Tetrasomy 18p

There are five major conditions involving large changes of chromosome 18. Each of these conditions has a wide variety of characteristics. Additionally, each of the conditions can vary in severity. Although every child with a chromosome change is different, these pages provide a general idea of the medical and developmental concerns that are associated with the conditions.

If you are unfamiliar with genetic concepts, we encourage you to visit our website. There, you will find several podcasts that introduce genetic concepts. Understanding basic genetics will help give the information provided here more meaning.

The diagram below illustrates a pair of normal chromosome 18's. The conditions of chromosome 18 occur when there are changes in one of these two chromosomes. The five most common changes are shown below.

Our Mission:
To help individuals with chromosome 18 abnormalities overcome the obstacles they face so they might lead healthy, happy, and productive lives.

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Background and Genetic Basis

The goal of this page is to describe the major features of Tetrasomy 18p. This information was obtained from a thorough review of the literature as well as from the experiences of the Chromosome 18 Clinical Research Center. This information may help you and your healthcare team make decisions about how to care for a person with Tetrasomy 18p.

As you read through this article, remember that no two people with Tetrasomy 18p are exactly alike. One person may have different medical and developmental concerns from another person with Tetrasomy 18p. Also, remember that no one with Tetrasomy 18p will have all of these features. In addition, people with Tetrasomy 18p share many features with their family members. They will also have their own unique skills and abilities which you will not find in the following list.

Research is critical. As we learn more about Tetrasomy 18p, we also learn more about how to best treat it. This will improve the health and development of people with Tetrasomy 18p.

Background

Tetrasomy 18p is a relatively rare condition. It has been called various names. You may also hear this condition called “isochromosome 18p”.

Genetic Basis of Tetrasomy 18p

Individuals with Tetrasomy 18p have 47 chromosomes instead of 46. The extra chromosome is made up of two additional copies of the short arm of chromosome 18. Therefore, there are a total of four copies of the short arm (the p arm) of chromosome 18. This is why the condition is called Tetrasomy 18p. The prefix “tetra” simply means “four”. For more information about basic genetic concepts, please visit our website at www.chromosome18.org.

Photographs courtesy of Rick Guidotti of Positive Exposure (www.positiveexposure.org)
Characteristics of Tetrasomy 18p

Problems in the Newborn Period

Newborns with Tetrasomy 18p (tet18p) often have problems at birth or shortly thereafter. The most common problem is nursing/feeding difficulties. Infants may have problems latching onto the breast or bottle. They may also have problems coordinating the suck-swallow motion that is required to nurse. Some infants may vomit frequently after eating. In some cases, a referral to an occupational or physical therapist may be recommended. In more serious cases, a feeding tube may be required. Supplementing their diet with a high-calorie formula may also be recommended to help the infant gain weight.

Infants with tet18p may have other problems. Jaundice is a build-up of bilirubin in the baby’s blood, leading to a yellowish color of the skin and eyes. Occasionally, this resolves on its own. However, many infants with tet18p require treatment for jaundice. Treatment is usually very easy. Typically, the baby is simply placed under a light. This helps break down the extra bilirubin in the baby’s blood.

Some infants with tet18p have breathing problems shortly after birth. To assist with the baby’s breathing, doctors may have to provide extra oxygen. This may be done by putting a tube into the baby’s nose.

Development

Tet18p changes the way the brain develops and works.

Infants, toddlers, and young children with tet18p may develop more slowly than those without tet18p. For example, it may take a little longer for them to roll over, sit, crawl, and walk. It may also take longer for them to reach for and grab toys, hold a bottle, and to feed themselves. Language skills may also develop later than their peers. On average, babies with tet18p roll over at about seven months, and walk and say their first words at about 2½ years. It is important to remember that these numbers are averages. This means that some babies reach their milestones earlier than these averages while others reach their milestones later.

People with tet18p typically have some degree of cognitive disability, though the degree of impairment varies among individuals. Based on standardized testing, people with tet18p typically have IQ scores in the moderate range of impairment.

Photographs courtesy of Rick Guidotti of Positive Exposure (www.positiveexposure.org)
Neurological Changes

People with tet18p usually have some changes in their muscle tone. They may have increased or decreased muscle tone. This is called hypertonia and hypotonia, respectively. Changes in muscle tone can lead to other difficulties. For example, infants with low muscle tone may have difficulty eating because the muscles surrounding the mouth are weak. Children with high muscle tone may also have spasticity. This means that there are uncontrolled muscle spasms when the muscle is stretched by someone else. Changes in muscle tone and spasticity may also lead to delays in meeting developmental milestones, such as sitting without support and walking. Physical, occupational, and speech therapy may improve some of these problems.

Seizures also happen more often in people with tet18p than in people without tet18p. If seizures are suspected, a doctor may request an electroencephalogram (EEG). They may also refer the patient to a neurologist to help manage the seizures.

Spina bifida has been reported in a small number of people with tet18p. Spina bifida, also known as myelomeningocele, is a type of birth defect. It is an opening in the spinal cord that occurs early in the first trimester of pregnancy. This can lead to paralysis of the legs, problems with the kidneys and urinary system, and hydrocephalus (extra fluid in the brain). Spina bifida is usually treated by a team of specialists, including neurosurgeons, therapists, and urologists. To find a spina bifida clinic in your area, visit the Spina Bifida Association website.

MRI Changes

Some people with tet18p have changes in the structure of their brain that can only be detected with an MRI. For example, several people have been identified with a thin corpus callosum. The corpus callosum is the bundle of nerves that connect the right and the left sides of the brain. Other people have been diagnosed with “enlarged lateral ventricles”. This simply means that the spaces that contain the cerebrospinal fluid in the brain are larger than expected.

Eyes and Vision

Eye problems are common in people with tet18p. The eyes may be misaligned (strabismus). People with tet18p may also have problems with their vision. They may be near-sighted, far-sighted, or have astigmatism.

Because vision problems are possible, people with tet18p should have regular eye exams.

Ear Infections

Recurrent infections are common in babies and toddlers with tet18p. They occur in approximately 50% of individuals. Untreated ear infections may lead to hearing problems. Therefore, it is important to identify and treat ear infections. Most of the time, medicine is prescribed to treat the ear infection. Some children may require surgery to insert tubes in the ears to reduce the number of ear infections.

Hearing

Although hearing loss does occur in some people with tet18p, the majority do not have any hearing problems. It appears that, even in the people with some hearing loss, the majority have a mild to moderate hearing loss. Although hearing loss is not very common in people with tet18p, it is still important to screen them for any potential hearing problems. This will help find and treat hearing loss early.
Palatal Abnormalities

The palate is the roof of the mouth. Many people with tet18p have changes in the shape of their palate. For example, the palate may be highly arched and/or narrow. In some cases, these changes may lead to some difficulties with speech. It is important to understand that this is not the same thing as a cleft palate (in which the palate does not completely close).

If a physician feels that a change in the palate may lead to speech problems, he or she may refer a patient to an ENT (a doctor who specializes in problems of the ears, nose, and throat).

Heart

Approximately 30% of people with tet18p have some type of heart defect. Many of the heart defects are “septal defects”, meaning that there is a hole in between the two sides of the heart. In some cases, these defects close without any intervention. However, in some cases, surgery is necessary.

Because heart defects are more common in babies with tet18p, they may have an ultrasound of the heart (echocardiogram) to rule out such defects.

Gastrointestinal Changes

Gastrointestinal problems are fairly common in people with tet18p. The most common concern is constipation. Reflux also occurs somewhat frequently. Structural malformations of the gastrointestinal system are not very common. However, pyloric stenosis and hernias have been reported. Pyloric stenosis is a closure or narrowing of the place where the stomach contents enter the intestines. A hernia occurs when some organs, often the intestines, push outside of the abdomen.

If there is a concern for gastrointestinal problems, a referral to a gastroenterologist is appropriate.

Genitourinary Changes

Males with tet18p may have some changes in the genital region. The testicles may not be fully descended (cryptorchidism). The opening of the urethra may not be at the end of the penis (hypospadias).

Some people have vesicoureteral reflux. This occurs when urine flows from the bladder up towards the kidneys. This can lead to recurrent urinary tract infections. However, structural changes in the kidneys occur in a small number of males and females with tet18p. A doctor may order an abdominal ultrasound to rule out structural changes in the kidney.

Musculoskeletal Changes

Some people with tet18p may have minor changes in their hands and feet. For example, they may have narrow feet, or they may have fingers and toes that are partially fused. They may have camptodactyly. Camptodactyly occurs when fingers are in a permanently “flexed” position. However, these issues seldom require medical intervention.

People with tet18p may have scoliosis or kyphosis. This simply means that they have an abnormal curvature of the spine. Flat feet are another common orthopedic concern.
Growth

Children and adults may have changes in their growth patterns. Children with tet18p may be small for their age. In a minority of cases, this is due to growth hormone deficiency. Treatment with growth hormone helps normalize growth and may improve a child’s development.

You can find several articles about growth hormone deficiency in people with chromosome 18 conditions on our website (www.chromosome18.org).

In addition to short stature, many people with tet18p have microcephaly, or a head size that falls below the 3rd percentile.

Facial Features

People with tet18p may have facial features that are slightly different from other family members. These changes do not affect a child’s health or development. They are simply small differences that might be noted by a doctor.

They may have changes in the structure or the placement of the ears. They may have a small mouth, and the area above the upper lip may be smooth. The upper lip may be thin. Their chin may be either larger or smaller than other children's chins.

Although people with tet18p may have facial features in common with one another, it is important to remember that they also have features in common with their family members.

Family Planning and Genetic Counseling

Many parents wonder, “If we have another child, what is the chance that our next child will have Tetrasomy 18p?”

The answer to this question depends on whether a chromosome change has been identified in one of the parents. In nearly all cases, neither parent has a chromosome change. In this situation, the chance that a couple will have another child with tet18p is low.

In a very small number of families, one parent has a chromosome change. If a parent has a chromosome change, there is a greater possibility that another child could have tet18p.

If you have questions about the implications of a chromosome change for other family members, we recommend contacting a genetics provider.
Care of an Individual with Tetrasomy 18p

Based on a thorough review of the medical literature as well as the findings and experiences of the Chromosome 18 Clinical Research Center, our team of scientists have compiled a list of recommended evaluations for an individual recently diagnosed with tetrasomy 18p.

- Genetics evaluation and counseling
- Parental chromosomes
- Periodic ophthalmology evaluations
- Periodic audiology evaluations
- ENT referral for management of chronic otitis media
- Cardiology evaluation
- Renal ultrasound
- Orthopedic evaluation for management of foot abnormalities
- Monitor for scoliosis and kyphosis
- Neurology evaluation for seizures, abnormal muscle tone
- Gastrointestinal/nutritional evaluation of failure to thrive, gastroesophageal reflux, constipation
- Endocrinology evaluation for short stature, to include evaluation for growth hormone deficiency
- Referral for developmental services and therapy

The information provided here is intended to be used in conjunction with conversations with your health care providers. This information should not replace professional medical advice, diagnosis, or treatment. If you have questions or concerns, you may find it helpful to talk with a clinical geneticist or genetic counselor.

For Additional Information

The information provided here is general information based on the literature as well as the experiences in the Chromosome 18 Clinical Research Center. However, every person with Tetrasomy 18p is different. Therefore, this information should not replace professional medical advice, diagnosis, or treatment. If you have questions or concerns, you may find it helpful to talk with a clinical geneticist or genetic counselor. You can locate a genetics provider at one of these sites:

GeneTests Clinic Directory (www.geneclinics.org)

National Society of Genetic Counselors (www.nsgc.org)
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With your help, we will achieve our mission:

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