Overview of 1p36 microdeletion syndrome

Each individual with 1p36 microdeletion syndrome (also known as monosomy 1p36) is unique. Most individuals experience minor changes in physical appearance and intellectual and developmental disabilities of varying degrees.

How common is 1p36 microdeletion syndrome?

1p36 microdeletion syndrome is one of the most common chromosome deletions. It is estimated that 1 in every 5,000 to 10,000 individuals have a 1p36 microdeletion; however, it is likely that many individuals who are affected do not get diagnosed.

What causes 1p36 microdeletion syndrome?

1p36 microdeletion syndrome is caused by a deletion ("missing piece") on the short arm ("p") of chromosome 1 (which is where the 1p comes from; the 36 is the precise location on the chromosome that is missing). The deletion results in the loss of several genes. The size of the deletion can vary between individuals with the condition, and it is thought that the larger sized deletions lead to more severe symptoms. Most of the time this deletion is said to be '*de novo*', which means that it was not passed down from a parent; it is brand new to that person in the family. In these cases, the 1p36 microdeletion occurs in the development of an egg or a sperm. About 5-10% of people with 1p36 microdeletion syndrome have inherited the deletion from an unaffected parent with a balanced chromosome rearrangement (called a translocation) that has a break at the 1p36 region.

A balanced translocation occurs when a piece of chromosome 1 breaks off and trades places with a piece of another chromosome. No genetic material is usually lost when this happens; the chromosome pieces just trade places. Because the parent usually has not lost any genetic material, they do not show symptoms for any genetic condition. However, when they have children, their egg or sperm can be missing genetic material from chromosome 1 leading to a higher chance of 1p36 microdeletion syndrome.

What are the health and developmental concerns associated with 1p36 microdeletion syndrome?

1p36 microdeletion syndrome is associated with a variety of health and developmental concerns; however, individuals with the condition can share many of the following characteristics: low muscle tone (hypotonia), growth and feeding problems, small head size

(microcephaly), a prominent forehead, deep-set eyes with straight eyebrows, low set ears, a small mouth, a long area between the nose and mouth (philtrum), and a small pointed chin, among other differences in physical features. Seizures occur in more than half of individuals and many can also have structural differences of the brain. People with this condition may also experience vision and/or hearing problems. Differences of the skeletal, gastrointestinal system, heart, kidneys, or genitalia can also be observed. Many individuals with 1p36 microdeletion syndrome also develop behavioral issues, such as self-harming or aggressive behaviors. Individuals are expected to need extra help and support with learning and development.

What is life like for people with 1p36 microdeletion syndrome?

People with 1p36 microdeletion syndrome can have loving relationships with friends and family, and learn and make progress in their social skills and communication at their own pace. Most individuals with 1p36 microdeletion syndrome will need support and care over their lifetime given the related medical issues.

If I have a baby with 1p36 microdeletion syndrome, what is the chance I will have another baby with this condition?

The chance to have another baby with 1p36 microdeletion depends on the chromosome status of the parents. If both parents are found to have the typical number of chromosomes without any type of rearrangement, the odds for having another baby with 1p36 microdeletion are assumed to be low (less than 1%). If one of the parents is found to have a balanced translocation involving the 1p36 region, the odds to have another affected pregnancy will be higher. These odds will depend on the specific type of translocation observed in the parent. Parents could consider meeting with a genetic counselor to discuss the specific chances and options for prenatal screening and testing or pre-implantation genetic testing.

How do parents of children with 1p36 microdeletion syndrome feel about raising a child with the condition?

Many families of children with 1p36 microdeletion syndrome share valuable information and support with each other through the 1p36 Deletion Support and Awareness. Their web site has lots of helpful information for families with a member who has 1p36 microdeletion syndrome.

What is the treatment for 1p36 microdeletion syndrome?

There is no cure for 1p36 microdeletion syndrome. However, medical management and therapeutic intervention can help individuals reach their full potential. Individuals with 1p36 microdeletion syndrome will require regular follow-up and treatment by a variety of health specialists to treat their specific needs. Intervention programs such as physical therapy, speech therapy, occupational therapy, and behavioral therapy can be beneficial to individuals with this condition.

What are the long term outcomes for individuals with 1p36 microdeletion syndrome?

It is important to recognize that the health and developmental effects of 1p36 microdeletion syndrome vary from person to person. Speech and other developmental issues common to 1p36 microdeletion syndrome can be improved with intervention programs. Individuals with 1p36 microdeletion syndrome may have moderate to severe intellectual disabilities, and may benefit from special education. Most individuals with this condition will need support and care over their lifetime given their health and developmental concerns. Generally, individuals with the condition do survive well into adult life.

Other Resources

Unique: 1p36 microdeletion syndrome

1p36 Deletion Support & Awareness

Click here to learn more about scheduling a genetic counseling appointment for pregnancy-related questions.