The Zygosity Puzzle

by Geoffrey Machin, M.D.

Boy/girl twins not identical, but monozygotic



This is probably a stupid question, but can identical twins be of opposite sex?

A: Did you notice a very interesting thing in the two most recent issues of TWINS Magazine? In a letter (Family Talk, November/December 2002, page 7), Darla from Regina, Saskatchewan, Canada, referred to her twins as monozygotic. The national twins organization in Canada switched terminology from "identical" and "fraternal" to monozygotic and dizygotic about a year ago, never again to use the ambiguous "identical" and "fraternal."

In Patricia Malmstrom's interview (Multiple Matters, January/February 2003, page 8) with Dr. Elizabeth Bryan, a pioneer worker with twins and their families in the UK, not once was the word "identical" used. Twins are either one-egg (monozygotic, MZ) or two-egg (dizygotic, DZ) and that is what Dr. Bryan and many twins' parents in the UK call them. What a great idea if the USA would catch up with the rest of the world!

In my experience, the biggest problem for parents who don't know the zygosity of their twins is that they expect MZ twins to be "identical." But MZ twins never are identical. There are always differences that allow parents to tell their MZ twins apart most of the time. In my experience of offering zygosity advice to twin parents, we find out that well over 90% of confused parents have MZ twins, when we have figured the answer. So why go on calling MZ twins "identical" when they aren't? I'll never understand it.

Which brings me to your question. Just exactly how unalike can MZ twins be-does it stretch as far as being of opposite sex? The answer is that MZ twins can be amazingly unalike, and there are boy/girl pairs. Two main reasons help

explain why MZ twins can be unalike, to small or large degrees. Different environmental experiences during prenatal life, e.g., donor and recipient of twin transfusion, very unequal fetal growth, can contribute to the differences.

Perhaps the most surprising thing is that some MZ twins are genetically different from each other. This simply means that, although they originally had the genetic composition of their original fertilized egg, one of them has undergone a genetic change during very early embryonic development. This could involve a whole chromosome, part of a chromosome or a change in a gene. All of these genetic abnormalities are seen in MZ twins, though they are rare.

How could you have girl/boy MZ twins? There are at least two possible mechanisms.

In one case, if the fertilized egg contains a Y chromosome (normal male chromosomes are 46, XY), genes on that Y chromosome ensure that the developing gonads are testicles. If there is no Y chromosome (as in a normal, 46, XX girl), the gonads become ovaries. But not all girls have normal female chromosomes (46, XX). A small number only have one X chromosome (45, X) and they have some developmental problems. These children have Turner syndrome. Now, suppose you have a 46, XY fertilized egg, but, early in cell divisions after fertilization, a cell loses its Y chromosome? Now you have two types of cells in the developing embryo-46, XY and 45, X. If the embryo splits to make twins, you can have a normal boy (46, XY) and a Turner syndrome girl (45, X). This has happened on several occasions and is well known to geneticists and researchers.

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Alternatively, the original fertilized egg could have one extra X chromosome (47, XXY). This condition is called Klinefelter's syndrome. If there are losses of chromsomes (as described above), you could have one 47, XXY cell that loses an X to become 46, XY (normal boy) and another 47, XXY cell that loses the Y to become 46, XX (normal girl). If MZ twinning occurs also, you end up with a normal boy and a normal girl.

These chromosome changes after fertilization are quite rare, but they have been fully investigated. They show that MZ twins are not always genetically "identical," although most people wrongly assume that they are-yet another reason to stop calling MZ twins "identical."

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