In layman's terms, what is Trisomy 18?

Dr. Melanie A. Manning answers questions about Trisomy 18. She's one of the medical geneticists at Lucile Packard Children's Hospital at Stanford.

Dr. Melanie A. Manning: Trisomy 18 is a genetic condition in which an individual has one extra chromosome more than the typical 46 we have in our cells. That extra chromosome is No. 18. Chromosomes 1 through 22 are the same in boys and girls, and we have two copies of each. The last pair determines our sex.

What causes the extra chromosome?

Manning: We don't understand all the mechanisms, but the extra chromosome can be in a sperm cell or an egg cell. When those eggs or sperm are being formed and the chromosomes are being divided, they can't contain 46 chromosomes each -- that would be too many for the final genetic code. So each egg has 23 and each sperm has 23. But sometimes the division of chromosomes doesn't happen as expected and you get an extra one. In that case, when the egg meets the sperm, you get a total of 47 chromosomes.

And why is that a problem?

Manning: Our chromosomes contain the genes that are the individual units of inheritance or the determiners of all of the features that make us human beings. If additional information is there that shouldn't be, it can cause organ system development to be altered. There are really only three common Trisomies: 13, 18, and 21. Those children tend to survive after birth because the developmental differences in the brain might not be as severe. You can have Trisomies of all of the other chromosomes, but that extra information is just too much and makes the development of the fetus not viable. We don't understand all of it but that extra information is just something that's not tolerated.

Do we know what causes the irregular chromosome division?

Manning: Maternal age is associated with an increased likelihood of not having everything go quite right. But it can also happen in sperm, and that's been shown to not necessarily be associated with paternal age. For that matter, it's not always associated with maternal age. There are plenty of children born with Trisomy 13 or 18 to parents who are only 18 or 21 years old. So there must be something we don't understand yet.

What are the symptoms of Trisomy 18?

Manning: Often the babies are small at birth, they're underweight and their length is shorter than what's typical for their gestational age. There can also be differences in their facial appearance -- they often have very petite features. Their respiratory system is often weak, so things like pneumonia can be a common problem. One of the most characteristic features is they have an unusual overlapping of their fingers. Their hands can be clenched but their second and fifth fingers overlap the two middle ones. And finally, for whatever

reason, the connections from the brain that tell the body to keep breathing are altered. So that's one of the reasons children with Trisomy 18 often expire -- they just stop breathing.

How common is this condition?

Manning: Some estimates say it's as common as 1 in 3,000 for live births, others say it's closer to 1 in 10,000. It's not as common as Trisomy 21 -- or Down Syndrome -- but it's still one of the more common that we would see that are live born. On the other hand, Trisomy 13 is even less common than Trisomy 18, and the birth defects are even more severe -- you see heart defects, severe abdominal wall defects, and clefting in the mid-portion of the face, which can be an indication of severe brain abnormalities.

It's possible to test the child for these conditions before birth, correct?

Manning: Yes, there are some different signs that might indicate abnormality. For Trisomy 18, an ultrasound might pick up that the baby keeps his or her hands clenched, or that there's a heart defect, or they're smaller than they should be. Noticing these clues can indicate that there might be a chromosome abnormality, and testing would be able to check which chromosome has been affected.

And finally, how long do children with Trisomy 18 usually live?

Manning: The vast majority of these conceptions are miscarried. More than half of those born alive will probably not survive past one month of age and 90 percent will not survive past one year. As with any child, it really depends on the severity of their birth defects and the general health of the child. Because those birth defects are so severe for children with Trisomy 18 -- even in the best-case scenario -- there can be some long-term survival but we would expect their development to be greatly affected despite their overall good-health.