# Trisomy Care Collaborative

## Information Guide for Families

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# Welcome to our program. We are here to support you wherever this journey takes you.

We know that you and your family are facing a challenging and uncertain journey. Our goal is to help you understand the decisions you might face, listen to you, support you, and communicate your care goals to your larger medical team.

*I will be forever grateful to Lurie's and the TCC team. When we first found out that our son Casey had Trisomy 13 at 12 weeks pregnant, we felt completely overwhelmed with worry and uncertainty and just didn't know how to even begin to navigate our journey.*

*When we finally found the incredible TCC team at Lurie's everything changed. We found a team that cared about Casey and whole-heartedly supported our goals of trying to give him the best shot at life we could give him.*

*We were able to ask them questions about anything and get guidance on decisions we didn't even know we would have to make. We just felt so taken care of and less alone. The access to support, information and guidance was invaluable.*

*We found out Casey had passed away at 26 weeks gestation while we were in with the TCC team having an ultrasound. All I can remember is the team surrounding us with love and took care of setting up all the next steps which involved going into hospital to be induced to deliver Casey sleeping. They were there for us every step of the way. They made what was a very challenging pregnancy feel that bit lighter for us. We will be forever grateful for their support.*

-- Casey's Parent (T13)

*"Learning about the diagnosis can fill you with fear and worry, but please know that the love you have for your child will carry you through – making the journey less frightening and more full of strength than you ever imagined."*

-- Maria's Mom (T18)

# Trisomy Care Collaborative (TCC)

## Who We Are

The TCC is a multidisciplinary group of clinicians – which means providers from many different specialties have come together to create this group. Our team includes members from Cardiology, Neonatology, Neurology, Otolaryngology, Pediatric Surgery, Cardiovascular Surgery, Palliative Care, Maternal-Fetal Medicine, Nutrition, Genetics, Nursing, and Social Work.

We know every patient and family is unique. Some families facing these genetic diagnoses will choose intensive in-hospital care, and we are here to give you an additional level of support for medical care planning and communication. And for those who decide on pregnancy termination or hospice, we are also here to give you education, support, and judgement-free care.

## What We Do

Our team is committed to improving personalized medical care for infants and children diagnosed with Trisomy 13 (T13) and Trisomy 18 (T18). We meet with families at different parts of their journeys with these diagnoses. We can meet prenatally (before birth) with expectant parents if testing shows that their child may be born with T13 or T18. For those families who choose intensive care after birth, we help make sure your child gets consistent care during their stay in the neonatal intensive care unit (NICU) or the cardiac intensive care unit (CICU).

We communicate with the primary medical team, help everyone follow clinical guidelines that our team developed based on the latest medical research, and advocate on your behalf. Our hope is that each child can reach their highest potential, while acknowledging and addressing the many challenges that your family may face. We also want to make sure that you feel supported no matter what you decide or what happens before and after birth.

## How We Can Help

We share knowledge of current recommendations and evidence-based practice when caring for children with T13 & T18. We use that knowledge to guide you and your medical team, helping you consider different pathways after birth. We can talk to you about what to expect (both short-term and long-term) based on your child’s unique characteristics, answer any questions you might have, and help streamline communication between different medical specialists. You can meet us once, or many times, depending on what you find helpful.

## What else is helpful to know?

- The TCC group meets every month to review patients and talk about their medical care. After the meeting, someone from the group will share feedback/recommendations with the medical teams & families.
- The TCC group can also meet as needed to discuss time-sensitive questions and support parents or medical teams.
- We can give second opinions about your child's medical care if they currently get care at another hospital.
- Members of your care team like Case Managers and Social Work can help you apply to different waiver programs. They will also help you get the medical equipment and support you need at discharge.
- Other trisomy parents have shared with us that it is important to clarify the **difference between palliative care and hospice**.
  - Palliative care is an added supportive layer of care that is focused on your baby living the best life given a significant medical diagnosis. It is **not** taking away treatment.
  - Hospice care is care for someone at the end of their life. It is focused on making them comfortable, spending time with their family (often not in the intensive care unit), and respecting their disease's natural progression.

# Prenatal Screening vs. Diagnostic Tests

When someone is pregnant, they can have screenings and tests done to see if there is a risk for their baby to be born with certain medical complications. Screenings come first to **see if there is a concern**, and then a diagnostic test is done to **confirm the concern** and make a diagnosis. Confirming a suspected genetic diagnosis lets your care team better treat the condition and understand how it will progress. Genetic testing can also help your care team understand the risk of future pregnancies being affected with these disorders.

## Prenatal Screening

It is common for someone who is pregnant to be offered a *non-invasive prenatal test* (also called "NIPT") early in a pregnancy to screen for extra or missing copies of the baby's chromosomes. One example of this is *cell-free DNA* (cfDNA) testing, which screens for a small number of genetic conditions that involve the baby having extra, or missing, chromosomes.

Humans typically have 23 chromosome pairs – which means we have 46 total chromosomes. The term **trisomy** means that there are 3 copies of a particular chromosome, instead of the typical 2 chromosomes to make the pairing. T13 means there are 3 copies of chromosome 13, and T18 means there are 3 copies of chromosome 18.

The care team uses an ultrasound to look at the developing baby and to see how the different parts of their body are forming. Most pregnant persons have a full anatomic ultrasound of their baby around

20 weeks of their pregnancy. A full anatomic ultrasound looks at all of the different organ systems in your baby's body.

If you have an abnormal NIPT screening or an abnormal ultrasound, this may lead to **increased concern** for specific genetic conditions, such as T13 or T18. But please know that these results alone **cannot confirm a diagnosis**. If there is a concern for a genetic condition, we will then do a diagnostic test.

## Diagnostic Tests

Diagnostic tests during pregnancy **confirm a suspected condition** that your doctor found during a prenatal screening. Your obstetrical provider can talk to you about these tests and order any needed tests during your pregnancy to confirm a suspected condition. Two types of diagnostic tests that your provider may talk to you about are *chorionic villus sampling* (CVS) or *amniocentesis*.

During these procedures, your provider takes a small sample of baby's genetic material either from the placenta (CVS) or from the fluid surrounding the baby (amniocentesis). Genetic testing can also be done after birth by taking a sample of your baby's blood.

That genetic sample is then tested to try and see if we can make a diagnosis that explains the concerns in the pregnancy. Once the genetic sample is analyzed in a lab, your provider will talk to you about the results. You will also meet a genetic counselor who will help you interpret and understand the results.

Many families have questions about the *variation of trisomy genetics* and hope to understand if the extra chromosome is seen in all cells (*full trisomy*), or only within some or certain types of cells (*mosaic* or *partial trisomy*). Your multidisciplinary medical team and genetic counselor can help you use this information to make decisions on how to best care for your pregnancy and your baby after birth.

# A diagnosis is confirmed, now what?

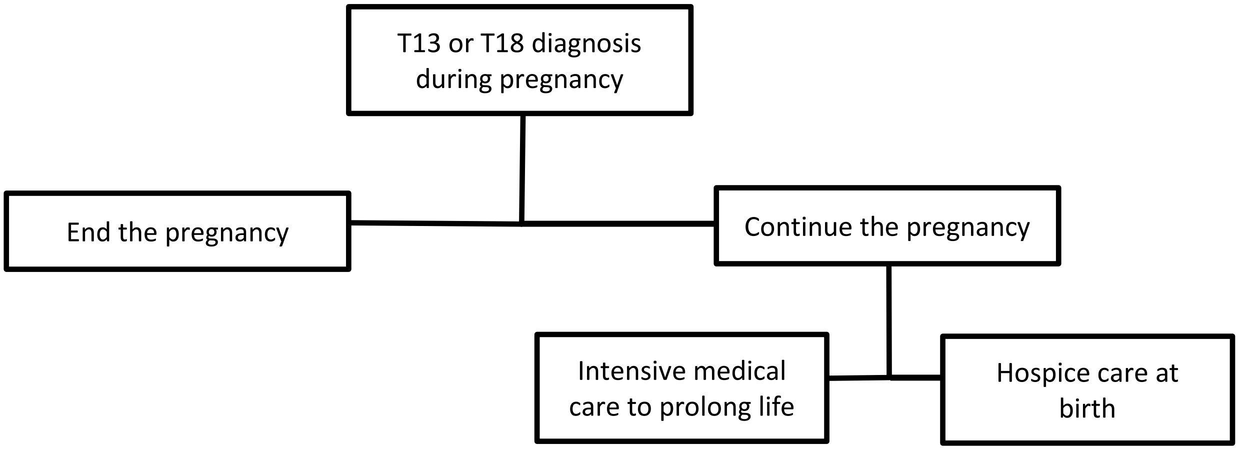
While awaiting diagnostic confirmation, many parents look for additional information about what they can expect. This can be overwhelming, as there is a lot of information online, including both medical facts and opinions. The available information can sometimes feel contradictory, or only seem to support one point of view. It is important to understand all available options and to have someone on your team who is both knowledgeable about the different decisions, and open to supporting you no matter what option you decide is right for your family.

## Potential decision points

After having comprehensive (complete) evaluation of your pregnancy and baby as well as a detailed discussion about the specific findings of the evaluation, your personal goals, and your values – parents can consider the following decisions:

- Continuing the pregnancy and deciding what to do after the baby is born: Parents can consider different options for their baby's care after birth, including intensive medical care or hospice support. Palliative care clinicians can help you think through these decisions and make a plan – for any scenario. They can also help you think through ways to combine these options.
- Ending the pregnancy: In the state of Illinois, pregnant persons are able to end a pregnancy any time before the gestational age reaches 23 weeks and 6 days (note that other state laws can be different). **We understand this is a very challenging and personal decision, and people have different views on this option. We respect that each family is unique and that having this option is important for some – for emotional, spiritual, or medical reasons.*

See below for another way to view these decisions points (which are **all** supported by our team):



A baby's life might be shorter if:

- They are born premature (less than 34 weeks old), even with medical care to prolong their life.
- They have significant congenital heart disease.

## More Helpful Information

- American Academy of Pediatrics (AAP): *Guidance for Caring for Infants and Children With Trisomy 13 and Trisomy 18: Clinical Report* https://doi.org/10.1542/peds.2025-072719
- Courageous Parents Network (CPN): https://courageousparentsnetwork.org/diagnosis/trisomy-18
- ParentWise: https://www.luriechildrens.org/en/patients-visitors/resources-support-services-for-families/peerwise-parentwise/
  Connect with other families at Lurie Children's who understand your experience.
- Positive Exposure: https://positiveexposure.org/images/
  This resource has images if you want to see people with these and other genetic differences.
- Prenatal Hospice & Palliative Care: https://www.perinatalhospice.org/
  A resource to help plan for a hospice birth.
- Support Organization for Trisomy (S.O.F.T.): https://trisomy.org
- The Edwards Syndrome Association: https://edwardssyndrome.org/
  *A local resource for T18*
- Trisomy 18 Foundation: https://trisomy18.org/

**En Español**

- S.O.F.T.: https://trisomy.org/publicaciones-y-documentos-en-espanol/#/
- CPN: https://courageousparentsnetwork.org/esp

**Faith-Based Support**

- Extra Chromosomes are Extra to Love: https://www.extratolove.org/
- Able Speaks: https://abelspeaks.org/
  (when continuing a pregnancy with a life-limiting diagnosis)
- Rory's Way: https://rorysway.org/

# Differences and Similarities

T13 and T18 are **unique conditions** with developmental differences associated with each condition.

There are also some differences that we often see in both conditions. Take a look below to learn more.

### Trisomy 13

Someone with T13 may have:

- Their brain structured differently (e.g., *holoprosencephaly*).
- A small area where their skin is missing (*Curtis aplasia*).
- A cleft lip or cleft palate.
- A smaller head size (*microcephaly*).
- Small or under-developed eyes.
- A defect in their abdomen (where your stomach and other organs are) where their organs are outside their body and not inside their abdomen (*omphalocele*).

### BOTH Trisomy 13 & Trisomy 18

Characteristics that we may see in someone with either T13 or T18:

- Breathing pauses that aren't usual (*apnea*).
- Their airway has a different structure, or is weak.
- They have weak bones.
- Their spine curves (*scoliosis*).
- Rocker bottom feet.
- They grow slower than other babies their age.
- It may take longer for them to reach certain developmental milestones, and they may not reach milestones past a child who is 4-15 months old.
- Heart defects found when they were born that impacts how it's structured or works (*congenital heart defects*).
- They are more likely to get infections like pneumonia or urinary tract infections (UTIs).
- Issues with their gastrointestinal (GI) tract—which is the system that helps you digest your food.
- Seizures.

### Trisomy 18

Someone with T18 may have:

- High blood pressure in their lungs (*pulmonary hypertension*).
- Tumors in their liver or kidneys.
- Their hip joint not fitting together the right way (*hip dysplasia*).
- Clenched fists with their 2nd and 3rd fingers overlapping.
- A small mouth/jaw.
- Their ears form in a different way.
- Eyes that are spaced apart wider than usual, and the openings are smaller than usual.

****Please note**—every baby is unique. Your baby may not have all of these symptoms. You may see many of these features, or just a few.

## Significant developmental differences

Most children with full T13 or T18 have significant developmental challenges. They tend to reach the developmental skills of a child who is 12-15 months old, regardless of how long they live. They also often develop these skills later than other children.

However, some children can have slightly more advanced skills. Kids with partial or mosaic trisomy may have an even wider range of developmental milestones.

The chart below shows the typical developmental milestones for babies without T13 or T18 who are between 4-15 months old. Many surviving children with T13 and T18 can reach these milestones, just at a slower rate. It is clear that many children can communicate (even if it's not using their words), laugh, play, and move around either on their own or with help.

**4-7 Months**

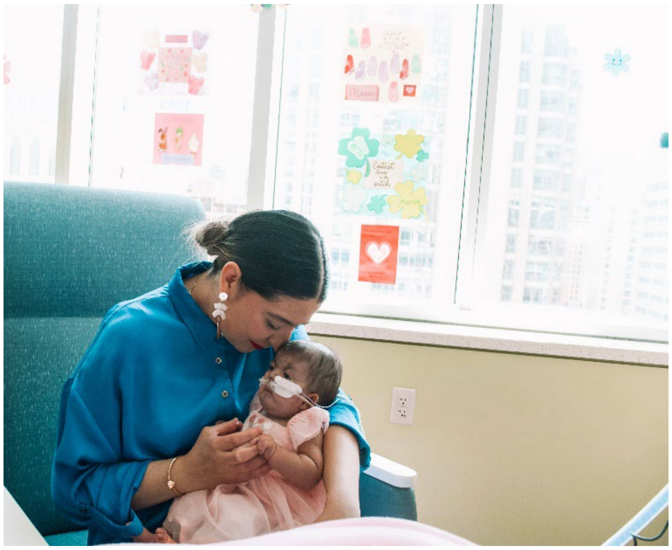
- Rolls (both ways)
- Reaches with 1 hand
- Responds to others expressing emotions
- Explores by bringing their hands to their mouth
- Knows familiar people

**8-12 Months**

- Sits without support
- Is fearful of strangers
- Imitates other people's gestures
- Looks at the right picture when you name the image
- Shows a preference for people/toys

**12-15 Months**

- Points to show you something
- Says "mama" or "dada"
- Can put a block (or something similar) into a container
- Plays games like pat-a-cake



# Other Helpful Information

## What is intensive medical care?

Babies who need support of their vital functions or surgery after birth will stay in the Neonatal Intensive Care Unit (NICU). In the NICU, medical teams can give your child the life-sustaining therapies such as invasive respiratory support (needing a breathing tube/use of a ventilator), medications that help their organs function, or surgery, as well as access to various sub-specialty teams. Many times these interventions work well to support babies so they can become ready to be discharged from the hospital. However, there are unfortunately times that babies are not able to survive even with these interventions.

Parents who chose intensive medical care for their child with T13 or T18 can expect:

- To meet with our perinatal palliative care team so that we can better understand your family's values and goals for your child's care.
- Your baby's first hospitalization could last many weeks, or even many months – it depends on their specific conditions. Some babies need up to 6-12 months in the hospital before first going home.
- Support from the TCC through that first hospitalization, as well as having multiple other subspecialists involved in your baby's care (depending on their specific conditions).
- A referral to your local Early Intervention (EI) Program when your baby is discharged from (leaving) the hospital.
- Routine screening tests for your child that include blood draws, abdominal (belly) ultrasounds due to the risk of developing tumors, and watching for seizures.
- Routine follow-up with a pediatric dentist, ophthalmologist, orthopedics, and possibly other medical professionals depending on your child's unique condition – in addition to regular well-child visits with their pediatrician.
- **If your child needs to have a tracheostomy** put in place before they leave the hospital, you will need home nursing in place before your baby can be safely discharged home. You may need go to transitional care facility while waiting for home nursing to be set up.

## What is hospice at birth?

This is a philosophy of care that focuses on maximizing your baby's comfort during what may be a short life, while also celebrating and making memories with your baby. It's important to know:

- The care practices of hospice do not try to bring death more quickly, but they also do not artificially prolong life. Hospice honors your child's natural abilities and limitations. It means we do not rely on medical devices or surgeries, and usually avoid the intensive care setting.

- This type of care focuses on your baby's comfort as the highest goal.
- We will care for your baby during their time at the hospital. When it's time to go home, they may be able to have home hospice support in place.
- Hospice supports the medical, emotional, and spiritual wellbeing of the entire family (including babies, parents, siblings, and grandparents).

## Common Medical Technology Words or Phrases You May Hear

- **Nasogastric (NG) tube**: This is a soft, flexible tube that goes into your baby's nose and the tip of the tube ends in their stomach. Babies born with T13 or T18 have a hard time taking their feedings *by mouth* (meaning swallowing their feedings). This is a temporary tube that lets your baby get the breast milk or formula that they need without them doing the work to drink it. You will work closely with your NICU team and Speech Therapist on how to best use the NG tube both in the hospital and possibly at home.
- **Gastrostomy tube (G-tube)**: This is a tube that goes through your child's skin, muscle, etc. on their belly and directly into their stomach. Your child will have surgery to put this tube in place. A G-tube gives you a more stable way to give your child the nutrition they need. It is usually offered for babies who will need this support for a longer time (as opposed to the temporary NG tube).
- **Fundoplication**: This is a surgery on your baby's stomach/esophagus to reduce symptoms of gastroesophageal reflux. Your child's care team may talk to you about it if your child is getting a G-tube. This is not always needed, but it helps some babies.
- **Nasal cannula** or **BiPAP support** (for older children): These are 2 different ways to give your child extra oxygen or needed pressure to their lungs/airway when they are at home.
- **Tracheostomy**: This is a surgery where a breathing tube is placed directly into your child's airway through their neck. This helps them breathe more comfortably if they need support from a ventilator. Kids with tracheostomies can live at home once their caregivers complete training on how to care for them using this device. You will have very detailed discussions with your care team about this procedure and using a ventilator if your baby needs it.
  Some reasons children get this procedure:
  - Some have something blocking their windpipe or their windpipe isn't strong enough to support their breathing.
  - Some need more pressure for air to reach their lungs and the ventilator does this for them. They may need it for months, or even years.

  Children who only need breathing support when they are sick will likely not need a tracheostomy.

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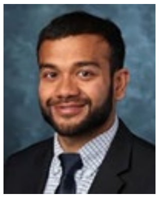

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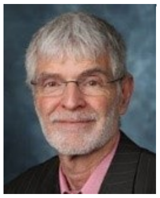

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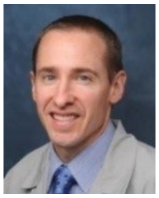

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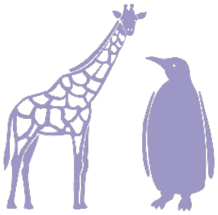

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**Jennifer Bathgate, DCN, RD, CSP, LDN**
Registered Dietician

## Lurie Children's Resources and Contact Information

- TCC: TCC@luriechildrens.org
- Chicago Institute for Fetal Health (CIFH): 1.866.338.2524
- CIFH website: https://www.luriechildrens.org/en/specialties-conditions/fetal-health/
- Lurie Children's Perinatal Palliative Care: https://www.luriechildrens.org/en/specialties-conditions/perinatal-palliative-care/

All,
for
your
one.®

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