Overview of important information about Jacobsen syndrome

Each individual with Jacobsen syndrome is unique. All individuals with Jacobsen syndrome have some degree of developmental and intellectual disabilities. Other common health concerns include a blood clotting disorder called Paris-Trousseau syndrome and abnormalities in the heart and bones.

What is life like for people with Jacobsen syndrome?

People with Jacobsen syndrome have loving relationships with friends and family, and learn and make progress in their social skills and communication at their own pace. Individuals with Jacobsen syndrome are expected to need support and care over their lifetime given the associated health concerns.

What causes Jacobsen syndrome?

Jacobsen syndrome is caused by a deletion on the long arm (also called the "q" arm) of the 11th chromosome. This deletion results in the loss of several genes. Most of the time (85%) this deletion is said to be "*de novo*", meaning that it was not passed down from a parent and it is brand new in the individual who is diagnosed with the condition. In this case, 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 (5-15%), Jacobsen syndrome can be inherited when one parent's chromosome 11 has been changed by a "balanced translocation." A balanced translocation occurs when a piece of chromosome 11 trades places with a piece of another chromosome. No genetic material is deleted during this process, the chromosome pieces just trade places. Because the parent has not lost any genetic material, they would not have Jacobsen syndrome. This translocation can become unbalanced when it is passed on, leading to Jacobsen syndrome. In an unbalanced translocation, some genetic material is lost when the parental chromosomes are passed down to the baby. Individuals who have Jacobsen syndrome caused by an unbalanced translocation have lost some genetic material on the end of the long arm (q) of chromosome 11.

What are the health and developmental concerns associated with Jacobsen syndrome?

Jacobsen syndrome is associated with a variety of health and developmental issues. Infants with Jacobsen syndrome often have feeding difficulties and delayed development. Most

individuals with Jacobsen syndrome have some degree of intellectual impairment, and many individuals with Jacobsen syndrome are diagnosed with attention-deficit hyperactivity disorder (ADHD). Behavioral issues such as compulsive behavior are also common. Crossed-eyes and drooping eyelids cause vision problems in many individuals with Jacobsen syndrome, but these issues can be corrected by surgery. Many individuals with Jacobsen syndrome also have skeletal abnormalities like a curved spine (scoliosis). One of the most serious health concerns associated with Jacobsen syndrome is abnormal development of the heart. A portion (around 20%) of individuals with Jacobsen syndrome do not survive early life because of severe heart defects. Around 90% of individuals with Jacobsen syndrome also have a bleeding disorder called Paris-Trousseau syndrome, which causes easy bruising and bleeding.

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

The chance of having another baby with Jacobsen syndrome depends on the type of genetic change that caused Jacobsen syndrome in the individual. In cases where Jacobsen syndrome is caused by a "*de novo*" deletion, there is not a significant chance of having another child with Jacobsen syndrome. When a parent has Jacobsen syndrome, there is a 50% chance of having a child with Jacobsen syndrome. If a parent has a balanced translocation on chromosome 11, there is a higher chance of having another child with Jacobsen syndrome.

What is the treatment for Jacobsen syndrome?

There is no "cure" for Jacobsen syndrome. However, medical management can help individuals reach their full potential. Individuals with Jacobsen syndrome will need regular follow-up and treatment by a variety of health specialists to treat their specific needs. Serious heart abnormalities may require surgical intervention, and surgery to correct skeletal abnormalities may improve quality of life. Surgery may also be performed to improve vision in individuals with Jacobsen syndrome when crossed eyes and drooping eyelids are impairing vision. Because many individuals with Jacobsen syndrome have a bleeding disorder called Paris-Trousseau syndrome, special care needs to be taken during surgery. Therapeutic interventions such as occupational, speech, physical, and behavioral therapies can be beneficial to individuals with Jacobsen syndrome.

What are the long-term outcomes for individuals with Jacobsen syndrome?

It is important to recognize that the health and developmental effects of Jacobsen syndrome

vary from person to person. Around 20% of individuals with Jacobsen syndrome do not survive the first 2 years of life, usually because of severe heart abnormalities. Special medical attention is needed during early life to avoid complications from heart abnormalities or bleeding, but people with Jacobsen syndrome are expected to need care and support over their lifetime. Life expectancy for individuals who survive early life is unknown, but individuals with Jacobsen syndrome have lived to adulthood.

How common is Jacobsen syndrome?

Jacobsen syndrome is believed to affect 1 in 100,000 people.

How do parents of children with Jacobsen syndrome feel about raising a child with Jacobsen syndrome?

Parents' experiences with Jacobsen syndrome are summarized in the document "Having a Son or Daughter with Jacobsen Syndrome/11q Deletion Syndrome: Perspectives of Parents"

Click below for other resources related to Jacobsen syndrome:

11q Research & Resource Group

National Institutes of Health: Jacobsen syndrome

National Organization for Rare Disorders: Partial Monosomy 11q (Jacobsen syndrome)