



Trisomy 13

A Handbook for Families



Support Organization for Trisomy 18,
13 and Related Disorders

Trisomy 13: A Guidebook for Families

Carol M. Stenson, Ph.D. Professor of Special Education, Idaho State University, Pocatello

Steven E. Daley, Ph.D.
Professor of Special Education, Idaho State University, Pocatello

Patricia A. Farmer, Ed.S. Regional Special Education Consultant, Idaho State Department of Education, and
Instructor of Special Education,
Idaho State University

Copyright © 1999, 2013

Support Organization for Trisomy (SOFT) 18, 13 and Related Disorders and the Munroe-Meyer Institute, University
of Nebraska Medical Center

Second Revised Edition All rights reserved.
ISBN 1-889843-30-X

Published by the Media Center, Munroe-Meyer Institute,

A University Affiliated Program, University of Nebraska Medical Center, 985450 Nebraska Medical Center, Omaha,
Nebraska 68198-5450.

**NOTE: This is an early version of the publication and the photos
have not yet been inserted.**

This book is dedicated to
Joey Watson
September 28, 1977 - January 7, 1984
Trisomy 13

Contents

Acknowledgments	7
Trisomy 13	9
What does a diagnosis of trisomy 13 mean for you and your child?	9
How do you know your child has trisomy 13?	17
Chromosomes and karyotypes	22
Are there educational opportunities for your child?	34
What else do you need to know?	45
Resources	48
Glossary of Terms	50

Acknowledgments

The authors wish to acknowledge John C. Carey, M.D., M.P.H., Associate Professor, University of Utah Medical Center, Salt Lake City, for his comments on the technical aspects of this book and for providing the karyotypes illustrating trisomy 13.

Trey and Diane Lackey and Pat O'Toole provided many of the photographs. We appreciate their support and encouragement of this project. Many of the quotations of parents used throughout this book are from letters published in *The Soft Touch*, a newsletter published by the Support Organization for Trisomy 18, 13 and Related Disorders.

Our thanks to these parents and to the many others who have offered their support, photographs and suggestions. Thanks also to Bruce Buehler, M.D., Rebecca Anderson and the Media Center of the Munroe-Meyer Institute, University of Nebraska Medical Center, Omaha, for their guidance in the preparation of this guidebook.

Finally, we hope that we have been of some assistance to the parents of children with trisomy 13 during what may be the most challenging days of their lives.

Cara Ann Carpenito May 29, 1986
Trisomy 13

Lauren Paige Hari January 24, 1988
Trisomy 13, Mosaic 6%

Michael John Giunta
March 9, 1987 - July 7, 1987
Trisomy 13
Trisomy 13

(tri so-mi thur-teen) [Fr. tri-, three, + G. soma, body from (chro- mo)some, + number 13]. 1) Having three copies of chromosome-13 instead of the normal two, resulting in 47 chromosomes instead of 46. 2) The clinical condition resulting from this chro- mosomal imbalance, i.e., low birth weight, distinctive physical appearance, variably severe internal organ malformations and profound developmental delay.

When your physician first tells you that your baby may have trisomy 13 (Patau syndrome) or that this diagnosis is suspected, you will be overwhelmed with questions. The first question usually is “What is trisomy 13?” This book was written to answer some of these questions, to share insights of parents of children with trisomy 13 and to give hope for the best possible outcome for your child.

What does a diagnosis of trisomy 13 mean for you and your child?

When your physician diagnoses trisomy 13, parents usually ask, “How long will my baby live?” The medi- cal complications associated with this condition make it likely the child may not survive infancy, and parents are routinely told this. But, children are not statistics. They continue to surprise and confound the medical community. Data collected by support groups such as SOFT (Support Organization for Trisomy 18, 13 and Related Disorders) indicate there are more first year survivors than have been reported in the medical literature. Your baby with trisomy 13 has already demonstrated a strong will to survive by just being born! In the absence of any immedi- ate life-threatening condition, accurate predictions of life expectancy are very difficult to make.

*I heard about how a woman felt after giving birth to a baby with Down syndrome. She said it had been like pre-
paring for a flight to Italy and landing in Holland ~ Holland not being such a bad place after all. Well, I felt like my
plane had been hijacked by terrorists, and we weren't going to land in a bed of tulips.*

There is a very good chance that your child will have to spend some time in the neonatal intensive care unit or NICU. This can be a strange and scary experience for parents who do not know what to expect. There is strange looking equipment that makes even stranger noises. Al- though this equipment looks forbidding and foreboding, it may actually provide a life-line for your baby during the first few days.

You might see . . .

- Open beds with overhead heaters designed to keep your baby's temperature regulated. A heat sensor at-

tached to your baby's skin tells the system when to cool or heat in response to changes in room temperature.

- Incubators, which are enclosed, see-through beds with openings on the side for tending the baby. This machine is also designed to help maintain your baby's body temperature.
- "Bili lights" over an open bed to correct jaundice, or hyperbilirubinemia, common to premature babies.
- Continuous positive airway pressure, or CPAP, a system in which air is forced into the baby's nose through tubes to assist in breathing.
- Respirators which actually take over and breathe for the baby when the baby can't breathe independently. One tube is placed into the baby's mouth with the other attached to the machine to pump oxygen according to the baby's needs.
- Heart and breathing monitors with buzzers to alert NICU nurses when the baby is having a breathing problem.

Since trisomy 13 is a relatively uncommon condition, many physicians may not be familiar with the syndrome and rely on descriptions from their medical books. These descriptions are not terribly positive. Textbooks give facts and figures, but seldom deal with the real issues facing families. While it is unfair to give false hope, not all children die in the first year of life. In fact, 10 percent survive. Again, so much is dependent on the degree of involvement of your baby that an accurate prognosis cannot be made in a medical text.

We encourage you to discuss your emotions as you face decisions concerning your child's care. You are not alone. There are parent organizations and support groups such as SOFT to help you. The goal of SOFT is to provide you with family contacts and with information when difficult decisions must be made (see page 48).

Sarah Hansen June 6, 1986
Trisomy 13

After being told there is a strong possibility that your child will not survive until the first birthday, you are then faced with the question of whether or not to bring your baby home. Many hospitals encourage parents to take their baby home, to spend quality time with their child. Others do not. Ultimately, the final decision rests with each family. In any case, it will probably be necessary for your baby to remain in the hospital until the essential and appropriate care is given and your baby's condition is stable.

We decided to bring Joey home with us for whatever that would mean. It was also our decision not to take heroic measures to prolong his life. His comfort would be our first priority, but it was our feeling that excessive medical intervention would only make what time he might have miserable.

When it is time to take your baby home, the nurses in the neonatal intensive care unit will work carefully with you to insure that you are able to care for your child. You may need special equipment to monitor the baby's heartbeat and breathing. If a cleft palate makes nipple feeding difficult or impossible, you may be trained in how to feed the baby using a tube, a technique known as gavage feeding. You and your physician may decide to feed the baby through a tube inserted directly into an opening in the stomach, called a gastrostomy.

We were scared to death each time Kelsey quit breathing during apneic spells, but never regretted bringing her home. We have had five wonderful years, and I hope we are given several more. If our time with her ended soon, I would not be bitter. Taking her home was not a decision for us, and she continues to amaze us all.

If your baby has a cleft palate or needs to be fed by tube, feeding may be a challenge. There are a number of devices which can be used to assist your child. One option might be the "Haberman Feeder" for babies who have sucking difficulties. The nipple remains free of air while allowing airflow into the bottle and responding to tongue action rather than a strong sucking reflex.

The feeder insures that each sucking action produces a strong flow of milk, but is designed so the baby is not overwhelmed with milk. If the baby cannot suck, the parent can control the flow of milk. The Haberman Feeder, developed in Great Britain by a mother whose baby had feeding difficulties, is available from Medela, Inc., P.O. Box 660, McHenry, Illinois 60051-0660, toll-free telephone 1-800-435-8316.

Whatever your baby's medical condition, insecurities about feeding are a major cause of parental anxiety. Be sure you have sufficient support from a specialist until you are comfortable with this aspect of your child's care. Chronic constipation is a problem with many babies with trisomy 13. Stool softeners are often prescribed as additives to the baby's food, and suppositories may also be indicated. Colic and G.E. reflux, or rising of food from the stomach to the esophagus can also be treated.

Perhaps the most frightening symptom for parents is apnea. Apnea is breathlessness or breath holding. It is most common in infancy, although some children continue to demonstrate apnea throughout their lives. Apneic spells vary in duration. For some children, slight stimulation is sufficient to induce breathing. For others, CPR (cardio-pulmonary resuscitation) may be required to restore breathing. Depending on the severity of this problem with your baby, you may choose to have apnea monitoring equipment in your home. There is no question, however, that all of the baby's primary care givers should be well-trained in CPR techniques.

When Joey was a baby, I didn't know CPR. I would become so frustrated when he stopped breathing that I would hold him up, shake him, look him right in the eye and scream, 'breathe, dammit, breathe!' Thank goodness it worked. I didn't know at the time that I was stimulating him.

Celeste Heinein
January 12, 1986 - July 17, 1986
Trisomy 13

Children with trisomy 13 have increased susceptibility to upper respiratory infections and pneumonia. This tendency can be particularly critical in infancy. In some cases, the baby may become so ill that oxygen, suctioning, antibiotics and even hospitalization are required.

Seizures are of major concern in children with trisomy 13. Many display some level of seizure activity ranging from mild absence or petit mal seizures to full tonic-clonic or grand mal seizures. These can be particularly troublesome if breathing stops during the seizure and the child has to be resuscitated. Often seizure activity can be controlled with medication. Be sure to let your pediatrician know if your child experiences breath-holding spells, blackouts, unusual repetitive motions, muscle jerks or full-blown seizures.

How do you know your child has trisomy 13?

Although most children resemble their parents, there are certain significant features of a child with trisomy 13 which will alert an informed physician or pediatrician immediately. Some of these characteristics will not affect the baby's life, but simply aid in the complex task of diagnosis. Some children will have potentially life-threatening complications. It is important to understand that the description of a syndrome includes all of the possible related features, but that few children with trisomy 13 have all of the clinical symptoms.

Infants with trisomy 13 can have low birth weight, even when the pregnancy is full-term. A moderately small head (microcephaly) with sloping forehead is often diagnosed at birth. Major structural defects of the brain may also be diagnosed shortly after birth. Often the front of the brain does not divide properly and a condition known as holoprosencephaly results. This defect of the central nervous system is also associated with alterations of facial development resulting in narrowly placed eyes and, in some cases, underdevelopment of the nose or nostrils.

Structural eye defects are often noted in babies with trisomy 13. These include microphthalmia, or an unusually small eye, and a keyhole defect of the iris tissue called coloboma. Faulty development of the retina (retinal dysplasia) occurs frequently. The bony ridges above the eyes are shallow and the eye openings (palpebral fissures) are usually slanted upward. Glaucoma or pressure in the eye also occurs sometimes.

Cleft lip, cleft palate or both are present in over half of all cases. The ears are abnormally shaped and sometimes low-set.

Some babies with trisomy 13 have birthmarks made up of tiny blood vessels close to the skin surface (capillary hemangiomas), especially on the forehead in the midline.

Scalp abnormalities (cutis aplasia) resembling ulcers may be present on the back of the head.

Cathy Jo was born with an extra finger, a unilateral cleft- lip and palate, large eyes, glaucoma, a scalp ulcer, numerous hemangiomas and, quite frankly, after this long I can't remember, nor care to remember, what else.

Unusual palm print patterns are often present and aid in initial diagnosis. These include a single diagonal crease on the palm, a sign of low muscle tone. Many trisomy 13 babies have polydactyly, extra fingers and toes. Many have deeply bowed, narrow fingernails. The fingers tend to be flexed in an over-the-thumb pattern.

Jasmine Woody April 7, 1982
Trisomy 13

All of Susie's cousins were jealous because they didn't have six fingers. They would come up to Susie and say, 'Give me six.' She kept her extra fingers for three years, and when we finally had them removed, I cried.

Tyheem Derek Frazier November 11, 1987 - February 15, 1990
Trisomy 13

In approximately 80 percent of cases, there are other congenital problems. These include ventricular septal defect, an opening between the two lower chambers of the heart, and atrial septal defect, an opening between the two upper chambers of the heart. Both of these malformations are sometimes associated with heart murmur and can result in disruption of the pumping action. Another common heart defect is patent ductus arteriosus. In this condition, normal closure of a duct fails to occur, and blood flow persists through a channel which usually closes soon after birth. Dextrocardia, a right shift in location or a mirror image type heart, sometimes occurs in the trisomy 13 syndrome. In addition to these problems, there may be other conditions which require surgical intervention. These include omphalocele, in which abdominal organs protrude into the umbilical cord, and kidney malformations.

Occasionally, failure of the testes to descend into the scrotum (cryptorchidism) and an abnormally developed scrotum are noted in males. Females may display bicornuate or horn-shaped branches on the uterus. These are not life-threatening conditions.

Your child will have some of the identified characteristics, but probably not all of them. Each baby must be evaluated individually and appropriate interventions should be identified by your medical team. If you do not understand what the team is telling you, ask them to tell you again. Physicians frequently assume that parents understand medical terms when they ask questions. If this is not true in your case, be sure your medical team understands that you have many questions and that you need answers in terms you can understand.

Alan Hoffman, Jr. January 14, 1985
Trisomy 13, partial

A specialist from Children's Hospital confirmed the diagnosis of trisomy 13. The next week was a blur. So many decisions to make on what to do and so many emotions— love, sadness, anger.

Chromosomes and karyotypes

Chromosomes are tiny, threadlike structures present in every cell of the body. Under the microscope, chromosomes can be distinguished by their length, their banding pattern and the location of a constriction site (centromere) which separates the short arm ("p") from the long arm ("q").

Each chromosome contains part of the genetic material inherited from parents which directs bodily development and gives each person a unique set of physical characteristics.

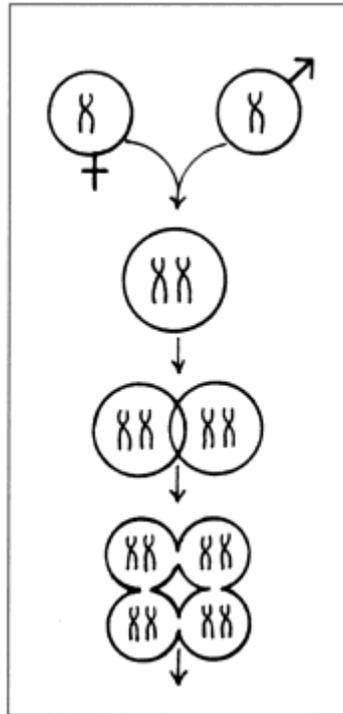
A karyotype is a "photograph" of a set of chromosomes arranged in a standardized order. While physicians can make very good predictions about the nature of a genetic disorder based on external or clinical characteristics, the diagnosis of trisomy 13 cannot be confirmed until the child's genetic material is "mapped" in a karyotype. The average human cell contains 22 pairs of autosomes (non-sex chromosomes) plus two X chromosomes if the child is a female and an X and a Y chromosome if the child is a male. When an egg and sperm are joined, each brings 23 chromosomes to the resultant new cell which then contains 46 chromosomes. The fertilized egg divides repeatedly as the pregnancy progresses, and each division results in a cell that is an exact duplicate of the initial cell (see Figure 1).

Rachel Kay Maurer
April 29, 1982
Trisomy 13
Mosaic 32%

Billy Vaughan September 15, 1986 -
December 29, 1989
Trisomy 13

Figure 1

An illustration of normal union and subsequent cell division of a sperm and egg. Only one pair of chromosomes is shown for simplicity.



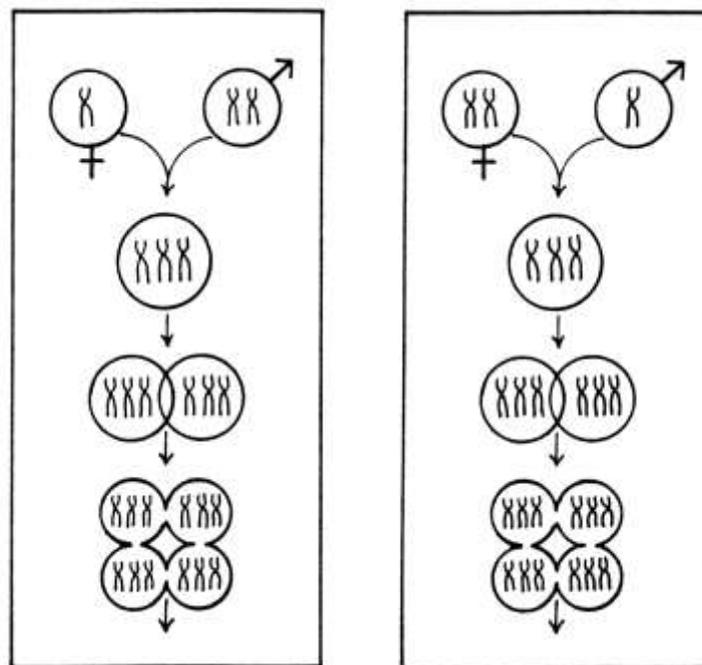
Sometimes, for reasons which are unclear, abnormalities in the number or the structure of chromosomes may occur. Trisomy 13 is usually an abnormality of chromosome number — a complete extra 13th chromosome is present in every cell. (The condition is also known as Patau syndrome. Dr. K. Patau was the first to link the symptoms to the presence of this extra chromosome.) Individuals who have some of the less common forms of trisomy 13 may represent abnormalities of structure as do all of the disorders that are called partial trisomy 13.

The word “trisomy” means that three copies of a particular chromosome are found in the cell instead of two. Most often, this error in chromosome number occurs at fertilization, and the blueprint for development is altered from the moment of conception. Usually this is the result of faulty chromosome division in the formation of the egg or the sperm.

The 13th pair of chromosomes fails to separate, leaving one gamete (egg or sperm) with 24 chromosomes and the other with 22. On fertilization, this will either result in too many or too few chromosomes. A fetus with a single copy of chromosome 13 will be miscarried; a fetus with three copies is at substantially higher risk for miscarriage but may be born alive. Figure 2 shows how a “meiotic nondisjunction” (error of chromosome separation in an egg or sperm) results in excess material in the developing embryo.

Nondisjunctional errors can happen with any pair of chromosomes. Most result in miscarriage, although live birth is possible with a few, including trisomy 18 and trisomy 21. Though the cause has not been established, these errors are known to occur much more frequently as mothers get older. Even though the chance of having a baby with a trisomy increases with age, it is important to note that these disorders can occur at any maternal age. Most babies with trisomies are born to women who are in their twenties, the most frequent childbearing years. About 50 percent of all pregnancies end within days of conception, and one in five confirmed pregnancies terminates spontaneously before 12 weeks gestation. Many of these natural terminations of pregnancy (miscarriages) are due to monosomies (only one copy of a chromosome) or trisomies, including trisomy 13.

Figure 2: Meiotic Nondisjunction



Full trisomy 13, with an extra, structurally normal chromosome 13 in every cell, is found in about 80 percent of the babies with Patau Syndrome who are karyotyped.

Nearly 20 percent of children with Patau syndrome have a partial trisomy of the 13th chromosome rather than complete trisomy. An extra piece of the 13th chromosome may be attached to another chromosome (translocation), or an extra segment may be found within the 13th chromosome itself (duplication). Translocations may involve any pair of chromosomes, and only cause developmental problems when they are unbalanced, that is, when extra or missing material results. The most common unbalanced translocations

Rebecca Lackey March 9, 1986

Trisomy 13 Danny Brown

August 13, 1985

Trisomy 13

found in Patau syndrome involve an entire extra 13 attached either to chromosome 14 or to another chromosome 13 (see Figures 3-6). The location of the chromosomal material has no effect on the child's symptoms.

In some children, an unbalanced translocation or duplicated segment involves a smaller portion of the chromosome, sometimes known as a partial trisomy. In these situations, the pattern of physical features can be different from full trisomy 13, depending on which portion of the 13th chromosome is involved. Your geneticist, genetic counselor or physician may be able to clarify the relationship between the partial trisomy, the syndrome features and prognosis.

Parents of a child with an unbalanced translocation should have chromosome studies on their own blood to determine whether one of them carries a balanced translocation or other chromosomal rearrangement. If so, the chance for another pregnancy with trisomy 13 is much greater than it would otherwise be, and unaffected siblings may also carry the balanced translocation.

Trisomy 13 mosaicism is a rarer form of the syndrome. "Mosaic" denotes the presence of two types of cells in the same individual, e.g., some with 46 chromosomes and some with 47. Two possible causes are suspected. In rare cases, the correct number of chromosomes may have been present at conception, but during the first few rounds of cell division one pair of chromosomes failed to separate properly. One of the daughter cells from that "mitotic non-disjunction" would contain a single copy of chromosome 13 and die. The other would contain three copies of chromosome 13 and survive, giving the developing embryo a normal cell line with 46 chromosomes and a second cell line with trisomy 13. More commonly, an embryo might begin with full trisomy 13 and "lose" one of the extra 13th chromosomes during early cell division. Some individuals with trisomy 13 mosaicism have the full spectrum of symptoms, while others have milder or fewer symptoms.

Errors in chromosome division are common and are outside of our control. There is no scientific evidence to suggest that exposure to environmental dangers or X-rays, living an unhealthy life-style, drinking, smoking, drug use, stress or poor parental health can cause trisomy 13. It is important to remember that nothing either parent did or failed to do could have caused or prevented the presence of the extra chromosome 13.

I spent weeks after my son was born wracking my brain trying to think what I had done that caused this. I finally narrowed it down to the fact that I had eaten red gelatin with mandarin oranges. In my mind at the time, that was the only thing that could possibly have caused it. It was then that I realized I was near a breakdown.

Figure 3: G-banded (standard staining technique) karyotype showing normal male chromosomes, i.e., 46, XY.



Figure 4: This karyotype shows the chromosomes of a boy with trisomy 13, i.e., 47, XY, +13,

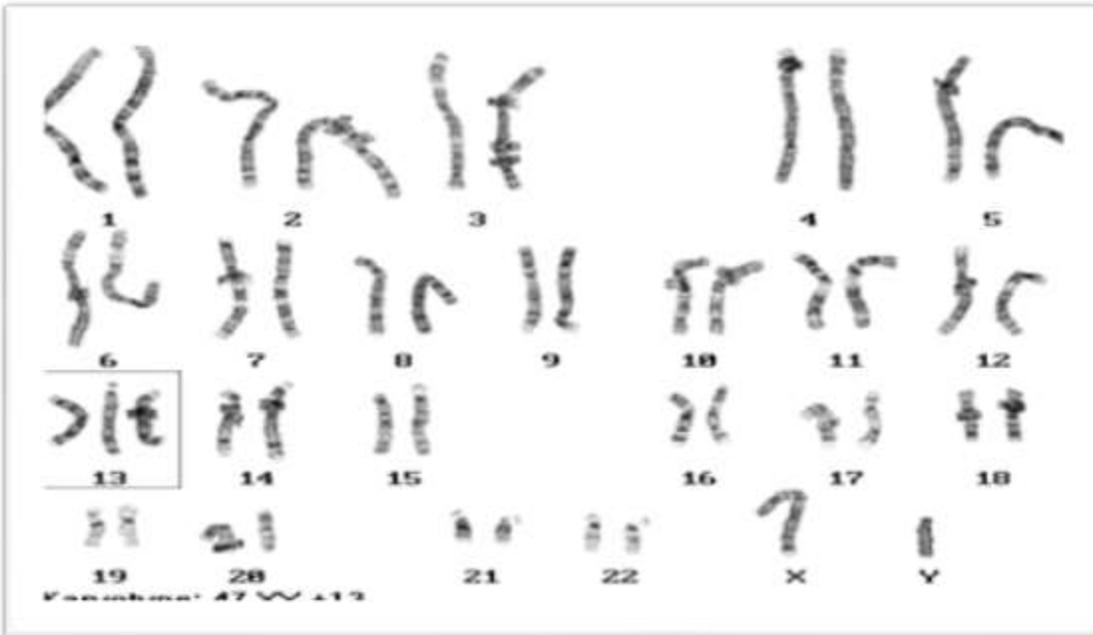


Figure 5: This karyotype shows an unbalanced 13/14 translocation. Note the attachment of a chromosome 13 onto a chromosome 14, producing a virtually complete trisomy 13.

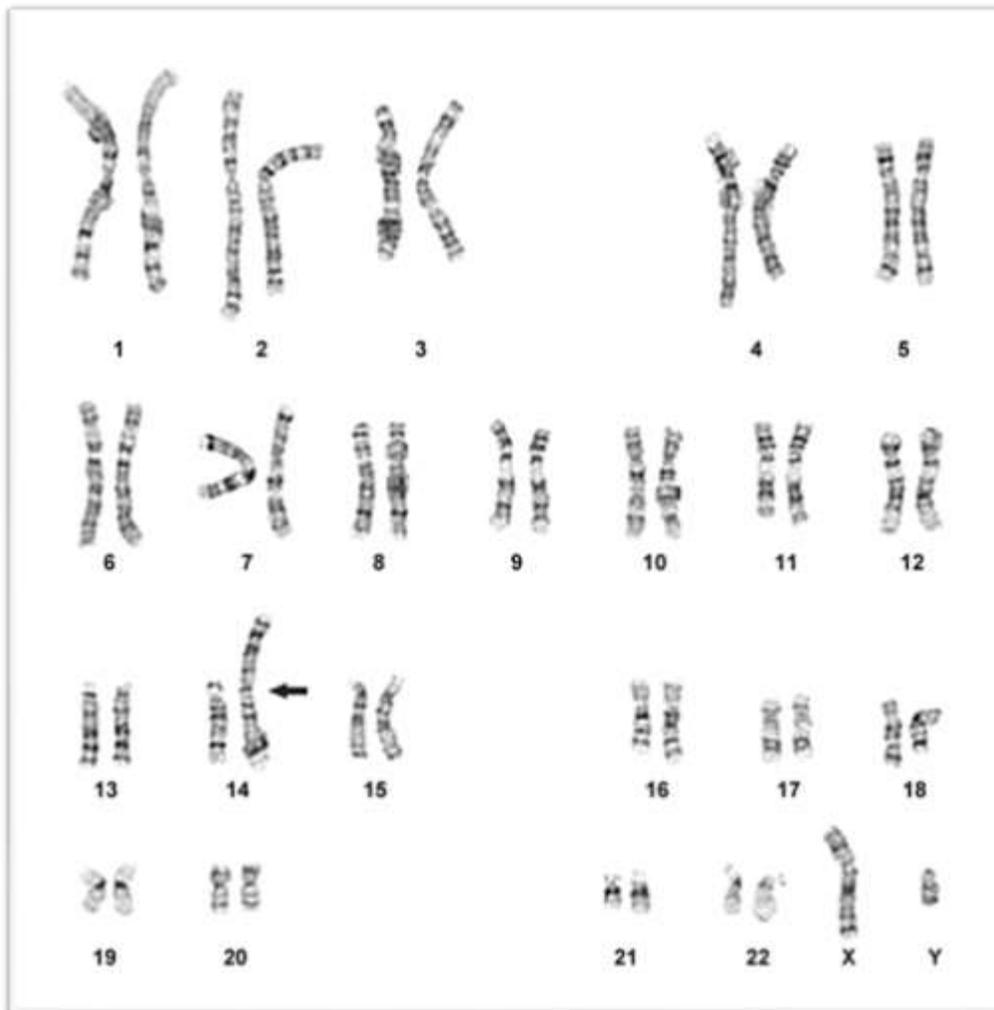


Figure 6: This study represents the karyotype of the mother of the child in Figure 5. Note the balanced translocation involving the 13 and 14; ie.. 45, XX, -13, -14, + t(13q; 14q).



Prenatal diagnosis either through amniocentesis or the newer test called chorionic villus sampling is an option for parents who have had a child with any form of trisomy 13. In future pregnancies, families can consult their physician or genetic counselor about the details of this option.

Trisomy 13 is found in only 1 in 7,500-10,000 babies each year. In the United States, there are about 400 liveborn babies per year with trisomy 13. However, trisomy 13 is a common chromosomal cause of fetal death, miscarriage and stillbirth.

All of this information may seem somewhat bewildering to parents facing the crisis of giving birth to a baby with trisomy 13. After reading this material, you probably will have many questions. It may be necessary to have the information explained several times in order to understand it completely. Be persistent! Let your physician know what you do and do not understand about your child's genetic makeup and medical conditions. If you do not understand the answers to your questions, ask them again!

Are there educational opportunities for your child? With more and more babies surviving the first years, we are seeing the need for early and continuing special education services. This section of the guidebook will assist you in understanding your rights as parents of a young child with disabilities and introduce you to some of the special education and related services that may be available for you, your child and your family.

Two pieces of federal legislation form the framework for providing the special education services needed by children with trisomy 13. Public Law 94-142, passed by Congress in 1975, requires that special education services be made available to all school-age children with disabilities.

Public Law 99-457, passed in 1986, extends this mandate downward to age three years, and, in addition, encourages states to make early intervention services available to children from birth through three years of age and their families. The Individuals with Disabilities Act (IDEA) of 1990 and the IDEA Amendments of 1997 replaced

PL94- 457, placing an emphasis on inclusive services for young children with disabilities. The diagnosis of trisomy 13 makes your child eligible for free and appropriate early intervention services if your state is one that provides services from birth to age three.

Joseph M. Watson
September 28, 1977 - January 7, 1984
Trisomy 13

Joey signing the word “more” for his mother.

We enrolled her into an infant stimulation program at the age of nine months. At that time, she was still being fed with the NG tube, she had no head control and could not put any weight on her legs. She began to walk at the age of four and threw the Pampers away at five. She runs, climbs, swings, dances and sings joyful noises that we love to hear.

Kelsey Lans August 18, 1985

Trisomy 13

These federal laws acknowledge that all children can profit from educational intervention and that no child shall be found ineligible because of the severity of the disability. These laws also provide specific regulations regarding parents' rights in all aspects of the educational process and provide an appeals process for parents if they disagree with the professionals. These rights pertain to parent input in assessment, program planning, determination of eligibility and placement decisions.

Every school district must make special education services available to all children with disabilities from three through 21 years of age. Early intervention services may be available in your state for children with disabilities from birth to three years of age. Many new parents do not understand what is meant when educational professionals discuss early intervention. Early intervention simply means that rather than waiting until your child is school-aged, the educational system realizes that interaction and teaching at a much earlier age offers tremendous benefits to both parents and children. Early intervention services can start before the child leaves the hospital nursery.

An example of an early intervention service that could be provided in the nursery is an occupational therapist working with you to show you how to hold and feed your baby. Early intervention services in the first two years often include home visits by specialists to work with you and your baby. The focus of these interventions is determined jointly by you and the early intervention specialist.

Depending upon your baby's needs, several different specialists such as a physical therapist, speech pathologist and teacher might work with you. The main purpose of early intervention is to provide you and your family with services that help your baby to develop basic abilities.

I had the feeling that they thought Bobby was going to die, and it didn't make any sense to do infant stimulation. I got so tired of people digging his grave. I chose to work with him on flexing his fingers, sitting, crawling and eventually walking. Services were not good when Bobby was born. Hopefully, they are better now.

There is strong evidence that early intervention increases social skills and the child's integration into the family setting. Services for infants and toddlers are usually provided in your home, although some specialized services may be available only in center-based settings. Services for preschool age children are usually center-based, including regular, community-based preschool and child care settings near your home.

The important thing to remember is that special education services are available for all children from ages three through 21, and that infant intervention programs may be available in your state. Early intervention services and related family support services in general are provided at no cost to the parent, and there are no income eligibility requirements.

Every state has a designated public agency responsible for providing early intervention services. The lead agency in your state may be your state department of education, health or social services. If you do not know which agency to contact, check with your pediatrician, hospital, local health department or the director of special education in your local school district.

Early intervention services usually include a complete developmental assessment and determination of your child's eligibility for services. Remember, the diagnosis of trisomy 13 automatically qualifies your child for the services provided in your state. You will be invited to meet with the specialists who will provide findings from their developmental evaluation of your child. You have the right to be a participating member of this team. You will be asked to provide your opinion about your child's immediate and long-term needs for learning and development.

Michael Litz September 29, 1985
Trisomy 13, Translocation

If you disagree with the results of the initial assessments or with the identified needs of your child, make your opinions known. Developing an Individual Family Service Plan, or IFSP (ages birth to three), or an Individual Educational Plan, or IEP (ages three to 21), can be overwhelming for parents. While the "experts" will tell

you what your child needs, remember you are the ultimate expert. Make your feelings known.

As a parent of a child with trisomy 13, you will work closely with agencies providing early intervention and special education services. All parents need to know their opinions are as important as those of the professionals.

Parents' rights are very clearly spelled out by the law. If you are uncertain about what the professionals are telling you about your child, ask them again. If you disagree with their decisions, persist in explaining your concerns.

If you cannot reach agreement with the professionals about your child's educational needs, you have specific rights under the law. In every state there is an agency responsible for providing free professional and legal support, counseling and advocacy services for parents of children with disabilities. Often these agencies are staffed by parents of children with disabilities.

In states that provide early intervention for children with disabilities from birth to three, a variety of services related to the special needs of your child could be offered. This could mean the development of an Individual

Family Service Plan (IFSP) for your family and your child. Managing the needs of a child with trisomy 13 often involves ongoing contact with several physicians and others in the hospital, consultation with specialists, diagnostic work-ups, and dealing with a variety of medical, surgical and health related issues.

In addition, you may be working with your insurance company, Social Security, and other public and private service agencies. Service coordination, also provided at no cost to you, is a part of early intervention services. You also may request services for your family as a part of early intervention for your child. These services may include counseling, respite care or additional information about trisomy 13 for family members.

What is your role in early intervention? Some parents like to be very involved in teaching new skills to their child and work closely with the early intervention specialists. Some parents would rather spend their time involved in whole family activities and prefer that the specialists teach their children. The “most appropriate” role in early intervention for you is the role you feel most comfortable assuming.

Services could include working on your child’s social skills, particularly in teaching your child how to interact with others. Depending on your child’s specific needs, there also could be programs aimed at the development of language and physical, cognitive and self-help skills. Additionally, early intervention services may be provided to help you understand your child’s strengths, weaknesses and developmental needs.

Getting a child qualified for special education is a reversal of positive thinking, because you must then point out all the things that she can’t do in order to get her all the help she needs.

When your child reaches the age of three, your child is entitled to a free, appropriate public education under the Individuals with Disabilities Education Act (IDEA). This includes, but is not limited to, educational services, speech pathology, physical therapy, transportation, occupational therapy and medical services (including vision and hearing) as needed to allow your child to attend school. All of these services are at no cost to parents.

Each child eligible for special education services has a written Individual Education Plan (IEP) specifying the services, placement, evaluation, and programming designed to help your child reach his/her potential. A multi-disciplinary team composed of a school district representative, the child’s teacher, members of the diagnostic team, and parents meets to establish goals and write the IEP.

I remember the first day the big yellow school bus came. I thought, ‘I really have a normal child.’ I wanted to cry and jump into my car and follow the bus to school. Seven years later, I still remember the number on that bus and get a lump in my throat when I see it around town. It’s funny what you remember.

It is important to understand that these are educational **rights**. Every school district in the country is required to provide services to children with disabilities regardless of available resources. Lack of funds or personnel is no excuse to withhold special education services.

Each state has a director of special education housed in the state department of education. If you have questions or concerns that cannot be addressed at the school district level, you may pursue them with the state department of education. If you still do not receive satisfaction, you may institute a due process procedure. In any case of dispute, be sure to communicate with other parents who may have gone through similar situations.

Faculty in the special education department at your nearest college or university are also available to give you information about your rights.

What else do you need to know?

When you became pregnant, you assumed you would have a “normal” child. This did not happen. Now, you may feel a desperate need to know what is “normal” for your child. The scientific literature will not necessarily give you the information you seek. The area of pediatric medicine grows and changes dramatically every day. It is impossible to project today what medical science will know five years from now. But there are some things that we do know which might help you come to understand your baby better.

There is a misconception that your child will not know you and will be a mere “survivor” devoid of personality. Those of us who have known a child with trisomy 13 know this is not true at all! Even though most children with trisomy 13 have severe to profound developmental disabilities, each has an individual personality.

With her curly, red hair and long, red eyelashes, I know she has a ‘blue-eyed soul.’ Siena has taught me that life is meant to be lived. She taught us about unconditional love. She has grown in our hearts and has touched many other lives as well.

Parents often report constant apprehension and fear about the potential death of a child with trisomy 13. Though this possibility is ever present, at least one child with trisomy 13 survived to the age of 21. The length of time you will have with your child will be in large part dependent on the number of life-threatening conditions your child experiences.

Each individual accepts the child with trisomy 13 in his or her own way. There is no denying that the care-giving demands of a child with disabilities place additional stress on the marriage, the family unit and the siblings.

Her brother is now 12 years old. While he has accepted her completely and loves her dearly, adolescence is taking its toll on his tolerance. He is beginning to understand what he misses because ‘we can’t take Cathy Jo.’ He worries about how he will take care of her when his mom and dad die.

There will be times when parents need to be alone together. Finding a baby-sitter who can cope with the special needs of a child with trisomy 13 is often a difficult task. However, most communities provide respite care through health and welfare agencies. Cost, length of stay and availability will vary widely from one area to another. You can probably get information on the respite care available through your health care team or local hospital.

Because of the serious nature of the physical disabilities that accompany trisomy 13, parents are often asked to participate in life or death decisions for the baby. Do you use heroic means to prolong life? Do you withhold drugs and therapies that might prolong life? Do you agree to have major surgery involving a general anesthetic, even though the potential complications of surgery and anesthesia might threaten life? These are all questions parents may face. The most important thing to remember is that no matter what decisions you and your medical team reach, you have done the best you can in the situation. There are no universally right or wrong answers to these questions.

The decision not to give intensive care to prolong the life of a very ill baby does not mean that loving care is withdrawn.

Justin, Lisa, Kyle, Megan the Clown, Heather, Trevor and Cal Perry
Heather April 23, 1981 Trisomy 13

During Rick's short life, many decisions had to be made. Do we put casts on his club feet? Do we give medication for seizures? Do we tube feed through the mouth or nose? Do we keep the tubes in or remove them after each feeding? I began to feel alone in the care of my son.

The pain of grief is hard to endure, but it is important that everyone in the family go through the grief process in his or her own way. For parents who give birth to babies with severe disabilities, the acute grief process may need to be completed twice — once when you grieve for the “normal” child you expected but did not have, and again if your baby dies. During the time between birth and death, many families experience a gentle grieving condition known as “chronic sorrow.” The grief process following the death of a child will probably be difficult for the other children in the family to understand and accept. We would all like to hold on forever, but there comes a time when we must let go.

Kenisha was a special child who comes along to a chosen few. With this I close, but never let our hearts and minds be closed to the emotions shared by our comrades, as we are very special children. Thanks again for the very special support by SOFT and the families. God bless us all.

Jared, Rebecca, and Erin Handel February 22, 1978
Trisomy 13

Resources

There are a variety of organizations created to assist parents and families. One group especially formed for parents of children with trisomy is **SOFT (Support Organization for Trisomy 18, 13 and Related Disorders)**. SOFT, a network of families and professionals, produces a quarterly national e-newsletter containing letters from parents, information on emerging medical trends, and articles by professionals regarding the special care required by children with trisomy.

Annually, SOFT sponsors an international conference dealing with many topics of interest and concern to parents, including sessions for families grieving the loss of a child. While there is no charge for SOFT materials or membership, individuals are encouraged to make contributions to SOFT when that is financially possible. Information for new families is available on the SOFT website, www.trisomy.org and the headquarters can be contacted at:

Barb Van Herreweghe
2982 South Union Street Rochester, New York 14624
(716)594-4621 Toll-free: (800) 716-7638
[Web Site: www.trisomy.org](http://www.trisomy.org)

The **National Center for Education in Maternal and Child Health** can direct you to agencies for educational, financial, medical and psychological help and identify the nearest genetic counseling center.
National Center for Education in Maternal and Child Health
Georgetown University 2000 N. 15 Street, Ste. 701
Arlington, VA 22201-2617
(703)524-7802

The **National Organization for Rare Disorders (NORD)** will refer you to parent and support groups as well as provide a newsletter and scientific articles on specific uncommon diseases.

National Organization for Rare Disorders
P.O. Box 8923
New Fairfield, CT 06812 (203)746-6518
Toll-free: (800)999-6673

Glossary of Terms

Words used in conjunction with trisomy 13.

amniocentesis - removal of a small amount of the fluid surrounding the fetus in the uterus, usually for genetic testing or prenatal assessment.

anomaly - a variance from the normal.

apnea - periods of interrupted breathing or breath holding.

ASD (atrial septal defect) - a hole between the two upper chambers of the heart which makes it difficult for the heart to pump sufficient oxygen-rich blood to the body's tissues; a heart murmur can often be heard.

auditory brainstem-evoked response - a test to determine if the hearing system can transmit information about sound to the brain. Sometimes abbreviated as ABR or ABER.

autosome - one of the numbered, non-sex chromosomes.

balanced translocation - a physical rearrangement can be seen in the chromosomes but the correct amount of genetic material is present.

bicornuate uterus - a uterus with horn-shaped branches.

capillary hemangiomas - a flat, red birthmark created by tiny blood vessels close to the skin surface.

CVS (chorionic villus sampling) - removal of a small portion of the interior placenta for genetic testing during early pregnancy.

chromosome - a small, threadlike piece of dark-staining material within the cell, made up of DNA (deoxyribonucleic acid) and proteins, which "carries" the genes.

cleft lip and/or palate - a gap in the soft palate and roof of the mouth, sometimes extending through the upper lip and nostril.

coloboma - defect of the iris tissue in the eye.

congenital - present at birth.

cryptorchidism - testes have not descended to the scrotum.

dermatoglyphics - ridged patterns of the fingers, palms, toes and soles of the feet; unusual patterns may suggest a chromosome abnormality.

dextrocardia - heart is shifted toward the right, or is formed in a mirror image of the normal heart.

fetus - the product of conception from the eighth week to the time of delivery.

GE (gastro-esophageal) reflux - splashing or rising of food and stomach acids from the stomach to the esophagus.

gastrostomy - method of feeding the child through a tube permanently inserted into the stomach through the abdomen.

gavage - feeding through a tube introduced into the mouth or nose.

gene - one of the 50,000 to 100,000 recipes or codes which direct development and function. Different genes govern different features.

holoprosencephaly - the front of the brain, or forebrain, does not divide properly.

karyotype - the photograph of a set of chromosomes arranged in a standardized order.

meninges - the three membranes enveloping the brain and spinal cord; namely, the dura mater, the pia mater, and the arachnoid.

microcephaly - unusually small head. micrognathia - unusually small jaw or chin. microphthalmia - unusually small eye.

mosaicism - in an individual, the presence of cells with different chromosome constitutions.

myelomeningocele - (spina bifida) protrusion of the spinal cord and its covering through an opening in the bony spinal canal.

nondisjunction - the failure of chromosome pairs to separate correctly during division, resulting in an abnormal number of chromosomes in the cells.

omphalocele - presence of abdominal organs in the umbilical cord.

palpebral fissure - eyelid opening.

partial trisomy - portions of an additional chromosome are present in body cells.

PDA (patent ductus arteriosus) - a small heart duct which normally closes soon after birth remains open, resulting in abnormal direction of blood flow.

polydactyly - extra fingers and/or toes.

prenatal diagnosis - the detection of specific genetic disorders prior to birth through the use of techniques such as amniocentesis, chorionic villus sampling, and ultrasound.

retinal dysplasia - faulty development of the retina in the eye.

scoliosis - a lateral or side-to-side curvature of the spine in an "S" shape.

sex chromosome - the X and Y chromosomes which determine the sex of an individual.

sternum - breastbone.

translocation - attachment of part or all of one chromosome to another chromosome; in some cases pieces of chromosomes "trade places."

trisomy - three copies of a particular chromosome, rather than the usual pair, are present in body cells.

ultrasound - (sonogram) a computerized method of visualizing internal organs, or a fetus in the uterus, through the use of sound waves which create a picture similar to an x-ray.

unbalanced translocation - extra and/or missing chromosomal material due to a rearrangement. This may be new in the individual, or may be inherited from a parent who has a balanced translocation.

VSD (ventricular septal defect) - a hole between the two lower chambers of the heart which prevents the heart from pumping blood correctly; a heart murmur is generally heard.