



# 18p-

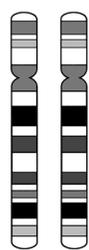
There are five major conditions involving large changes of chromosome 18. Each of these conditions has a wide variety of characteristics. Additionally, each of the conditions can vary in severity. Although every child with a chromosome change is different, these pages provide a general idea of the medical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

If you are unfamiliar with genetic concepts, we encourage you to visit our website ([www.chromosome18.org](http://www.chromosome18.org)). There, you will find several podcasts that introduce genetic concepts. Understanding basic genetics will help give the information provided here more meaning.

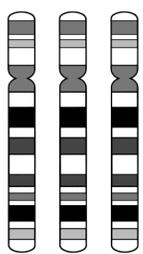
The diagram below illustrates a pair of normal chromosome 18's. The conditions of chromosome 18 occur when there are changes in one of these two chromosomes. The five most common changes are shown below.

### ***Our Mission:***

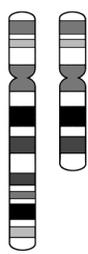
***To help individuals with chromosome 18 abnormalities overcome the obstacles they face so they might lead healthy, happy, and productive lives.***



Normal Chromosomes



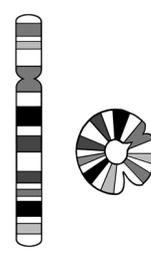
Trisomy 18



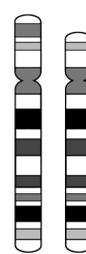
18q-



Tetrasomy 18p



Ring 18

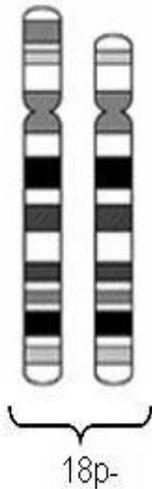
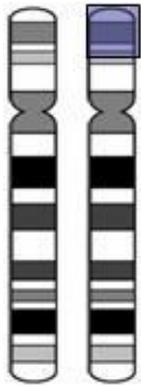


18p-

The Chromosome 18 Registry & Research Society  
7155 Oakridge Drive  
San Antonio, Tx 78229  
Phone: 210-657-4968

Administrative Director: [Gellwanger@chromosome18.org](mailto:Gellwanger@chromosome18.org)  
Executive Director: [CJTraa@chromosome18.org](mailto:CJTraa@chromosome18.org)

## Background and Genetic Basis



*The loss of part of the short arm of the chromosome causes 18p-. The deletion may include the entire short arm of chromosome 18 or only a part of the arm.*

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

As you read through this article, remember that no two people with 18p- are exactly alike. One person may have different medical and developmental concerns from another person with 18p-. Also, remember that no one with 18p- will have all of the features discussed below. In addition, people with 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.

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

### **Background**

18p- was first described in the 1960s. Since then, over 100 case reports have been published, describing the features of 18p-.

18p- has been called several different names. You may hear this condition called “monosomy 18p”, “18p deletion syndrome”, or “18p- syndrome”.

### **Genetic Basis of 18p-**

The term “18p-” means that part or all of the short arm of chromosome 18 is missing, or deleted, from one of the two copies of chromosome 18. For more information about basic genetic concepts, please visit our website at [www.chromosome18.org](http://www.chromosome18.org).

To the left is a diagram showing the difference between a normal pair of chromosome 18's and one with a deletion of the short arm.

In most individuals, the deletion is the only chromosome change present. However, in some cases, the deletion results from a more complicated chromosome rearrangement. For example, some people have 18p- because of an unbalanced translocation. An unbalanced translocation may lead to 18p- and a duplication of another piece of chromosome. In this case, predicting what type of problems a child might have is more difficult. People with an unbalanced translocation may have features of 18p- as well as features of the chromosome duplication.

# **Characteristics of 18p-**

## **Development**

In many individuals, 18p- changes the way the brain develops and works.

Infants, toddlers and young children with 18p- may develop more slowly than those without 18p-. 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 also develop later than their peers. Almost all children with 18p- will require some type of therapy, such as speech, occupational, or physical therapy.

People with 18p- may have some mental impairment, though the degree of impairment varies among individuals. Based on standardized testing, people with 18p- appear to have IQ scores ranging from average to severe mental impairment.

People with 18p- may also have some challenges with adaptive skills. The phrase “adaptive skills” refers to a person’s social skills as well as their level of independence. In the case of 18p-, a person’s IQ may not be directly related to their adaptive skills. Often a person’s adaptive skills are higher than their IQ score might suggest.

It is important to remember that every person with 18p- is different. Some may be more severely affected than others. At this time, we cannot predict exactly how a deletion of 18p will affect a child’s development.

## **Holoprosencephaly**

Some people with 18p- have a condition called holoprosencephaly. Holoprosencephaly is a type of birth defect in which the brain fails to divide into two separate halves during early embryonic development. This term includes a wide range of severity. In some babies, this condition is so severe that they do not survive in the womb or they may die shortly after birth.

Other individuals may have milder forms of the condition. For example, an MRI may show that their brain has minor changes, such as a missing corpus callosum (the connection between the two halves of the brain). There may be changes in the facial features as well, such as a cleft lip and palate or hypotelorism (eyes that are closely set to one another). Another minor feature of holoprosencephaly is a single incisor (front tooth) located at the midline of the mouth.

Individuals with holoprosencephaly may have a number of different health concerns. Many have developmental delays. Seizures and hydrocephalus (build-up of fluid in the brain) may also occur. Some people with holoprosencephaly may have hormone problems caused by a change in the structure of the pituitary gland in the brain.

## Neurological Changes

People with 18p- may have neurological problems that are unrelated to the presence or absence of holoprosencephaly. For example, they may have low muscle tone (hypotonia). 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. Seizures have also been diagnosed in a small number of people with 18p-. Children with holoprosencephaly have a higher likelihood of having seizures.

Dystonia has been reported in a handful of people with 18p-. Dystonia is an involuntary contraction of muscles. Because the muscles cannot relax, people with dystonia may have changes in their posture. Tethered cord has also been reported in some people with 18p-. A tethered cord occurs when the spinal cord is attached to the bony part of the spine. This can lead to bowel and bladder problems as well as back pain or curvature of the spine.

If there are neurological concerns, a person may be referred to a neurologist for a complete evaluation. If there is a concern of seizures, a primary care physician or a neurologist may request an electroencephalogram (EEG).

## Eyes and Vision

Vision problems are often found in people with 18p-. Near-sightedness and far-sightedness are fairly common. Also, the eyes may be misaligned (strabismus). Another common eye problem is ptosis. Ptosis occurs when the eyelid droops. In some cases, ptosis can cause problems with vision by covering the eye. When this happens, a person's vision is reduced.

Because vision problems are possible, people with 18p- should have regular eye exams. In some cases, surgery may be required to treat strabismus or ptosis.

## Ear Infections

Babies, toddlers, and children with 18p- may have more ear infections than other children. Ear infections may lead to hearing problems if left untreated. 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.

Because there is a risk for hearing loss due to the ear infections, people with 18p- should have regular hearing screening. This will help find and treat hearing loss early.

## Cleft Lip and Palate

Cleft lips and palates are more common in babies with 18p- than in the general population. The palate is the roof of the mouth. Sometimes the palate does not form correctly during development. This results in an opening in the roof of the mouth. A cleft lip occurs when the tissue that forms the upper lip does not fuse during prenatal development. Cleft lip and palate may lead to dental, hearing, speech, and feeding problems.

In most cases, a team of professionals is involved in the management of cleft lip and palate. This team may include surgeons, speech pathologists, dentists, audiologists, geneticists, and other professionals. To locate a team in your area, contact the [Cleft Palate Foundation](http://www.cleftline.org) ([www.cleftline.org](http://www.cleftline.org)).

## **Heart**

Heart defects are found in about 25% of babies with 18p-. There are several different kinds of heart defects that have been diagnosed in babies with 18p-. There doesn't appear to be a specific type of heart defect that is more commonly associated with 18p-.

Because heart defects are more common in babies with 18p- than in those without 18p-, an echocardiogram (ultrasound of the heart) may be recommended to look for defects.

## **Gastrointestinal and Other Abdominal Changes**

Babies and children with 18p- may have some problems with digestion. The most common digestive problem is chronic constipation. A physician may prescribe dietary changes or medication to help manage constipation.

Hernias may also occur in babies with 18p-. A hernia occurs when some organs, often the intestines, push outside of the abdomen. This problem is usually corrected by surgery.

There have been a very limited number of reports in the scientific literature of people with 18p- and a rearrangement of their internal organs or additional organs. For example, in one individual, the location of the internal organs was reversed (situs inversus). In other individuals, there was an extra spleen (accessory spleen), or a malrotation of the intestines. In these case reports, the changes did not require surgical correction. It should be noted, however, that these types of malformations may require surgical intervention on occasion.

## **Genitourinary Changes**

Males with 18p- 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). In some cases, surgery may be required to correct these concerns.

## **Growth**

Children and adults may have changes in their growth patterns. Children with 18p- are often small for their age. In some cases, this is due to growth hormone deficiency. Growth hormone deficiency has been reported frequently within the literature as well as by research participants at the Chromosome 18 Clinical Research Center.

If there is a concern regarding growth, a person can see a pediatric endocrinologist to rule out growth hormone deficiency. Drs. Jannine Cody and Daniel Hale have written an article for the Chromosome 18 Registry & Research Society about growth hormone deficiency in children with chromosome 18 changes. To read it, visit [www.chromosome18.org](http://www.chromosome18.org).

In addition to short stature, many people with 18p- have microcephaly, or a head size that falls below the 3rd percentile.

## Thyroid Changes

Some people with 18p- have thyroid problems. Thyroid hormones regulate a number of functions in the body, including how fast the heart beats and how quickly a person burns calories. Some signs of low thyroid hormone include fatigue, weight gain, and depression.

People with 18p- should be screened for thyroid problems once a year. This is because thyroid problems can arise at any time in their life. This screening can be done through an annual blood test. If a thyroid problem is found, an endocrinologist may prescribe medications to treat the problem.

## Immunology

Low levels of IgA are found in some people with 18p-. IgA is a protein that helps fight off infections. People who have a low level of IgA are more likely to get infections and colds than those with normal levels of IgA. For example, they may have lots of ear and sinus infections.

In most cases, IgA deficiency is managed by treating infections, allergies, and asthma early.

## Facial Features

People with 18p- 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.

For example, people with 18p- may have ears that are lower-set and look slightly different from a "typical" ear. They may have an extra fold of skin covering the corner of the eye. The lower jaw may be slightly smaller than in people without 18p-.

Although people with 18p- 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.



*Photographs courtesy of Rick Guidotti of Positive Exposure ([www.positiveexposure.org](http://www.positiveexposure.org))*

## **Family Planning and Genetic Counseling**

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

The answer to this question depends on whether a chromosome change has been identified in one of the parents. In most cases, neither parent has a chromosome change. In this situation, the chance that a couple will have another child with 18p- is very low.

In a small number of families, one parent has an 18p deletion. If a parent has a deletion, there is a 50% chance that they will have a child with 18p-.

In some families, the deletion of 18p results from a more complicated chromosome rearrangement in a parent, such as a translocation. In this situation, the likelihood that another child would have a chromosome change depends on what type of rearrangement the parent has and which chromosomes are involved.

If you have questions about the implications of a chromosome change for other family members, we recommend contacting a genetics provider.

## **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 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](http://www.geneclinics.org) (www.geneclinics.org)

[National Society of Genetic Counselors](http://www.nsgc.org) (www.nsgc.org)



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

## Additional References

Awaad Y, Munoz S, Nigro M (1999): Progressive dystonia in a child with chromosome 18p deletion, treated with intrathecal baclofen. *J Child Neurol* 14: 75-77.

Bühler E, Bühler U, Stalder G (1964): Partial monosomy 18 and anomaly of thyroxine synthesis. *Lancet* Jan 18;1:170-171.

De Grouchy J (1969): The 18p-, 18q-, and 18r syndromes. *Birth Defects Orig Art Ser* 5(5): 74-78.

Faust J, Habedank M, Nieuwenhuijsen C (1976): The 18p- syndrome: Report of four cases. *Europ J Pediat* 123: 59-66.

Fischer P, Golob E, Friedrich F, Kunze-Mühl, Doleschel W, Aichmair H (1970): Autosomal deletion syndrome: 46,XX,18p-: a new case report with absence of IgA in serum. *J Med Genet* 7: 91-98.

Klein C, Page C, LeWitt P, Gordon M, de Leon D, Awaad Y, Breakefield X, Brin M, Ozelius L (1999): Genetic analysis of three patients with an 18p- syndrome and dystonia. *Neurology* 52: 649-651.

Movahhedian H, Kane H, Borgaonkar D, McDermott M, Septimus S (1991): Heart disease associated with deletion of the short arm of chromosome 18. *Del Med J* 63(5): 285-289.

Pearl W (1989): Heart disease associated with deletion of the short arm of chromosome 18. *Pediatr Cardiol* 10: 174-176.

Schinzel A, Schmid W, Lüscher U, Nater M, Brook C, Steinmann B (1974): Structural aberrations of chromosome 18: the 18p- syndrome. *Archiv für Genetik* 47: 1-15.

Sabater J, Antich J, Lluch M, Pérez del Pulgar J (1972): Deletion of short arm of chromosome 18 with normal levels of IgA. *J Ment Defic Res* 16: 103-111.

Thompson R, Peters J, Smith S (1986). Intellectual, behavioral, and linguistic characteristics of three children with 18p- syndrome. *Dev Behav Ped* 7(1): 1-7.

Tonk V, Krishna J (1997): Case report: Denovo inherited 18p deletion in a mother-fetus pair with extremely variable expression, confirmed by fluorescence in situ hybridization (FISH) analysis. *Eur J Obstet Gynecol Reprod Biol* 73(2): 193-19

## Board of Directors

### Officers

#### *President*

Jannine D. Cody, Ph.D.

#### *Vice President for Member Relations*

Catherine Burzio

#### *Vice President for Public Relations*

Ben Flowe, Jr.

#### *Secretary*

Kristen Earl

#### *Treasurer*

Denise Parker

### Staff

Executive Director Claudia Traa

Administrative Director Gloria Ellwanger

If you found this information helpful, or would like to learn more about the Chromosome 18 Registry & Research Society, we encourage you to become a member of our organization.

To become a member, visit our website at [www.chromosome18.org](http://www.chromosome18.org) or call us at 210-657-4968.

With your help, we will achieve our mission :

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