



## **Overview of Important Information about Langer-Giedion syndrome**

Each individual with Langer-Giedion syndrome is unique. Most individuals with Langer-Giedion syndrome have some degree of developmental and intellectual delay. Other common health concerns include the development of bone tumors (osteochondromas) that can cause pain, limit the range of motion in joints, and put pressure on blood vessels and nerves.

### **What is life like for people with Langer-Giedion syndrome?**

People with Langer-Giedion syndrome can have loving relationships with friends and family, and learn and make progress in their social skills and communication at their own pace. Some individuals with Langer-Giedion syndrome are able to live semi-independently, while those with more severe symptoms may need support and care over their lifetime.

### **What causes Langer-Giedion syndrome?**

Langer-Giedion syndrome is caused by a deletion or genetic change on chromosome 8. Two missing genes, *EXT1* and *TRPS1*, cause some of the symptoms seen in Langer-Giedion syndrome. There may be additional deleted genes causing Langer-Giedion syndrome that have not been identified yet. 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 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.

### **What are the health and developmental concerns associated with Langer-Giedion syndrome?**

Langer-Giedion syndrome is associated with a variety of health and developmental concerns, including frequent upper respiratory infections such as sinus infections, sore throats, and tonsil infections. Individuals with Langer-Giedion syndrome tend to be shorter than average, and may have a variety of other skeletal and limb differences. These differences include joined fingers (syndactyly), cone-shaped ends of their long bones, and the development of bone tumors (osteochondromas). These bone tumors are noncancerous, but can cause pain and put pressure on blood vessels, nerves, and the spinal cord. These bone tumors can also limit range of motion if they develop in joints. Some individuals with Langer-Giedion syndrome also develop Coxa plana, which is associated with decreased blood flow at the top of the femur (the large bone in your thigh) that causes this part of the bone to break down. This can cause pain when walking and limit range of motion in the hip joint. The bone can



regrow, but the breakdown may happen repeatedly. Low muscle tone (hypotonia), a wandering eye, delayed speech, and mild to moderate intellectual delay are common in individuals with Langer-Giedion syndrome.

### **If I have a baby with Langer-Giedion syndrome, what is the chance I will have another baby with this condition?**

Because most cases of Langer-Giedion syndrome are caused by a de novo deletion on chromosome 8, it is expected that the chance to have another child with Langer-Giedion syndrome is 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 Langer-Giedion syndrome was inherited from a parent with a chromosome 8 deletion, there is a 50% chance of having another child with Langer-Giedion syndrome.

### **What is the treatment for Langer-Giedion syndrome?**

There is no cure for Langer-Giedion syndrome. However, medical management and therapeutic intervention can help individuals reach their full potential. Individuals with Langer-Giedion syndrome will need regular follow-up and treatment by a variety of health specialists to treat their specific needs, and surgery may be required to remove bone tumors. Intervention programs such as physical therapy and speech therapy can also be very beneficial to individuals with Langer-Giedion syndrome.

### **What are the long-term outcomes for individuals with Langer-Giedion syndrome?**

It is important to recognize that the health and developmental effects of Langer-Giedion 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. Some individuals with Langer-Giedion syndrome are able to live semi-independently, while others may require more care. Life expectancy is expected to be normal for most individuals with Langer-Giedion syndrome.

### **How common is Langer-Giedion syndrome?**

Langer-Giedion syndrome is very rare; there are no estimates of how often this syndrome occurs.



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**

[Langer-Giedion Syndrome Association](#)