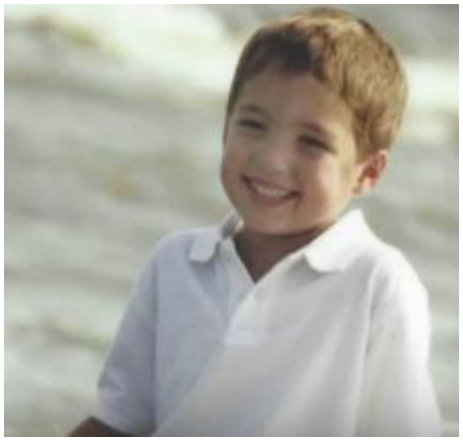
Overview of Important Information about Jacob’s syndrome

Each individual with Jacob’s syndrome is unique. Some individuals with Jacob’s syndrome may be above average height, but Jacob’s syndrome does not affect other physical features. Sexual development is also normal. Individuals with Jacob’s syndrome have an increased risk of learning disabilities, delayed speech, and behavioral problems. A small number of those with Jacob’s syndrome are also diagnosed with autism spectrum disorders. The symptoms of Jacob’s syndrome vary greatly from person to person, but they are generally mild and some individuals with Jacob’s syndrome may remain undiagnosed.

What is life like for people with Jacob’s syndrome?

People with Jacob’s syndrome can have loving relationships with friends and family, go to school with their peers, and develop their social skills and communication. Individuals with Jacob’s syndrome live healthy, full, and independent lives. Most men with Jacob’s syndrome are able to have children of their own.

What Causes Jacob’s syndrome?



Jacob’s Syndrome (47, XYY)

In a typical genome, individuals have two sex chromosomes to determine biological sex. Women usually have two X chromosomes (one from mom and one from dad), and men usually have one X chromosome (from mom) and one Y chromosome (from dad). Individuals with Jacob’s syndrome have one X chromosome and two Y chromosomes, so they have 47 chromosomes total and their sex chromosomes are XYY. Jacob’s syndrome is usually caused by a cell division error in the sperm before conception. Rarely, the cell division error may

occur after conception. These individuals may have mosaic Jacob's syndrome, meaning some of their cells are XYY and some are XY.

What are the Health and Developmental Concerns Associated with Jacob's syndrome?

Jacob's syndrome is associated with few health or developmental concerns. Increased height is one of the main symptoms of Jacob's syndrome and may be noticeable as early as 5 years old. Jacob's syndrome does not cause infertility or abnormal sexual development. Speech delays and learning disabilities such as dyslexia occur in some individuals with Jacob's syndrome. Some individuals with Jacob's syndrome may also develop behavioral issues. There is a small increased risk of autism spectrum disorders associated with Jacob's syndrome.

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

Most cases of Jacob's syndrome are not inherited, but are caused by a cell division error in the sperm before conception. Mosaic Jacob's syndrome is also not inherited. The chance of having another baby with Jacob's syndrome does not increase in these cases.

What is the treatment for Jacob's syndrome?

There is no "cure" for Jacob's syndrome, but therapeutic intervention can help individuals struggling with learning disabilities or behavioral issues reach their full potential. Speech therapy can be beneficial to individuals with speech delays. School support for learning disabilities and/or educational therapy can also be beneficial. Individuals with Jacob's syndrome tend to respond well to intervention measures, and in some individuals these issues may resolve completely.

What are the long-term outcomes for individuals with Jacob's syndrome?

It is important to recognize that the health and developmental effects of Jacob's syndrome vary from person to person. Therapy and other interventions such as speech therapy and support for learning disabilities can be beneficial to individuals with Jacob's syndrome. Men with Jacob's syndrome are expected to live full, independent lives.

How common is Jacob's syndrome?

Jacob's syndrome is estimated to occur in 1 in 1,000 newborn boys.

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.

Resources

The Association for X and Y Variations (AXYS) https://genetic.org/
The Focus Foundation: http://thefocusfoundation.org/