

*"Health is the greatest gift, contentment the greatest wealth, faithfulness the best relationship." - Buddha*

## 9<sup>th</sup> 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://emedicine.medscape.com/article/942792-treatment#showall>
- ✚ [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)

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