9th May is Cornelia De Lange Syndrome Awareness Day

Cornelia de Lange Syndrome

Cornelia de Lange Syndrome (CdLS) is a genetic disorder present from birth, but not always diagnosed at birth. It causes a range of physical, cognitive and medical challenges and affects both genders equally.

It is often termed as Bushy Syndrome and is also known as Amsterdam dwarfism. It is a genetic disorder that can lead to severe developmental anomalies. It affects the physical and intellectual development of a child. Exact incidence is unknown, but it is estimated at 1 in 10,000 to 30,000.

Causes:

The vast majority of cases are due to spontaneous genetic mutations. Multiple genes have been associated with the condition.

Features and characteristics:

Following are the features and characteristics that help in spotting this disorder:

- Low birth weight (usually under 5 pounds/2.5 kilograms).
- Delayed growth and small stature.
- Developmental delay.
- Limb differences (missing entire limbs or portions of limbs).
- Small head size (microcephaly).
- Thick eyebrows, which typically meet at midline (synophrys).
- Long eyelashes.
- Short upturned nose and thin downturned lips.
- Long philtrum.
- Excessive body hair.
- Small hands and feet.
- Small widely spaced teeth.
- Low-set ears.
- Hearing impairments.
- Vision abnormalities (e.g., ptosis, nystagmus, high myopia, hypertropia).
- Partial joining of the second and third toes.
- Incurved 5th fingers (clinodactyly).
- Gastro esophageal reflux.
- Seizures.
- Heart defects.
- Cleft palate.
- Feeding problems.
- Hypoplastic genitalia.
**Medical Care:**
- Early intervention in patients with Cornelia de Lange syndrome (CdLS) is necessary for feeding problems, hearing and visual impairment, congenital heart disease, and urinary system abnormalities.
- Early intervention for psychomotor delay is also indicated.

**Surgical Care:**
Surgery may be necessary for the following conditions:
- Cleft palate.
- Nasal polyps.
- Gastroesophageal reflux disease.
- Pyloric stenosis.
- Intestinal malrotation/volvulus.
- Undescended testis.
- Lacrimal duct stenosis.
- Hip dislocations.

**Reference Links:**
- [http://en.wikipedia.org/wiki/Cornelia_de_Lange_Syndrome#Features_and_characteristics](http://en.wikipedia.org/wiki/Cornelia_de_Lange_Syndrome#Features_and_characteristics)
- [www.cdlsusa.org/what-is-cdls/characteristics-of-cdls.htm](http://www.cdlsusa.org/what-is-cdls/characteristics-of-cdls.htm)
- [www.cdlsusa.org/what-is-cdls](http://www.cdlsusa.org/what-is-cdls)

**For any enquiry or assistance please contact:**  wellness@medicaretpa.co.in

**Disclaimer:** No information contained here should be relied on in making health decisions. Always check with your doctor or other health care provider.