human leukocyte antigen (HLA) genes, some of which encode cell surface recognition proteins that the body uses to distinguish its own cells from foreign material. Because there are hundreds of different versions of the 200+ HLA genes, each person's combination is almost unique. However, because these genes are located in a group on chromosome #6, they are often inherited together in a block known as a haplotype (see Figure 1).

Figure 1. Location of HLA genes.



Because a transplanted organ is seen as foreign tissue to a host's immune system, it is important to try to "match up" as many of these genes as possible to minimize the rejection of the new organ. The chance of matching is highest among related individuals.

The nurse told Karen that her lab report results indicated that her HLA haplotype is type 1 and 3 and her three sons were types 2 and 5, 2 and 5, and 1 and 6, respectively.

Questions

1. How many HLA haplotypes should this woman's sons each share with her?
2. According to these data, which sons cannot be hers?
3. What are some hypotheses to explain these data? Write down as many explanations as you can, no matter how far-fetched.
4. What tests should be done next to evaluate your hypotheses?

Part II—Pedigree

Karen sought help from Dr. Margot Kruskall, a doctor at the Beth Israel Deaconess Medical Center in Boston, Massachusetts. Karen assured Dr. Kruskall that these were her children, conceived naturally, without the help of any reproductive technologies. Dr. Kruskall decided to rerun all of the HLA tests on Karen and her sons' blood. The tests yielded similar results. This time Karen's husband was also tested. The tests indicated that he had an HLA haplotype of 5 and 6. Dr. Kruskall also verified that Karen had a normal chromosome complement, 46 XX.

Questions

1. Make a pedigree of Karen's family, displaying their HLA haplotype combinations.
2. Draw a Punnett square using Karen and her husband's haplotypes showing the possible haplotype combinations of their offspring. Determine the probability of each of their sons' haplotype combinations and record it on your pedigree.
3. Which of your hypotheses from Part I are no longer likely, given this information, and why? Which of your hypotheses are still likely?

Part III—Family Members

Dr. Kruskall and her colleagues were stumped. They decided to test more of Karen's family members. Her two brothers had haplotypes of 1 and 3 and 2 and 3, while her mother had haplotypes of 3 and 4. Karen's father is deceased and could not be tested.

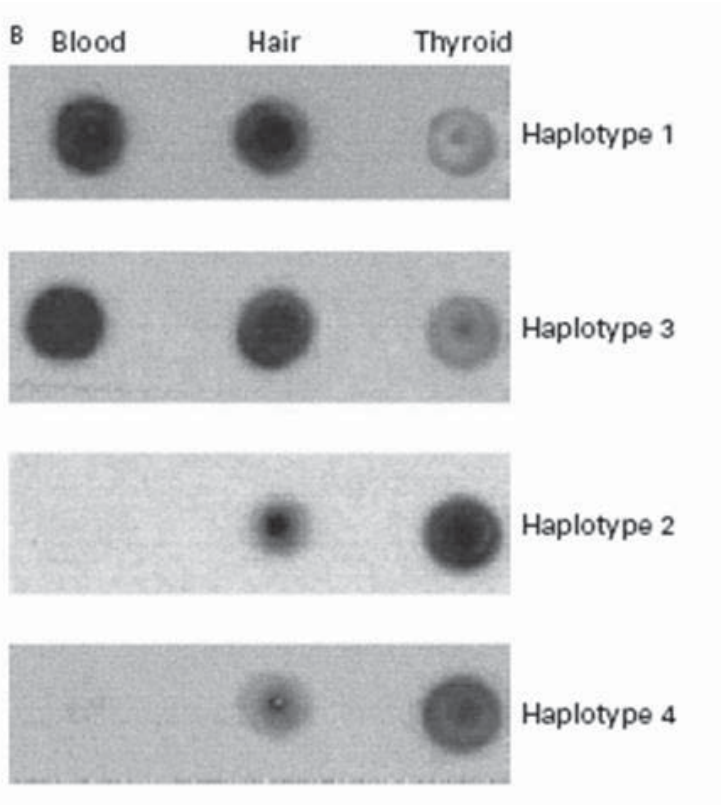
Questions

1. Add these data to your family tree.
2. Using what you know about genetics, deduce the likely haplotype combination of Karen's father and add it to the pedigree.
3. What do these data indicate about Karen's relationship to her sons?

Part IV—Hair and Thyroid

The data suggested that Karen's two sons were related to her one brother, so they must, in theory, be related to her as well. Because Karen's blood cells provided no match, the team of researchers decided to test some of her other tissues as well, including her hair and thyroid gland. The results are shown in Figure 2.

Figure 2. HLA haplotype testing of Karen's blood, hair and thyroid tissues.



(Figure originally published as Panel B of Figure 1 in: N. Yu et al., 2002, The New England Journal of Medicine 346 (20), 1545–1552. Copyright © 2002 Massachusetts Medical Society. All rights reserved. Used with permission.)

Questions

1. What HLA haplotypes does Karen have in her blood? Her hair follicles? Thyroid?
2. What do these results indicate?

Part V—Conclusion

Karen is a whole body chimera, otherwise known as a tetragametic chimera, meaning she resulted from the fusion of four gametes—two eggs and two sperm (see Figure 3). Therefore, her mother released two eggs that were separately fertilized by two sperm. This usually results in fraternal twins, but in Karen’s case, these two zygotes fused into one organism. One zygote went on to give rise to some of Karen’s cells, while the other zygote formed other cell types, giving Karen different genetic make-ups depending on what cell was examined.

The results were hard for Karen to cope with. Breaking the news to her sons was the hardest part for her. “I felt that part of me hadn’t passed on to them,” she explained.

Karen ended up accepting a kidney transplant from her husband and she is now a mixture of three genotypes.

Question

1. What are the implications of the discovery of Karen’s condition?

Figure 3. Karen is the fusion of two zygotes, each resulting from the separate fertilization of two eggs by two sperm. If this fusion had not occurred, fraternal twins would have resulted. Therefore, different parts of Karen’s body carry different sets of genetic information.

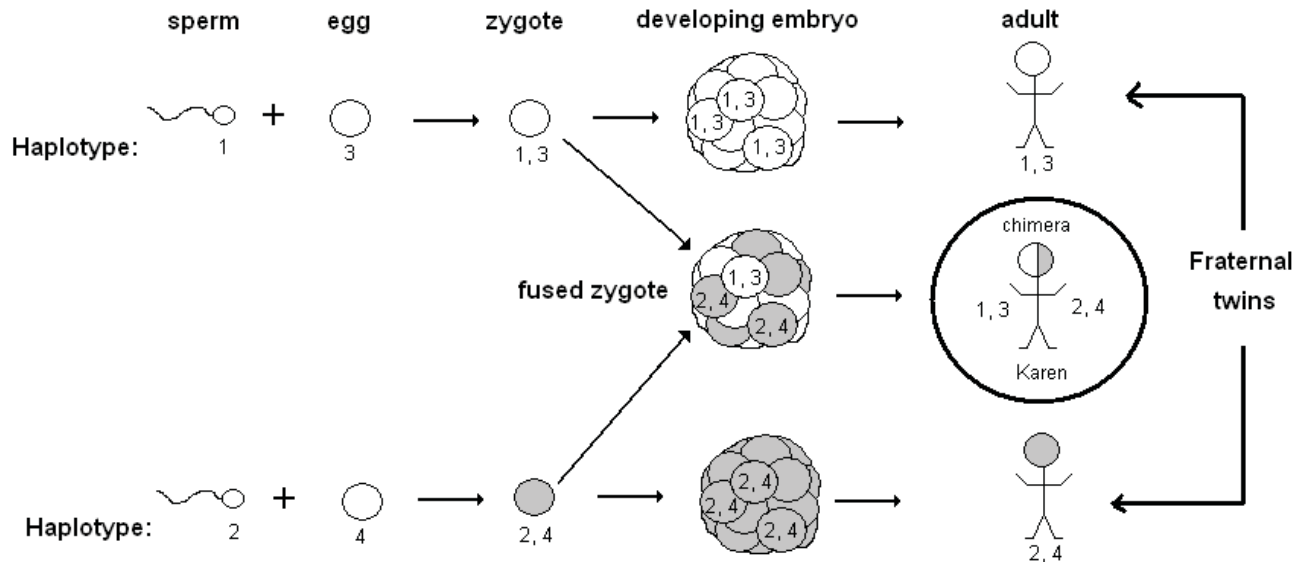


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