What is Trisomy 18?
Around 1 in every 5,000 babies is diagnosed with Trisomy 18, also known as Edwards syndrome. Normally, a person has 23 pairs of chromosomes. Chromosomes are the packages of genetic information, made of DNA, that contain the instructions the body uses to build a person. Chromosomes come in 23 pairs, with most people having 46 total chromosomes. Trisomy 18 is caused when a person has three copies of chromosome #18 instead of the usual two, for a total of 47 chromosomes.

This extra chromosome affects a baby’s development, resulting in a number of medical issues which may include: heart defects, digestive tract abnormalities, cleft lip, joint contractures, vision and hearing problems, slow pre- and postnatal growth, seizures, and hypotonia (weak muscles). All babies that survive with Trisomy 18 have significant intellectual disability (usually in the severe range).

There are three types of Trisomy 18:
Full Trisomy 18
» The existence of a third copy of chromosome 18 in all of the cells.
» About 95% of cases of Trisomy 18 are this type.

Mosaic Trisomy 18
» The existence of a third copy of chromosome 18 in some of the cells.
» About 5% of cases of Trisomy 18 are this type.

Partial Trisomy 18
» The existence of a part of a third copy of chromosome 18 in the cells.
» Less than 1% of cases of Trisomy 18 are this type.

When a couple has a baby with Trisomy 18, it is usually unexpected. Nevertheless, some risk factors exist. For example, the chance of having a baby with Trisomy 18 is higher in older mothers. In other cases, Trisomy 18 can be inherited due to a familial chromosome rearrangement called a translocation. Trisomy 18 is never the result of anything a mother or father did, or didn’t do.

How is Trisomy 18 Diagnosed?
Pregnancies at increased risk for Trisomy 18 may be identified through screening tests such as non-invasive prenatal testing (NIPT) and ultrasound examinations. The diagnosis can be confirmed prenatally with better than 99% accuracy through chorionic villus sampling or amniocentesis. The diagnosis can be confirmed shortly after birth through blood testing.

If I Have Been Told My Unborn Baby May Have Trisomy 18, What Can I Expect During My Visits at The SSM Health St. Louis Fetal Care Institute?
When a family visits the Fetal Care Institute for the first time, a Level II detailed fetal ultrasound will be performed. If an amniocentesis has not yet been done, this test is also available.

When a family with a presumed diagnosis of Trisomy 18 in their baby visits the Fetal Care Institute, they have the opportunity to meet with a team of experts who can provide information about their baby’s diagnosis. This will involve maternal-fetal medicine specialists, genetic counselors, clinical geneticists, social workers, Footprints coordinators, and neonatologists. Additionally, based on the specific issues in a given pregnancy, the family may meet at a follow-up visit with other pediatric specialists. A Fetal Care Institute nurse serves as the family’s primary contact and coordinates the mother’s and baby’s care throughout the pregnancy.

The goal of the Fetal Care Institute team is to provide families with the support, information, knowledge and options to make the decision that is best for their baby. We recognize that the diagnosis of Trisomy 18 affects a family greatly and our hope is to be able to provide families with the resources they need during a difficult and overwhelming time.
How Does Trisomy 18 Affect My Baby?
In addition to having birth defects and cognitive impairment, about 60% of babies diagnosed with Trisomy 18 pass away before they are born. Of those who are born alive, approximately 10% are expected to survive the first year of life. We don't know why many of these babies pass away before birth or within their first year of life. We also do not understand why females with Trisomy 18 seem to survive longer, on average, than males.

Babies with Trisomy 18 can have multiple life-threatening medical issues at birth and throughout their lives. Because every baby’s situation is different, the Fetal Care Institute brings together an experienced team of specialists to meet with families to provide information, answer questions, and prepare them for the birth of their unique baby.

What are The Treatment Options For Babies With Trisomy 18?
Even though there is no cure for Trisomy 18, there are medical treatments that may be provided after birth that may improve the quality and duration of life for these babies. There is no known prenatal treatment that will improve the outcome for a baby with Trisomy 18, but our team can provide a family with support, education, and a safe environment in which to receive their care. Furthermore, treatment after birth may be available for some of the birth defects caused by Trisomy 18.

What are My Options if My Baby is Diagnosed With Trisomy 18?
The diagnosis of Trisomy 18 is just the beginning of a journey that can be challenging for any family. Our goal is to provide the information and options available for these babies and their families, and to be there as a support system throughout the pregnancy and beyond.

Families who receive a prenatal diagnosis of Trisomy 18 sometimes make the difficult decision to stop the pregnancy. Other families choose to continue the pregnancy. Continuing the pregnancy does not have any known risks for the mother, other than those associated with pregnancy itself. The team at the Fetal Care Institute will help the family develop a plan of care for the pregnancy and immediate newborn period. That plan can include the presence of the newborn medicine team at delivery to assess the baby’s medical issues and assist with the transition after delivery.

The decisions facing families at the time of the diagnosis of Trisomy 18 and throughout the pregnancy are difficult and personal. We strive to provide the options, resources, information, and support to help families make the decisions that respect and reflect their wishes on their child’s behalf.

How Does Trisomy 18 Affect Delivery?
Because of the complex nature of Trisomy 18, babies often face medical challenges during delivery and in the newborn period. Every baby’s situation is different, and the St. Louis Fetal Care Institute works with each family to prepare for their baby’s delivery and to create an initial working care plan to follow after birth. This plan can include medical interventions or be restricted to comfort measures, depending on the baby’s medical condition and the desires of the family.

What Can I Expect After My Baby is Born With Trisomy 18?
Babies with Trisomy 18 have varied outcomes. For some, the medical challenges they face will be significant enough that they will pass away shortly after delivery, while a small number of families can go on to celebrate their baby’s first birthdays and beyond. Babies that survive the newborn period will have significant cognitive impairment and other medical issues that will require advanced care throughout life. The pediatric specialists at SSM Health Cardinal Glennon Children’s Hospital remain available to provide the best possible care for every baby with Trisomy 18 from birth and beyond.