



[www.LivingWithTrisomy13.org](http://www.LivingWithTrisomy13.org)

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## **Trisomy 13 Prenatal Diagnosis Information Package**

Compiled by Kylie Sheffield (mum of Daniel, full Trisomy 13, due 29 July) in Consultation with LWT13 Families

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### **Introductory note**

If you have recently been advised that your unborn child may have a potentially life-threatening condition, chances are you are currently experiencing a range of responses and emotions including confusion, disbelief, anger, denial and grief. You may have been told that your baby will die in utero, during labour or very shortly after birth or that survival will mean a life of profound disability and suffering. If your baby has a chromosomal disorder like Trisomy 13 (T13), you will almost certainly have been told that the condition is 'not compatible with life' and 'inappropriate to treat' or that your baby is 'genetically programmed not to live'. Your doctors have probably informed you that most parents in your position choose to end their baby's life and, in some cases, may even have pressured you to terminate your pregnancy.

Following this devastating news, parents are asked to make choices based on information that is often outdated, incomplete or grossly inaccurate. This information package has been compiled in the belief that access to current and accurate material can alleviate much of the confusion and loneliness experienced during those first days or weeks after detection or diagnosis; and empower parents to make informed decisions for their babies.

My husband Paul and I received a prenatal diagnosis of full T13 for our second son Daniel when I was twenty-two weeks pregnant and have spent the last four months collecting information to support the many decisions we will make for our baby. If you have received a T13 diagnosis prenatally, you are not alone. Help IS available. Read through this package, view the recommended resources and most importantly, don't be afraid to keep asking questions until you are satisfied with the answers you receive.

## **Detection – markers and what they might mean**

For many families the initial signs that something is wrong are detected during a routine prenatal ultrasound or fetal anomaly scan at around the sixteen to twenty-week mark. These signs are commonly referred to as ‘markers’. Simply defined, ultrasound markers are deviations from the normal anatomy seen on an ultrasound scan. Two common examples are echogenic intracardiac foci, which appear as white, golf ball-like features on the heart; and choroid plexus cysts, black spots in the brain which indicate a collection of fluid. In isolation, these and other markers are often of no significance and can appear in perfectly healthy babies. Occasionally, and more particularly if multiple markers are found, their presence may indicate an underlying chromosomal abnormality<sup>1</sup> or the presence of a similarly serious syndrome.

Having detected multiple markers, your ultrasound operator will most likely seek a second opinion, contact your obstetrician and schedule a more detailed scan. Further diagnostic testing in the form of amniocentesis may also be offered at this point.

The period of uncertainty following initial detection is extremely painful and traumatic, but it is important to remember that while prenatal ultrasound can detect specific anomalies, it CANNOT provide a definitive diagnosis of your baby’s condition. Nor is it possible, at this stage in your pregnancy, to predict how some internal organs will develop. Anomalies which initially appear to be significant may become increasingly less so or disappear completely as your baby continues to grow. For these reasons, no decision about your baby’s future should be based on the results of initial ultrasound findings alone.

### **Recommended reading**

Beryl R Benacerraf, MD. ‘The sonographic diagnosis of fetal aneuploidy.’  
<http://patients.uptodate.com/topic.asp?file=antenat/4468>

## **Further diagnostic testing – making the right decision for you and your baby**

Proceeding with amniocentesis is an obvious choice for some parents while others find the decision extremely difficult. The procedure carries a number of risks, most notably a 1 in 200 chance of miscarriage. Other potential complications include vaginal bleeding, amniotic fluid leakage and infection. If your baby is suspected to have a chromosomal condition, your doctors may warn of an increased likelihood of miscarriage following amniocentesis ‘because so many babies with these conditions are likely to spontaneously abort anyway.’ At this point you may wish to consider what exactly you hope to gain from agreeing to undergo amniocentesis and whether you are prepared for the further choices you will face should the procedure return a diagnosis of T13 or similar.

Where T13 is suspected at this stage of pregnancy, a definite diagnosis by amniocentesis may:

- allow expectant parents the option to end the pregnancy should they be unwilling to give birth to a baby with serious and potentially fatal chromosomal defects;

- assist parents and their doctors in managing the pregnancy and allow them to plan the birth most appropriate to the baby's individual needs, ensuring the availability of adequate support facilities and specialised medical staff;
- influence the obstetrics team's advice in terms of how the baby should be delivered (should amniocentesis confirm the presence of a chromosomal disorder deemed to be not compatible with life, some obstetricians are unlikely to recommend a Caesarian section during labour, regardless of any signs of fetal distress—the mother's health and viability for future vaginal deliveries may be prioritised due to the poor expected outcome for her baby).

While knowing there are sound reasons to justify amniocentesis does not necessarily make the decision to proceed any easier, ensuring you have all the available facts will at least allow you to make an informed decision.

### **Recommended reading**

Elizabeth Steine, CNM. 'Should I have an amniocentesis?'

<http://www.askyourmidwife.com/amniocentesis.html>

Alessandro Ghidini, MD. 'Patient information: Amniocentesis'.

<http://patients.uptodate.com/topic.asp?file=pregnan/6937>

Robert G. Resta. 'Genetic Counseling: Coping with the Human Impact of Genetic Disease'.

[http://www.accessexcellence.org/AE/AEC/CC/counseling\\_background.html](http://www.accessexcellence.org/AE/AEC/CC/counseling_background.html)

Wendy Hogarth. 'Amniocentesis: The Struggle to Choose'. Viewed May 2007

[http://www.humanehealthcare.com/Article.asp?art\\_id=755](http://www.humanehealthcare.com/Article.asp?art_id=755)

Monica Rafie (founder and administrator of BeNotAfraid.Net). 'A Different Kind of Crisis Pregnancy: When There is "Bad News" About Baby'.

[http://www.humanlife.net/view\\_reports.htm?rpId=23](http://www.humanlife.net/view_reports.htm?rpId=23)

### **Diagnosis: T13 – What now?**

Assuming your amniocentesis returned a positive T13 diagnosis, you will most likely be offered the option to terminate your pregnancy. At this point it is essential that you have access to current and accurate information. Request to see written reports of all test results and have your medical professional explain what they mean. Questions you may wish to ask your practitioner include:

- What form of T13 does my baby have (full, partial, mosaic, translocation) and what exactly does this mean?
- What are my baby's specific anomalies?
- I would like to seek further advice and undergo further diagnostic testing before making my decision—can you refer me to the appropriate specialists?
- Are you aware that a number of T13 babies do survive and that there are children and adults living with the condition today?
- What exactly will termination entail?
  - Can you explain the actual procedure?
  - What medications will I need to take and what are their known side effects?

- What are the potential complications and what risks do they present to my future health and fertility?
- What will my baby experience?
- Who will be present during the procedure?
- Will I have access to counseling before and after the procedure and can you recommend any support organisations?

Whether or not to continue your pregnancy following a negative prenatal diagnosis is an extremely personal decision and no one has the right to judge another's chosen course. This pack simply aims to help you gather the necessary information to ensure you understand the implications of whichever path you choose. After hearing your medical practitioner's views, it may be time to consider an alternative perspective. For links to articles and personal accounts, see <http://www.livingwithtrisomy13.org/trisomy-13-termination.htm>.

## **Continuing with your pregnancy following prenatal diagnosis**

You are no doubt aware that there are many more decisions ahead, and that the road will be long and at times very tough. If you are fortunate enough to have found compassionate, open-minded and well-informed physicians, you are off to a great start. Sadly, this is not always the case. Regardless of whether you are considering comfort measures alone or plan on pursuing extensive medical treatment for your child, a thorough knowledge of your options for pregnancy management, labour and delivery, and postnatal care is essential. Pre-armed with the following information you will be well equipped to discuss these issues with your medical team and, if necessary, provide evidence to counter the standard blanket prognoses of 'not compatible with life' and 'inappropriate to treat'.

## **What is Trisomy 13?**

### **Trisomy 13 Fact Sheet**

By Dr John Carey, SOFT Medical Director

<http://www.trisomy.org/trisomy13.php>

### **Definition**

Trisomy 13 (T13) syndrome is a disorder of human chromosomes which occurs in approximately 1 in 10,000 live born infants. T13 is due to the presence of an extra #13 chromosome. Approximately 80% of infants with T13 syndrome will have a full trisomy while the remainder will have a trisomy due to a rearrangement called a translocation or have mosaicism (two different cell lines).

Infants born with T13 have a recognizable pattern of physical features that often allow the health professional to make the diagnosis of the syndrome. Notable physical birth defects and sometimes, anatomic changes of internal organs are present. Findings of significance include small head size (microcephaly); small eyes (microphthalmia) or sometimes absent eye or faulty development of the retina. Cleft lip or cleft palate or both occur in about 60% of children. In addition, there are a number of less medically significant physical findings that are helpful in diagnosis. These include variations of ear shape, changes on the palm of the hand, and extra fingers and toes. Changes in foot development, including changes to the heel, the so-called rocker bottom foot, can occur.

## Heart Defects

About 80% of children with T13 will have a congenital heart defect. These can include:

- ventricular septal defect – 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 – 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 – a defect involving the lack of closure of the channel that usually closes near the time of birth;
- dextrocardia – location of the heart on the right side of the chest; on occasion more medically serious heart defects can occur in T13.

## Medical Problems

The major implications of T13 involve a predisposition to congenital malformations (birth defects) mentioned above, an increased mortality in infancy, and a developmental disability in older children. In addition, older infants can have visual difficulties because of the findings mentioned above and a hearing loss. The increased mortality is related to difficulties with breathing due to either interrupted breathing (apnea), or problems of lung development. In addition, gastroesophageal reflux and feeding problems can occur and predispose to aspiration adding to this risk. Usually the heart defects are not serious enough to be a major health threat in the newborn period.

## Important and Common Birth Defects in T13

- Omphalocele 10%
- Holoprosencephaly 60% (an anatomic defect of the brain involving failure of the forebrain to divide properly)
- Kidney defects 30%
- Skin defects of the scalp 20%

## Common Disorders in infants and young children with T13

- feeding difficulties
- gastroesophageal reflux
- slow post natal growth
- apnea
- seizures
- hypertension
- kidney defects
- developmental disabilities
- scoliosis

**Routine follow-up care of infants with T13**

- Routine child care/anticipatory guidance
- Cardiac evaluation
- Eye evaluation
- Hearing test
- Referral for infant pre-school/ early intervention program
- Ongoing support
- Scoliosis check through childhood
- Routine immunization including chicken pox

**For publications and additional information relating to T13, contact:**

Support Organization for Trisomy 18, 13 and Related Disorders  
2982 South Union St.  
Rochester, NY 14624-1926  
1-800-716-7638

Information from Dr. John Carey, SOFT Medical Director

## T13 survivors

Despite frequent medical advice that T13 is not compatible with life there are many known survivors. The LWT13 Web site currently lists 65 children and adults living with various forms of this condition including Full T13. Specific anomalies and developmental challenges vary, as does the extent of ongoing medical intervention required. The children listed below have been randomly selected from the LWT13 site. Their full stories and the stories of the many others like them can be found at <http://www.livingwithtrisomy13.org>



### **Kristopher, born 13 December 2005**

- Kristopher was born full term weighing 6 lbs 8 oz and measuring 18.75 inches. After being diagnosed prenatally with Dandy Walker Syndrome, postnatal MRI and CT scans confirmed that this was not the case. Kristopher was diagnosed with T13 following a postnatal blood test.
- Kristopher's anomalies at birth included a bilateral cleft lip and palate as well as an extra finger on his left hand. Tests showed he had an insignificant VSD, which did not require any medication or surgery.
- At 9 and a half months old, Kristopher's development was somewhat delayed—he could not roll over all the way and had trouble holding his head up. He was declared legally blind and had failed two hearing tests.
- Kristopher has undergone surgery to repair his cleft lip and is preparing to have his palate repaired. Kristopher is fed only through a peg tube and his most significant challenges to date have been seizures and reflux. He is on medication for the reflux, high blood pressure and seizures.



### **Natalia, born 25 August, 2000**

- Natalia has Full T13 and was diagnosed postnatally.
- Natalia's mum ThereseAnn, also founder of the LWT13 Web site, reports that Natalia is G-tube fed, happy, presently in good health, and able to hum a few of her favorite tunes.

- She is currently attends first grade where she walks with a reverse K-walker. Natalia is able to ride a special adaptive trike and at the age of six, began attempting to walk independently.
- Visually, Natalia is legally blind, has had cataracts removed and uses glasses.
- In ThereseAnn's words she is 'progressing slowly, but always moving forward.' Her mum describes her as a 'joy'.



**Aaliyah, born 18 July, 2003**

- Aaliyah is full T13 and started attending special needs preschool in September 2006. She takes a few steps unsupported and has begun to speak. Aaliyah is able to negotiate obstacles, enjoys music, can hum a number of songs and loves watching 'Elmo' on television.
- Aaliyah's teacher reports she is 'a big help' at school.
- Aaliyah's health is good, though she is prone to catching cold in the winter months.



**Elisha, born 26 August, 2005**

- Elisha was diagnosed with full T13 when her mother was five months pregnant. She was born naturally at 41 weeks and stopped breathing around three to six hours after birth. After being bagged and given oxygen she was revived.
- At birth Elisha's anomalies included a cleft lip and palate, extra digits on each hand and foot, and extended pupils.
- When she was still thriving after her first birthday, Elisha's parents decided to proceed with surgery to correct her cleft lip and palate. Unfortunately the surgery was delayed due to other ailments including pneumonia, roto virus and hydration problems.
- Tests on Elisha's heart found no underdevelopment and confirmed that everything was functioning normally.

- Elisha's parents have arranged for further tests to assess Elisha's sight and hearing.
- Elisha cannot yet sit or walk and her development is assessed at around three to sixth months.



**Amber Lyn, born 17 June, 1997**

- Amber is mosaic T13 but was not diagnosed until just a week before her sixth birthday. At birth she appeared healthy with only some very minor problems including 'a skin tag on her pinky, a reddish purple spot on her forehead, apnea and bad reflux'.
- Amber now attends school and will start the fifth grade later this year and mum Jaylinn reports that she 'couldn't be happier'.
- Problems Amber has encountered include ADHD and a lag in her fine motor skills.
- She currently enjoys good health and is involved in dance, swimming and girl scouts.

## **Further recommended reading**

### ➤ **Trisomy 13 Research**

Baty BJ, Blackburn BL, Carey JC. 'Natural history of trisomy 18 and trisomy 13: Growth, physical assessment, medical histories, survival, and recurrence risk'. American Journal of Medical Genetics 1994; 49(2):175-88.

<http://www.medscape.com/medline/abstract/8116665?prt=true>

Robert G Best, PhD et al. 'Trisomy 13 (Patau Syndrome)'.

<http://www.emedicine.com/ped/topic1745.htm>

Sonja A. Rasmussen, MD, MS, Lee-Yang C. Wong, MS, Quanhe Yang, PhD, Kristin M. May, PhD and J. M. Friedman, MD, PhD. 'Population-Based Analyses of Mortality in Trisomy 13 and Trisomy 18'. Pediatrics Vol 111 No 4 April 2003, 777-784

<http://pediatrics.aappublications.org/cgi/content/abstract/111/4/777>

### ➤ **Resuscitation options in immediate neonatal period**

'International Guidelines for Neonatal Resuscitation: An Excerpt From the Guidelines 2000 for Cardiopulmonary Resuscitation and Emergency Cardiovascular Care: International

Consensus on Science'. Pediatrics Vol 106 No 3 September 2000, e29.  
<http://pediatrics.aappublications.org/cgi/content/full/106/3/e29>

## Other resources

### Web Sites

- Be Not Afraid – <http://www.BeNotAfraid.net>
- Emmanuel's Foundation – <http://www.emmanuelfoundation.org>
- Carrying to Term Pages – <http://www.geocities.com/tabris02/>
- Hope for Trisomy 13 and 18 – <http://www.hopefortrisomy13and18.org>
- Michaels Feat – <http://www.michaelsfeat.org/resource.htm>
- Living With Trisomy 13 – <http://www.livingwithtrisomy13.org>
- Prenatal Partners for Life – <http://www.prenatalpartnersforlife.org>
- Support Organisation for Trisomy 18, 3 and Related Disorders (S.O.F.T) – <http://www.trisomy.org>

### Articles

- Chitty et al. 'For Debate: Continuing with pregnancy after a diagnosis of lethal abnormality: experience of five couples and recommendations for management'. BMJ 1996;313:478-480 (24 August)  
<http://www.bmj.com/cgi/content/full/313/7055/478>
- Louise Locock et al. 'The parents journey: continuing a pregnancy after a diagnosis of Patau's syndrome'. BMJ 2005;331:1186-1189 (19 November)  
<http://www.bmj.com/cgi/content/full/331/7526/1186>

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<sup>1</sup> 'What are ultrasound markers?', 2006, viewed March 2007,  
 <<http://www.babycentre.co.uk/pregnancy/antenatalhealth/scans/ultrasoundmarkers/>>