TRISOMY 13

NEW AND EXPECTANT PARENT RESOURCE GUIDE

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TO ALL THE
CHILDREN AND FAMILIES
IMPACTED BY TRISOMY 13

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INTRODUCTION:

The purpose of this book is to assist the expectant family that has received a possible or confirmed diagnosis of trisomy 13 (Patau syndrome). This book will help you understand your pregnancy and related decisions; and be a guide for you throughout your pregnancy and at the time of birth. We have bolded terms throughout the book that are defined in the Glossary.

PART 1: WHAT IS TRISOMY 13?

Trisomy 13 is a condition of human **chromosomes** that is caused by an extra or third 13th **chromosome**, hence trisomy 13. This extra 13th chromosome leads to a specific pattern of physical findings known as the trisomy 13 **syndrome**, also known as **Patau syndrome**.
(NOTE: genetic testing before or after birth must be done to confirm the diagnosis.)

The trisomy 13 syndrome has an impact on the health of the children who have the condition. It includes three major implications: the occurrence of medically important birth defects (especially of the brain and heart), a higher than expected frequency of infant mortality (making it a potentially life-limiting condition), and a developmental disability in older infants and children.

The actual reason or cause of the extra 13th chromosome is not known even after years of research. There is no way to have prevented its occurrence prior to conception, and there is nothing you could have avoided or done to stop it from happening.

> **"I was scared and sad and angry (why me, why us?). This was my welcome to parenthood."**

There are three types of **chromosome** findings seen in persons with the trisomy 13 syndrome. Approximately 90% will have a full or complete trisomy 13 (see Figure 1) in all body cells. The remainder (about 10% of infants) will have a partial trisomy, an extra portion of the 13 chromosome due to a rearrangement of part of the chromosome (usually attached to another chromosome, called a **translocation**), or have **mosaicism** (a mixture of two different cell populations, usually some normal cells and some trisomy 13 cells).

Those infants who have trisomy 13 due to a **translocation** usually have a special type of rearrangement called a Robertsonian translocation. In this chromosome change, the long arm (called the **q arm**) of chromosome 13 is attached to the long arm of another chromosome (usually the number 14 chromosome) resulting in 3 number 13 chromosome long arms. These 3 copies of the chromosome arm cause the **trisomy 13** or **Patau syndrome.**

In translocation types of trisomy 13 it is recommended that parents have a chromosome study done to see if they carry a balanced form (no missing or extra important chromosomal material) of the translocation. It is especially important for parents of infants with a translocation type of trisomy 13 to discuss the result with a genetic counselor or medical geneticist.

Chromosome study showing trisomy 13

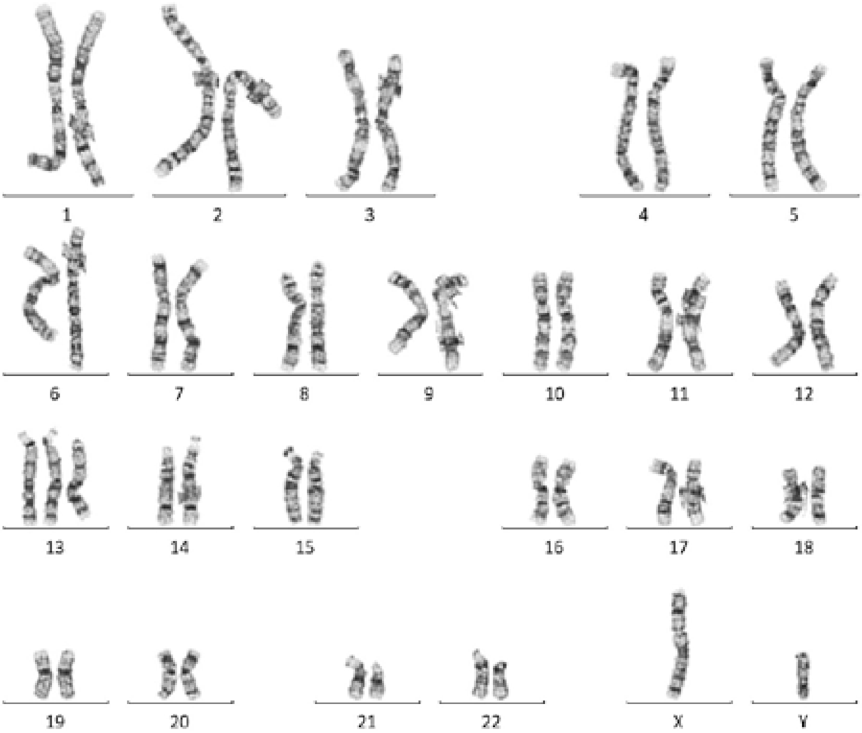

Figure 1: Courtesy of Dr. Erica Andersen and Monica Theriot, ARUP Laboratories

Note the 23 pairs of numbered chromosomes with chromosome 13 having 3 (trisomy) rather than 2 chromosomes.

PART 2: THE DIAGNOSIS OF TRISOMY 13 BEFORE BIRTH


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Prenatal testing to detect trisomy 13 can be divided into two basic groups: screening tests and diagnostic tests. Screening tests include quadruple screen, **noninvasive prenatal screening or testing (NIPS or NIPT,** and often called **cell-free DNA test)**, and ultrasound. These tests do not make a definitive diagnosis of trisomy 13. Diagnostic testing will be described further below.

The quadruple screen is an older test, requires a blood sample from the mother, and looks at specific hormones and proteins. It has essentially been replaced in recent years by the cell-free DNA test.

The NIPS or NIPT (cell-free DNA) test is a newer screening test that also requires a blood sample from the mother. This test looks for fetal DNA fragments that are from the placenta that are in the mother's bloodstream. The amount of fetal DNA in the mother's serum is measured. This test is typically offered at 10 weeks or later. While almost all fetuses who have full trisomy 13 will show the excess DNA, not all fetuses with a positive result will have trisomy 13/Patau syndrome. It is important to ask the doctor or genetic counselor how likely it is that a positive result predicts Patau syndrome **(positive predictive value PPV).**

An **ultrasound** can be used in the first trimester to look for extra fluid and thickness at the back of the fetus' neck. This type of ultrasound is typically performed by a maternal fetal medicine specialist (MFM). If there is extra fluid or thickness, this can be a sign of a chromosomal abnormality. An ultrasound during the second or third trimester of pregnancy can be used to look at the fetus' organs and various structures between 18-22 weeks. Findings such as suspected heart defects, brain abnormalities, and a cleft lip could be associated with trisomy 13 and may lead to your care provider suggesting further ultrasounds or diagnostic testing. Many of the physical findings seen on prenatal ultrasound are not medically important but provide clues to the diagnosis before birth (or in the newborn period). These can include close-set eyes, small jaw, and extra finger or toe.

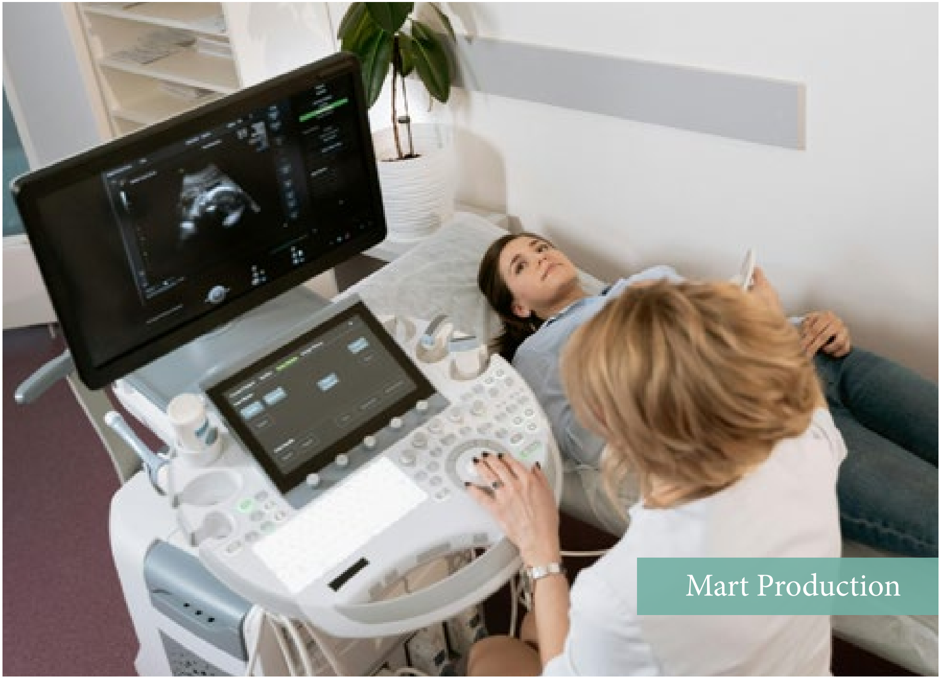
> **"I didn't find anything positive online when I googled Trisomy 13 while I was pregnant. It was all doom and gloom, and my fragile heart couldn't handle that."**

Since quadruple screen, NIPS or NIPT, and ultrasound are screening tests, a definitive diagnosis prenatally requires diagnostic testing. Diagnostic tests include **chorionic villus sampling** (CVS) at 10-12 weeks or **amniocentesis** at 15-16 weeks. Both are considered invasive tests and have less than 1% of pregnancy loss over normal risk for pregnancy loss.

CVS is performed in the first trimester, and involves taking a small sample of the developing placenta for the study. Due to this, CVS carries a slightly higher risk of pregnancy loss than amniocentesis. The benefit of CVS is that the procedure is performed earlier in pregnancy, and thus the result is back sooner (12-13 weeks of pregnancy compared to 16-17 weeks for an amniocentesis). Knowing early is important for families who would consider termination because abortion laws are changing rapidly and are becoming increasingly more restrictive depending on the state (see Part 9).

Amniocentesis can be performed anytime after 15 weeks and involves obtaining a sample of the fluid surrounding the developing fetus. Both the CVS and amniocentesis use ultrasound to guide the needle during sampling. These tests are most often performed by maternal-fetal medicine (MFM) specialists, or “high-risk OB’s.” While there are rare exceptions, CVS and amniocentesis are generally considered very accurate. Risks of both procedures include miscarriage, premature rupture of membranes, infection, or bleeding from the placenta; however, these are typically very rare.

PART 3: OTHER TESTS DURING PREGNANCY



Mart Production

Once the diagnosis of trisomy 13 is confirmed, the need for additional monitoring and testing during the pregnancy is offered to assist in providing the best possible outcome for both the mother and baby. This section will explain what these tests are and why they are performed.

If a genetic difference is suspected, the pregnancy is typically considered a high risk pregnancy, and will be followed by both a general obstetrician-gynecologist (OB-GYN) and a maternal-fetal medicine (MFM) specialist. They can perform the ultrasound mentioned above looking for changes in the organs (often called a “level 2 ultrasound”). Heart defects are commonly associated with trisomy 13, and they may either perform or recommend a special heart ultrasound of the fetus called a fetal **echocardiogram.**

A fetal echocardiogram evaluates the anatomy and function of the chambers of the heart. This test is typically done after 20 weeks gestation and looks for various heart defects such as ventricular septal defects (VSD), or other more complex cardiac defects (see Part 4). This specialized ultrasound will look at how the blood flows through the heart and vessels leading to and from the heart. The images from the echocardiogram will be interpreted by pediatric cardiologists (see Part 5) and MFMs to help determine what, if any, treatments need to be considered following birth. This echocardiogram may be performed again during pregnancy, or once the baby is born.

The most rewarding part of the journey was seeing Nora laugh and smile. Everyone who met her was touched by her spirit and it was amazing to see how she changed perceptions of teachers, nurses, physicians, therapists, and other parents who were given this prenatal diagnosis."

If a change in brain development or another organ is suspected, the care providers or team may suggest an MRI of the head and/or chest and abdomen.

There are other complications that can arise during pregnancy. **Fetuses** (babies inside the womb) with trisomy 13 are at an increased risk for miscarriage and stillbirth. The risk for the loss during pregnancy decreases as the pregnancy progresses.

Some mothers may require additional testing in the third trimester of pregnancy. These tests are typically performed along with fetal growth ultrasounds done every 3 to 4 weeks in the third trimester. They may include: fetal non-stress testing (NST), measurement of the fluid around the baby (amniotic fluid indexes, or AFI), and/or a biophysical profile (BPP). Each office has their own protocol for performing these tests.

- Non-stress test: Monitors are placed on the mother's abdomen to record contractions as well as the baby's heart beats. This test typically takes between 20 and 60 minutes.
- Amniotic fluid index (AFI): Ultrasonographic measurement of the fluid around the baby. Pregnancies complicated by genetic conditions are often associated with fluid abnormalities. This is performed to look at how the placenta is performing, how well the baby is swallowing, as well as making and voiding urine.
- Biophysical profile (BPP): Completed during an ultrasound and looks at the baby's heart rate, breathing, movement, muscle tone, and the amount of amniotic fluid around the baby. This test can be done after 28 weeks.

Other more specialized ultrasounds can be performed if there is concern for problems with growth. Umbilical cord dopplers are used to evaluate how well the blood flows from the placenta to the baby. They may also be performed more frequently depending on other ultrasound findings or growth problems.

The OB/GYNs and MFMs will use these third trimester tests to help determine the best timing of delivery. More concerning findings may warrant earlier delivery. However, otherwise uncomplicated pregnancies may be able to continue until closer to your due date. Multiple factors go into the timing of delivery, and this is a discussion that should occur with your care provider.

PART 4: WHAT DOES TRISOMY 13 MEAN FOR YOUR BABY AND FAMILY?

> **“During pregnancy, we were told Muhammad had a major heart defect called pulmonary atresia with VSD. The doctors believed he would be born with a genetic condition. He was later diagnosed with full trisomy 13. I felt like the earth beneath my feet had dropped out.”**

As mentioned in Part 1, trisomy 13 is sometimes referred to as Patau syndrome, named for the doctor who first described the condition. It includes the occurrence of medically important birth defects. We will discuss the birth defects and other medical issues in this section.

Birth Defects

Almost all infants with trisomy 13 will have **birth defects** (physical differences in an organ or body part); however, each child is different, and the occurrence of these individual challenges is highly variable. The most common and important problems are defects of the heart and brain.

About 80% of children with trisomy 13 will have a heart **malformation**. The actual defect varies, but the most common include:

- Ventricular septal defect (VSD): an opening between the lower chambers of the heart which prevents the heart from pumping blood correctly (a heart murmur is generally heard from this finding)
- Atrial septal defect (ASD): an opening between the two upper chambers of the heart making it difficult for the heart to pump sufficient oxygen-rich blood to body tissues (a heart murmur is often heard)
- Patent ductus arteriosus (PDA): a heart defect involving a persistent opening of the channel between two major blood vessels leaving the heart. This is normal during pregnancy,but this channel usually closes near the time of birth. Lack of closure can contribute to pulmonary hypertension (discussed below)

The majority of heart lesions are usually not those that cause major health problems in the newborn period (first 4 weeks of life). But sometimes, the heart defect in combination with respiratory issues, can lead to breathing difficulties and heart issues in the first two weeks of life. Only about 1 in 6 of children with trisomy 13 will have a more complicated or serious heart defect noted before or soon after birth.

These heart defects include dextrocardia, tetralogy of fallot, a double outlet right ventricle (DORV), and hypoplastic left heart syndrome (HLHS). The potential option for heart surgery should be discussed with the specialists prenatally and in the newborn period.

Other Medical Issues

Along with cardiac defects, the other common medically important conditions associated with trisomy 13 are concerns around breathing (respiratory). These difficulties can include:

Obstructive apnea: blockages of the upper airway due to floppy tongue (glossoptosis) or problems with the respiratory passages (the trachea and/or bronchi).

Central apnea: where the brain does not give the message to breathe (this finding is likely less common than previously thought). Central apnea can occur in babies that have differences in brain development (see below).

Aspiration pneumonia: often due to difficulty feeding/swallowing. This is when milk or food goes into the lungs causing irritation.

Pulmonary hypertension: high blood pressure in the arterial blood vessels of the lung; secondary to high blood flow through the lungs and/or obstructive apnea.

Because of the various challenges and differences, older infants and children are followed by the medical specialists who are experts in the particular problem. These specialists are usually located at children's hospitals. Your primary care provider can make referrals to the appropriate medical center for you (see Part 5).

Some hospitals have comprehensive care teams that help organize the care of **children with medically complex conditions.**

> **"At first everything was very scary. Every medical event was a "first" and we had to learn how to calmly and confidently handle things in the beginning."**

Other common birth defects and conditions seen in trisomy 13 (Patau syndrome) and their estimated frequency include:

- Differences in brain development including a problem in the formation of the fetal brain called **holoprosencephaly**; this defect is variable ranging from mild to more serious forms; it can be associated with physical differences in the face including changes in eye socket and nose formation: 50-60%
- **Cleft lip with or without cleft palate**, or a cleft palate by itself: 60-70%
- Differences in eye development often resulting in small or almost absent eyes: 60%
- Extra fingers and/or toes on the outer side of the hand or foot (polydactyly): 60%
- Esophageal atresia, blockage of the esophagus, with or without a connection to the trachea (windpipe): 5%
- Spina bifida (opening in the back from a defect in the spine): 3%
- **Omphalocele**: 10%
- Physical defects of the kidneys sometimes resulting in cysts: 50%
- **Aplasia cutis congenita** (scalp defect): 35-40%
- Seizures: about 25-50% of infants and young children develop a seizure condition, occasionally in the newborn period but usually later in childhood

- Feeding difficulties/challenges in the ability to breast or bottle feed, sometimes referred to as dysphagia: very common
- Developmental disabilities - significant limitation in the development of skills such as sitting, walking, and speaking are present in all children

> **“We were filled with so much love for our son and felt relief that he was breathing on his own.”**

PART 5: SPECIALISTS AND TEAMS


Frankie Gilmore Photo + Video

Now that your baby has been found to have features associated with Patau syndrome, your medical team may expand to include a number of specialists who can provide further diagnosis of current or potential medical needs as well as treatment options. Parents often find it can be helpful to engage a number of these specialists before the baby is born (prenatally).

Additionally, some communities have Fetal Care Centers where families might be referred.

A **Fetal Care Center** is often based at a children's hospital and provides teams of specialists making access to the appropriate professionals easier for parents. The number of fetal centers in the U.S. has increased over recent years. A current list can be found at fetalhealthfoundation.org/treatment-centers/

> **"Trisomy is an amplified version of why we go into medicine in the first place. There is a group of people that you can develop the capability to help."**

Prenatal Consults:
Prenatal consults can help parents better understand the capabilities and care provided at the hospital where you are delivering, and may help determine where you deliver your baby. Please keep in mind that consultations with some of the health care professionals listed below will be dependent on the ultrasound findings seen on the prenatal ultrasounds. Most families will consult with a neonatologist, an obstetrician and/or MFM specialist, a geneticist or genetic counselor, and a palliative care professional. If the baby has a heart defect, then meeting with the pediatric cardiologist and possibly the heart surgeon is important. If the baby has spina bifida, meeting with the pediatric neurosurgeon can be helpful.

- Obstetrician/Gynecologist (OB/GYN) -
  A physician who specializes in maternal health. They will be the primary physician during the pregnancy and will be responsible for directing and managing routine OB appointments, labor/delivery admission, and will most often perform the delivery. They may also work with Certified Nurse Midwives (CNMs) who may also be involved in the prenatal care/delivery. After medical school, training for an OB/GYN includes a four-year residency in obstetrics/gynecology.

- Maternal Fetal Medicine (MFM) -
A physician who specializes in taking care of high risk mothers and babies with medical issues primarily during their pregnancy. This doctor will likely be the first doctor you are referred to by your primary OB/GYN. They will help manage your pregnancy and provide recommendations regarding monitoring as well as delivery. After medical school, their training includes four years of residency in obstetrics and gynecology (OB/GYN) followed by a three-year fellowship.

- Medical Geneticist -
A physician who specializes in the evaluation, diagnosis, management, treatment, and counseling of individuals with inherited disease. After medical school, they usually complete an integrated residency program of two years of internal medicine, pediatrics or OB/GYN followed by two years of medical genetics; some train in a specialty like pediatrics and then complete a two-year categorical residency in medical genetics and genomics.

⊕ Genetic Counselor -
a healthcare professional who is trained in medical genetics and counseling. Certified genetic counselors work closely with physicians in OB/GYN, MFM, and medical genetics. They are very knowledgeable in genetic testing and genetic conditions.

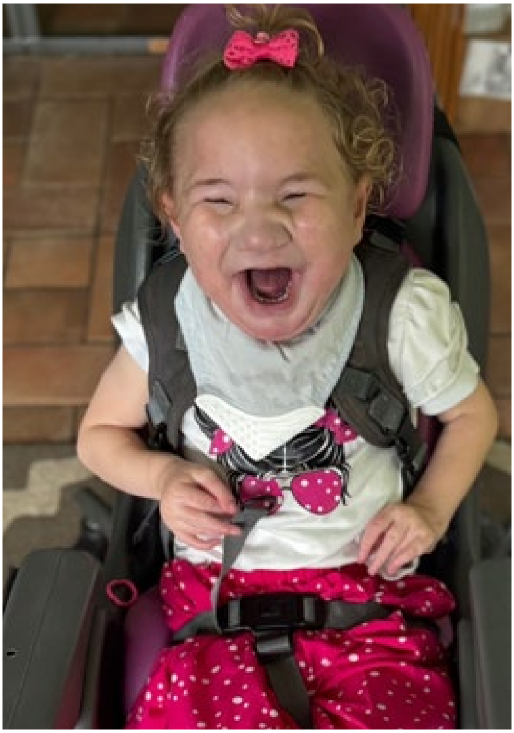
⊕ Pediatric Cardiologist -
A pediatrician that specializes in taking care of children with problems of the heart's structure and rhythm. They can look at your baby's heart before they are born by fetal echocardiogram (ultrasound of the heart) or MRI. They often work with pediatric cardiothoracic surgeons for planning regarding surgical repair of a heart defect. After medical school, their training includes three years of residency in general pediatrics followed by a three-year fellowship in pediatric cardiology.

⊕ Pediatric Cardiothoracic Surgeon -
A surgeon who specializes in taking care of children (and some adults) with serious heart or lung conditions that require surgical intervention. It may be helpful to meet with this specialist prior to delivery depending on the specific heart defect and/or your geographic location. After medical school, their training includes five years of residency in general surgery followed by a two to three years cardiothoracic surgery fellowship followed by an additional two to four years fellowship in pediatric cardiothoracic surgery.

⊕ Neonatalogist -
A pediatrician who specializes in the care of newborn infants, especially those who are ill or born early. They work in the Neonatal Intensive Care Unit (NICU). You can meet with them prior to your delivery to help set up a care plan for your child and also find out the type of care they will provide to your child. After medical school, their training includes three years of residency in general pediatrics followed by a three-year fellowship in neonatal care.

⊕ Pediatrician -
A primary care physician who specializes in taking care of children. It is important that this doctor is comfortable taking care of children with complex medical needs. After medical school, their training includes three years of residency in general pediatrics.

- Palliative care specialist - Physician or nurse with special training who provides support and guidance for families whose baby has a potential life-limiting condition such as trisomy 13 (Patau syndrome). These specialists support families in decision making both prenatally and after birth. A palliative care physician's training includes a residency in another area, like pediatrics, and a fellowship in palliative medicine.



Postnatal Consults:
Depending on your baby's medical needs, you may meet with additional specialists after your baby is delivered. These postnatal consults can happen in the hospital or after discharge. Based on your child's changing needs, additional specialists may become part of your care team throughout your child's life and sometimes even into adulthood. For other resources regarding physicians who may be part of your child's team, visit https://www.healthychildren.org/English/family-life/health-management/pediatric-specialists/Pages/default.aspx

- Pediatric Surgeon -
  A surgeon who specializes in treating newborns, infants, children, and teens. They may also meet with you before your baby is born if there are conditions that can be treated while in the womb or will need surgery immediately after birth. After medical school, their training includes five years of general surgery residency followed by a two-year fellowship in pediatric surgery.

- Pediatric Pulmonologist -
  A pediatrician who specializes in treating children with breathing problems and lung diseases. They manage conditions such as asthma, cystic fibrosis, and complex airways/lung disease that require ventilators and other breathing equipment. After medical school, their training includes three years of residency in general pediatrics followed by a three-year fellowship in pediatric pulmonology.

- Pediatric Neurologist -
  A pediatrician who specializes in treating children that have problems with their nervous system. This includes the brain, spinal cord, nerves, and muscles. They also have different tests like EEG that they can use to help look at electrical activity in the brain. After medical school, their training includes a combined child neurology residency program of two years of general pediatrics and three years of child neurology.

- Pediatric Neurosurgeon -
  A surgeon who specializes in children with birth defects or diseases of their brain, spinal cord, nerves, and muscles such as holoprosencephaly, hydrocephalus, and other conditions that can be associated with trisomy 13 (Patau syndrome). Depending on the condition, they may continue management of the condition into adulthood. After medical school, their training includes one year of general surgery followed by five or more years of neurosurgical residency with an additional year of training in pediatric neurosurgery.

- Pediatric Otolaryngologist (ENT) -
  A surgeon who specializes in treating diseases of the ears, nose, throat and upper airway. They perform surgeries such as ear tubes, **tracheostomy** placements, and are important in the evaluation of the upper airway; they use cameras that look into these areas to check the structure. They also play an important role in the assessment for potential hearing loss in trisomy 13 (Patau syndrome). After medical school, they complete one year of general surgery training followed by four years of ENT residency and one-year fellowship in pediatric ENT.

- Pediatric Orthopedic Surgeon -
  A surgeon who specializes in children who have bone deformities or broken bones. After medical school, they complete five years of residency in orthopedics followed by a one-year fellowship in pediatric orthopedics and/or pediatric spinal deformity.

- Pediatric Plastic Surgeon -
  A surgeon who specializes in conditions needing repair (reconstruction) of tissues such as a cleft lip or a scalp defect; they frequently work as part of a craniofacial or cleft palate team that includes orthodontics, speech and language therapy, feeding specialists, audiology, medical genetics, and oral surgery. These surgeons also can work with hand surgeons for deformities of the hand. After medical school there are actually 2 pathways. In the first, they complete a five-year residency in general surgery or an approved alternative subspecialty pathway such as neurosurgery, otolaryngology, orthopedic surgery, urology, or oral and maxillofacial surgery followed by a three-year residency in plastic surgery followed by a one-year fellowship in pediatric plastic surgery. In the second way they complete a six-year residency in plastic surgery followed by a one-year fellowship in pediatric plastic and craniofacial surgery.

- Pediatric Gastroenterologist -
  A pediatrician who specializes in children with diseases of the gastrointestinal tract, liver, and nutritional issues. This includes food allergies, difficulty swallowing, feeding, constipation, and problems with the pancreas or intestines.

They can also perform procedures like placing feeding tubes and using cameras to look at anatomy. Some hospitals have feeding or dysphagia teams often led by a pediatric gastroenterologist. They also commonly work with nutritionists who help with your child's diet. After medical school, they complete a three-year pediatric residency followed by a three-year fellowship in gastroenterology.

- Pediatric Ophthalmologist -
  A doctor who specializes in the medical and surgical care of eye diseases and vision difficulties in children. They manage conditions such as retinopathy of prematurity, lazy eye (amblyopia), eyes with high pressures (glaucoma), blocked tear ducts, and eyes that don't look in the same direction (strabismus). They also take care of children with vision difficulties such as nearsightedness (myopia), farsightedness (hyperopia), brain blindness (cortical visual impairment, and astigmatisms). They can treat these conditions with glasses, medicine, therapy, or surgery. You might also be seen for the differences in eye development that some children with trisomy 13 (Patau syndrome) may have. After medical school, they complete a one-year internship followed by a three-year fellowship in ophthalmology followed by one-year fellowship in pediatric ophthalmology and strabismus.

- Comprehensive Care Teams -
  An established team of specialists who work together with the other specialists for the goal of providing better communication among team members and with families. These specialized teams are available at some children's hospitals, and some hospitals in the U.S. are establishing teams specifically for the care of children with trisomy conditions (see Weaver in References).

PART 6: DECISION OPTIONS IN PREGNANCY, DELIVERY, AND THE NEWBORN PERIOD

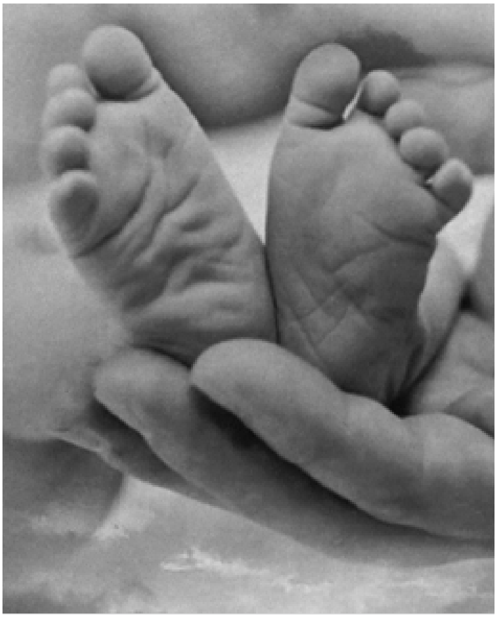
Many families ask what options they have and how to make decisions regarding their pregnancy after receiving a diagnosis of trisomy 13. There is no correct answer for this, just what is correct for your family and your baby.

After the diagnosis is made during pregnancy, families will face the hard decision to either continue or terminate the pregnancy (see Part 9). If continuing the pregnancy is chosen, further discussions with your OB care provider and other specialists regarding the care of your baby before and after birth are important. Conversations will focus on the type of care and interventions you want to pursue. This can range from purely comfort care to partial or full **intensive intervention**. It is not uncommon for you to meet with a multidisciplinary team consisting of your obstetrician, MFM, genetic counselor or medical geneticist, neonatologist, and hopefully a perinatal palliative care specialist. A pediatric cardiologist and/or heart surgeon are helpful when a heart defect is present in the baby.

> **"Decisions become acts of profound love, each choice a testament to the hope and heartache held within a parent's embrace."**

Deciding on a path of pure comfort care or intensive interventions are informed by the goals that a family establishes with their care providers. We have learned from both parents and research that recognizing the goals of care for the baby before and after birth is very important and provides a road map throughout the prenatal and postnatal periods.

The common decision points include:



- Fetal monitoring during labor. This would be performed to evaluate if the baby is tolerating labor.
- Cesarean birth if the baby is in distress before or during labor or has other reasons for the procedure. These discussions can occur with your pregnancy care provider during pregnancy and prior to labor.
- **Resuscitation** after birth in the delivery room. A newborn specialist or palliative care care specialist/team are helpful in this discussion.
- Medical and surgical interventions after birth. A family may choose full or some interventions or take a "wait and see" approach; there is much variability in how families proceed in choices surrounding care.
- Compassionate comfort care. Support for this decision is given by a pediatric palliative care doctor, nurse, or team. Pediatric palliative care teams are increasingly available in fetal centers, children's hospitals, and neonatal intensive care units throughout the U.S. and Canada.

Families tell us that it is helpful to have discussions about these choices prior to birth. Final definite decisions do not need to be made in the first meeting and ongoing discussions are helpful.

Decisions regarding medical and surgical interventions after birth:
Recent research shows that intensive and/or surgical interventions increase the chance that an infant with trisomy 13 (Patau syndrome) will live past the early months and first year of life and into childhood. Interventions include:

- Oxygen administration
- Respiratory technologies, such as CPAP and a ventilator to support breathing when needed
- Feeding support such as through nasogastric (NG) tubes or gastrostomy tubes (G-tube)
- Surgeries, including cardiac surgeries to correct heart defects, tracheostomy to support breathing, or other surgeries to address concerns such as spina bifida and esophageal atresia.

Ongoing Support
Parents caring for a child with medical complexity say that there are many stresses and challenges. We know that the risk for postpartum depression increases. Mental health resources are available for postpartum depression, anxiety, and depression. Pediatric palliative care specialists and primary care providers can give ongoing emotional support. Support groups including SOFT and local groups provide a unique perspective and are an important part of the parents' support network. If needed, parents are encouraged to ask their providers for other resources and services. Parents can also consider online services if that meets their needs.

PART 7: PREPARING YOUR BIRTHPLAN

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Having information about your baby's medical concerns and talking with your medical team can help you create a birth plan. A birth plan serves as a guide for how you envision your delivery, allowing you to consider your options and open communication with your birth team as you set care goals. While having a plan is important, it's also good to remember that sometimes babies, especially those with trisomy, have their own plans, and things may unfold differently!

> **"I learned more about trisomy 13 and the symptoms and treatments. I was able to bring awareness to the doctors and we worked together with them in treating my son's medical issues."**

Some of the following considerations may be limited by your pregnancy's specific health concerns and the hospital where you will be delivering. A birth plan lays out various choices at each decision landmark.

BIRTH PLAN

Parents names:

____

Your baby's name:

____

Name of your OB/GYN(s) and phone number(s):

____
____
____
____

Name of your baby's doctor(s) and phone number(s):

____
____
____
____

Name of important support person(s) and number(s) (friends, family, clergy, etc):

____
____
____
____

LABOR AND DELIVERY WISHES

Choose as many as you wish:

○ Vaginal birth
○ Cesarean birth
○ Fetal heart monitoring during labor
○ Who should cut umbilical cord

Labor Comfort

- ○ I would like to be able to move around as I wish during labor
- ○ I would like to be able to drink fluids during labor
- ○ An intravenous (IV) line for fluids and medications
- ○ A heparin or saline lock (this device provides access to a vein but is not hooked up to a fluid bag)
- ○ A birthing ball
- ○ A birthing stool
- ○ A birthing chair
- ○ A squat bar
- ○ A warm shower or bath during delivery
- ○ Music (what type:____________________)
- ○ A quiet labor room
- ○ I don't have any preferences

I would like the following people with me during labor (check hospital or birth center policy on the number of people who can be in the room):

____

____

It's OK [ ] **It's not OK** [ ] for people in training (such as medical students or residents) to be present during labor and delivery

I would like to try the following options if they are available (choose as many as you wish):

Anesthesia Options (choose one):

- ○ I do not want anesthesia offered to me during labor unless I specifically request it
- ○ I would like anesthesia. Please discuss the options with me
- ○ I do not know whether I want anesthesia. Please discuss the options with me

Delivery

I would like the following people with me during delivery (check hospital or birth center policy):

____

____

____

____

For a vaginal birth

○ To use a mirror to see the baby’s birth
○ For my labor partner to help support me during the pushing stage
○ I prefer to avoid an episiotomy unless it is necessary
○ For the room to be as quiet as possible
○ For one of my support people to cut the umbilical cord
○ For the lights to be dimmed
○ To be able to have one of my support people take a video or pictures of the birth(Note: Some hospitals have policies that prohibit videotaping or taking pictures. Also, if it is allowed, the photographer needs to be positioned in a way that does not interfere with medical care)
○ For my baby to be put directly onto my chest immediately after delivery, if possible and safe to do so (please discuss any exceptions you have based on your baby’s medical needs)
○ To attempt breastfeeding my baby as soon as possible after birth
○ Other: ____

For a cesarean birth

- ○ I would like the following person to be present with me: ______________________________
- ○ I would like one of my support people to hold the baby after delivery if I am not able to
- ○ I would like one of my support people to go with my baby to the nursery
- ○ I would like my baby to go to NICU if indicated
- ○ Other: ____

BABY CARE PLAN

Medical Care

- ○ If baby appears to be in distress, I desire a cesarean section if otherwise indicated
- ○ We desire to have NICU staff in attendance for the birth
- ○ If baby needs respiratory help we desire:
- ○ All interventions indicated to include: oxygen supplementation, pressure support, and/or intubation if indicated
  - ○ Only oxygen and pressure support
  - ○ Only oxygen support
  - ○ No intervention
- ○ If baby is in severe distress and would otherwise need CPR, we request:
  - ○ All interventions to include: chest compressions, IV insertion, necessary medications and fluids
  - ○ Medications only
  - ○ No intervention

- ○ We desire our baby to be admitted to the NICU if otherwise required
- ○ We desire measures such as OG, NG tubes as indicated for feeding
- ○ We desire comfort care only
- ○ No NICU admission. Let us have a quiet, separate room
- ○ No invasive feeding measures such as orogastric tube, nasogastric tube, or G-tube for feeding
- ○ We choose to be consulted and involved in all end-of-life care decisions and to ensure that our baby receives care that is consistent with comfort, dignity, and our values
- ○ Wishes for delaying routine procedures or providing them while baby is in parent's or support person's arms
- ○ We desire confirmation testing for trisomy 13, Patau syndrome
- ○ We desire all indicated consults while in the NICU
- ○ Other: ______________________________

Feeding the Baby

Keep in mind that the baby's ability to breathe and swallow may be compromised and may require necessary interventions to support breathing and feeding.

I would like to (check as many as you wish):

- ○ Breastfeed attempts
- ○ Bottle-feed attempts
- ○ Tube feeding
- ○ A pacifier
- ○ Sugar water
- ○ Breastmilk
- ○ Formula
- ○ IV nutrition (TPN)

POST BIRTHING PLAN

We understand that our baby's condition may be life-limiting, and we are committed to ensuring that our baby receives the best possible care while maintaining comfort and quality of life.

- ○ Would you like to go home at discharge
- ○ Code status at discharge
- ○ Name of support team (home health, hospice, etc) that will be assisting at home
- ○ Anticipated needs at home
- ○ Plan if emergency care needs arise at home
- ○ Other: ______________________________

Plans if baby dies before discharge

- ○ Plans to ensure baby is comfortable during the dying process
- ○ We desire to keep baby in room with family
- ○ We desire for organ/tissue donation if eligible
- ○ We desire for further testing after death
- ○ Funeral home information
- ○ Wishes for your child's funeral and burial arrangements
- ○ Special wishes about transportation of baby's body
- ○ Discuss the possibility of organ donation
- ○ Other: ______________________________

MEMORY MAKING

Wishes for memory-making and support

- ○ Do you wish to have any siblings or family members involved and if so, when______________________________

○ Do you wish to have a photographer (not all hospitals have a photographer on staff)

○ Keepsakes: footprints, handprints, heartbeat recording, hand molds, foot molds, locks of hair, crib card, ID bands, blankets, clothing, etc

○ Wishes for baby and special outfits

○ Spiritual rituals and/or wishes to be followed during delivery and after-birth care

○ Other: ____

PART 8: ADVOCACY


Frankie Gilmore Video + Photo

Communicating about what you believe your baby needs can sometimes feel difficult, especially with all the information coming your way and the many decisions that need to be made. It's natural to feel overwhelmed.

> **"Nothing is guaranteed. Absolutely nothing, not the negative things that you will be told and not the positive things either. Trust your gut...and heart. If you can handle doing the research, do it. If not, connect with support groups to help you get through all of this. You are not alone."**

Medical advocacy means working with healthcare providers to gather the information you need to make the best decisions for your child's care. However, it's equally important to remember to advocate for your own emotional and mental health. Seeking support, including counseling, can be an essential part of coping with the challenges of caring for a child with complex needs. There are simple, helpful steps you can take to become a strong advocate for yourself, your family, and your baby.

- Be aware: Learn about trisomy 13 and other conditions your child has. (see Resources, Care book, and trisomy.org)

- Learn needs: Ask questions to learn about the specific needs your child may have and explore the available care options for each of those needs. Take time to familiarize yourself with these options and consider what may be the best fit for both your family and your child (and remember, it's okay to adjust your decisions as needed). Example: Will your baby require breathing support? Will surgery be necessary, and if so, what type(s)? Will feeding assistance be needed? Inquire about any educational classes or resources that can help you become comfortable with any equipment you may need for caring for your child at home.

- Be assertive: This doesn't mean being aggressive, but it does mean expressing what you feel your child needs, listening to feedback, and having an open conversation with care providers. Everyone does want what is best for your child, but there may be a difference of opinion on what "best" looks like.

- Be self confident: You know your child better than anyone. You know your family better than providers. You are the best resource!

- Get support: online support groups (SOFT Facebook groups) and meet in person with local trisomy families by connecting with SOFT online.

"I was too upset to talk to anyone about it at first. I wish I had joined the Facebook community earlier. But I did when I was ready."

Remember your Patient Rights:

- For your needs to be met with dignity, respect, courtesy, and in a responsive and timely manner

- To receive information from your physician and have the opportunity to discuss risks and benefits of having or not having the treatments, and costs

- For your physician to provide guidance about what they consider the ideal course of action based on objective professional judgement
- To ask questions about your health, recommended treatments and to have those questions answered
- To make decisions about your care and your child's care and to have those decisions respected by the care team
- To have physicians and other staff respect your privacy and confidentiality
- To obtain copies of your medical records
- To obtain a second opinion

> **"My husband followed, as a team of doctors quickly rushed her into the next room while I lay there, feeling alone. But then I heard the most beautiful sound – Zoey crying!"**

PART 9: OTHER CHOICES

Cottonbro Studio

Some parents feel overwhelmed and frightened and don't know if they can do this. Every parent is different. This part will discuss other available options.

> **"In the tender space between anticipation and sorrow, parents facing the birth of a trisomy baby find themselves at a crossroads where every potential path is paved with profound love, deep reflection, and the quiet strength of cherishing each moment given."**

Pregnancy Termination/Abortion

After receiving a prenatal diagnosis and learning about trisomy 13, some parents will choose to terminate the pregnancy. An obstetric provider, medical geneticist, or genetic counselor can provide safe options for termination in your area or surrounding region. Abortion care has changed rapidly in the U.S. since 2022, so it is crucial that you understand the current laws in your state. This may mean traveling to a different state for the termination. At this time, restriction is based largely on gestational age/presence of a heart beat.

As a general rule, termination is typically safer the earlier it can be performed. Most commonly, pregnancies with a baby with a chromosome condition are not diagnosed and confirmed until late first trimester or in the second trimester, which may limit a family's options regarding termination in their state.

There are two basic modes of termination: medical and surgical. Medical termination involves the administration of particular medicines that cause an abortion; it is done in the first trimester usually by 10 weeks and can often be performed as an outpatient. Surgical termination, particularly in the second trimester, is performed by a procedure called dilation and evacuation (D&E) and is done as an in-patient.

The issue of mental health and pregnancy termination is complex and research can be conflicting. As with any person receiving unexpected news, it is important to consider utilizing mental health resources for help both during and after the process. Because pregnancy termination can be associated with depression and other reactions, some women will appreciate referral to supportive resources.

Adoption

It is possible for birth parents to choose to place their child for adoption. There are families that seek out adopting children with trisomy 13 (Patau syndrome).

> **"Joey came to us unexpectedly through adoption. We were excited about this new little life coming into our lives. The fact that he had trisomy 13 was just an added bonus!"**

The first step when making the decision to place a child for adoption is to talk with a social worker and an adoption agency. The second step is to choose the adoptive family. You will need to share as much medical information as you have about the child to ensure that the adopting family is prepared for all that the child will need. You also need to decide the amount of involvement you want with the family for the remainder of your pregnancy and also with the family after the child is born.

RESOURCES

www.trisomy.org

SCAN HERE TO ACCESS LINK ABOVE

Care Book https://trisomy.org/resources/parenting-a-child/care-book-trisomy-18-trisomy-13/#/

SCAN HERE TO ACCESS LINK ABOVE

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GLOSSARY

AMNIOCENTESIS — a prenatal diagnostic technique where a needle is placed into the uterus and a small amount of amniotic fluid is withdrawn usually for performing testing

ANENCEPHALY— a birth defect where there is a problem in the development or formation of the fetal brain and skull

APLASIA CUTIS CONGENITAL — an area on the top back part of the scalp that does not form completely leaving a patch of partly missing skin,usually an ulcer, and is treatable

BIRTH DEFECTS — congenital abnormalities of the structure of organs or body parts

CELL-FREE DNA SCREENING — analysis of DNA fragments released into the bloodstream; see noninvasive prenatal screening

CHILDREN WITH MEDICAL COMPLEXITY — a group of children and youth with special health care needs including at least 3 multiple significant chronic health problems and functional limitations

CHORIONIC VILLUS SAMPLING (CVS) — a prenatal diagnostic technique where a small amount of placental tissue (chorionic villus) is taken for performing testing

CHROMOSOME —threadlike structures seen only by microscope in the nucleus of cells and carrying the genes; they have 2 arms, a long and short arm, separated by a central narrowing called a centromere

CLEFT LIP AND /OR CLEFT PALATE — lack of closure of the lip causing an opening of the upper lip and/or the roof of the mouth

ECHOCARDIOGRAM — an ultrasound study of the heart, if prenatal, a fetal echocardiogram

GLOSSARY CONTINUED

FETAL CARE CENTER — multidisciplinary teams recently established in the U.S and Canada to help manage complex pregnancies especially those with fetuses with birth defects

FETUS — developing baby in the womb before birth

GENE —the fundamental unit of heredity and made up of specific DNA

HOLOPROSENCEPHALY — a birth defect where there is a problem in the development or formation of the fetal brain. This is often associated with facial deformities

INTENSIVE INTERVENTION — Medical treatment that is meant to prolong life, including ventilators and surgeries

MALFORMATION — an abnormal or irregular structure or defect that happens during development before birth. They can affect any part of the body

MOSAICISM — 2 or more genetically different cell lines in a person

NONINVASIVE PRENATAL SCREENING (NIPS) — maternal blood sample obtained at 10-11 weeks gestation to study cell-free DNA derived from the placenta that crosses into the maternal bloodstream

OMPHALOCELE — a birth defect where the abdominal wall doesn't close properly creating an opening where the abdominal organs, such as the intestines and liver, can develop and protrude outside the body within a thin transparent sac

GLOSSARY CONTINUED

PATAU SYNDROME — the pattern of physical findings and differences originally described by Dr. Klaus Patau in 1960 and caused by trisomy 13

POSITIVE PREDICTIVE VALUE (PPV) — the chance that a positive result of a screening test will predict a condition, for example, trisomy 13

Q ARM — the longer arm of a chromosome; the shorter is called a p arm

RESUSCITATION — process or act of reviving or correcting difficulties in an acutely ill person

STILLBIRTH — a fetus (baby in the womb) of greater than 20 weeks of pregnancy who dies in the womb prior to birth

SYNDROME — a recognizable pattern of multiple birth defects

TRACHEOSTOMY — a surgical procedure that creates an opening in the neck to windpipe (trachea) so a tube can be placed to help air and oxygen to get to the lungs

TRANSLOCATION — an exchange of chromosomal material between 2 chromosomes

TRISOMY — an extra copy of a chromosome

TRISOMY 13 — 3 copies of the # 13 chromosome and the cause of Patau syndrome

ULTRASOUND — a procedure using sound waves to look at tissues and organs; also called a sonogram

TRISOMY 13
New and Expectant Parent Resource Guide

Prepared By Support Organization for Trisomy 18, 13, and Related Disorders (SOFT)

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