Overview of Important Information about Cri-du-chat syndrome

Each individual with Cri-du-chat syndrome is unique. Most individuals with Cri-du-chat syndrome have some degree of development and intellectual delay, and are usually moderately to severely affected. Other possible health concerns include congenital (born with) heart defects, seizures, scoliosis, and the development of a hernia in the lower abdomen (inguinal hernia).

What is life like for people with Cri-du-chat syndrome?

People with Cri-du-chat 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 Cri-du-chat syndrome will need support and care over their lifetime given the related medical issues.

What causes Cri-du-chat syndrome?

Cri-du-chat syndrome is caused by a deletion on the short arm (p) of chromosome 5. The symptoms of Cri-du-chat syndrome are thought to be caused by the loss of multiple genes on this chromosome due to the deletion. 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 in the individual who is diagnosed with the condition. In these cases, the deletion occurs in the development of an egg or a sperm and is then copied over into every cell in the developing baby’s body.

In some cases (10-12%), Cri-du-chat syndrome can be inherited when one parent’s chromosome 5 is part of a balanced translocation. A balanced translocation occurs when a piece of chromosome 5 breaks off and trades places with a piece of another chromosome. No genetic material is lost when this happens; the chromosome pieces just trade places. Because the parent 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 5 leading to a higher chance of Cri-du-chat. The specific chance for Cri-du-chat depends upon the chromosome segments involved in the parent’s translocation.

What are the health and developmental concerns associated with Cri-du-chat syndrome?

Cri-du-chat syndrome is associated with a variety of health and developmental concerns. Small head size (microcephaly), low birth weight, delayed growth, and low muscle tone

(hypotonia) are some of the early symptoms of Cri-du-chat syndrome, as well as a high-pitched, cat-like cry. Delayed development, delayed or absent speech, and intellectual disability are also found in individuals with Cri-du-chat syndrome. Some babies may be born with a heart defect. Scoliosis and seizures are other common health concerns associated with Cri-du-chat syndrome. Many individuals with Cri-du-chat syndrome also develop behavioral issues, such as self-harming or aggressive behaviors.

If I have a baby with Cri-du-chat syndrome, what is the chance I will have another baby with this condition?

Because most cases of Cri-du-chat syndrome are caused by a de novo deletion on chromosome 5, the chance of having another child with Cri-du-chat syndrome is expected to be small. However, if the de novo deletion occurred early enough to be present in more than one egg or sperm, there could be a slight increased risk to have another child with the condition.

If Cri-du-chat syndrome was inherited from a parent with a chromosome 5 translocation, the chance of having another child with Cri-du-chat depends upon the specific pieces of chromosomes exchanged. If you are found to have a chromosome translocation, meeting with a specialist, such as a genetic counselor, can help to better understand what your specific risks may be.

How do parents of children with Cri-du-chat syndrome feel about raising a child with it?

Many families of children with Cri-du-chat share valuable information and support with each other through the Five P Minus Society. Their web site has lots of helpful information for families with a member who has Cri-du-chat syndrome.

What is the treatment for Cri-du-chat syndrome?

There is no cure for Cri-du-chat syndrome. However, medical management and therapeutic intervention can help individuals reach their full potential. Individuals with Cri-du-chat 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 Cri-du-chat syndrome.

What are the long-term outcomes for individuals with Cri-du-chat syndrome?

It is important to recognize that the health and developmental effects of Cri-du-chat syndrome vary from person to person. Speech and mobility issues can be improved with intervention programs. Individuals with Cri-du-chat syndrome may have moderate to severe intellectual disabilities, and may benefit from special education. Most individuals with Cri-du-chat syndrome will need support and care over their lifetime given their health and developmental concerns. Life expectancy is expected to be normal for most individuals with Cri-du-chat syndrome.

How common is Cri-du-chat syndrome?

Cri-du-chat syndrome is believed to affect between 1 in 20,000 and 1 in 50,000 people.

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

Click here to learn more about scheduling a genetic counseling appointment for infertility or preconception questions.

Click here to learn more about scheduling a genetic counseling appointment for questions about pediatric or adult genetic conditions.

Other Resources

Five P Minus Society