

Seckel Syndrome

➤ **What is Seckel Syndrome?**

- Seckel Syndrome is a rare genetic condition of dwarfism that involves intrauterine growth retardation (smaller size at birth), microcephaly (abnormally small head), intellectual disability, short build, large eyes, narrow jaw, deformed hips, bent fifth finger, and birdlike features (receding forehead and chin with beaked nose). Seck Syndrome is observed equally in males and females. This condition is also known as bird-head dwarfism, nanocephaly, and microcephalic primordial dwarfism, Seckle type primordial dwarfism, SCKL.

➤ **What are the signs/symptoms?**

- Some clinical features of Seckel Syndrome include:
 - The circumference of the head is small for the age of the infant
 - Intellectual disability
 - Low birth weight
 - Short Stature
 - Defects in dentition—crowding of teeth, defects in enamel development (hypoplasia)
 - Large eyes, sloped eyelids, eyes are crossed
 - Bird-like features (sloping forehead, beaked nose, unusually small jaw)
 - Ears with no ear lobes
 - Hip, forearm, elbow bone defects and dislocation
 - Bent fifth finger
 - Club foot
 - Excess body hair,

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

- Seckel Syndrome is a rare genetic condition that is inherited in an autosomal recessive manner. This means that the chance of both the copies of the gene to get affected is rare. When one of the copies is affected, the individual is a carrier. Seckel Syndrome is also characterized by microcephaly. When the occipitofrontal head circumference at birth is less than 2 standard deviation of the mean value based on the age, ethnicity and gender, the child is diagnosed with Seckel Syndrome.

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

- The disorders observed in Seckel Syndrome range from structural deformities to blood disorders. There is no confirmed evidence of life expectancy but individuals with Seckel Syndrome are known to have a life expectancy of more than 50 years.

References: <https://www.medindia.net/patients/patientinfo/seckel-syndrome.htm>

