

Jacobsen Syndrome

➤ **What is Jacobsen Syndrome?**

- A condition characterized by the deletion of several genes on Chromosome 11. Signs and symptoms vary among individuals with Jacobsen Syndrome but often include Paris-Trousseau Syndrome (a bleeding disorder that also causes easy bruising), distinctive facial features, delayed development of motor skills and speech, and cognitive impairment. The estimated incidence of Jacobsen Syndrome is 1 in 100,000 newborns. More than 200 affected individuals have been reported. .

➤ **What are the signs/symptoms?**

- The signs and symptoms of Jacobsen Syndrome vary. Most common are:
 - Delayed development of motor skills and speech, cognitive impairment and learning disabilities.
 - Behavioral features such as compulsive behavior and ADHD
 - Paris-Trousseau Syndrome
 - Distinctive facial features:
 - Small and low set-ears
 - Wide-set eyes with drooping eyelids
 - Skin folds covering the inner corner of the eyes
 - Broad nasal bridge
 - Down-turned corners of the mouth
 - A thin upper lip
 - Small lower jaw
 - Large head
 - Skull abnormalities that give the forehead a pointed appearance.
 - Congenital heart defects
 - Short stature
 - Feeding difficulties in infancy
 - Frequent ear and sinus infections
 - Skeletal (bone) abnormalities

➤ **What is Jacobsen Syndrome a result of?**

- Jacobsen Syndrome is caused by deletion of genetic material at the end of the long (q) arm of Chromosome 11. The size of this deletion varies among individuals but the deleted area almost always includes the tip of Chromosome 11. The specific features of Jacobsen Syndrome relate to the loss of multiple genes within the deleted region. In about 85% of cases, the deletion is due to a random error during the formation of the egg or sperm, or an error in cell division in early fetal development. Typically this is not inherited however individuals with Jacobsen Syndrome can pass the deletion on to their children. .

➤ **What are the complications of Jacobsen Syndrome?**

- Low platelet counts can lead to needing blood or platelet transfusions before or during surgeries.
- Surgery may be needed to repair various malformations associated with the condition.
- Congenital heart defects (rapid heartbeat or fluid accumulation)
- Eye abnormalities.
- Abnormalities of the joints, tendons, muscles and bones.

References:

<https://rarediseases.info.nih.gov/diseases/307/jacobsen-syndrome>

