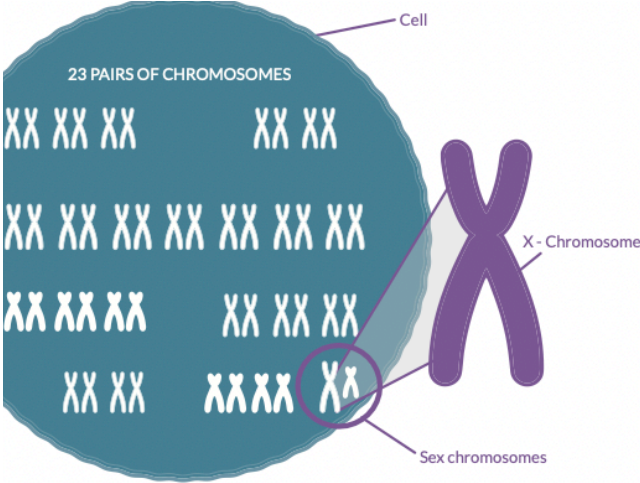


# THE GENETICS OF CdLS

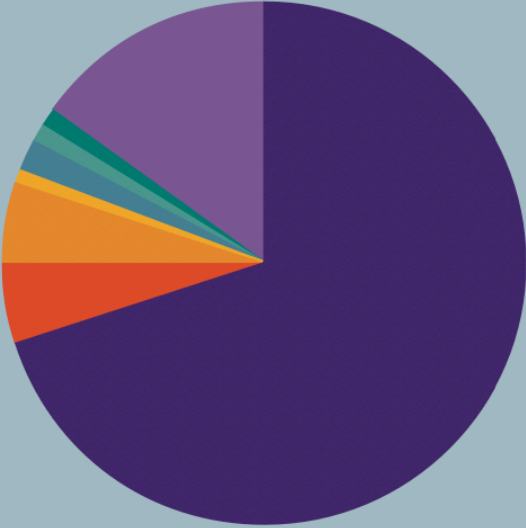
Genetic testing is often used to identify or confirm a CdLS diagnosis. To date there are seven gene variants that can cause CdLS. In the future, researchers may identify additional gene changes that cause CdLS. Research helps us better understand why and how CdLS occurs.



- 1. Five of the seven genes identified (NIPBL, SMC3, RAD21, ANKRD11, BRD4) are located on one of these other 22 pairs of chromosomes.
- 2. Two of the seven genes (SMC1A and HDAC8) identified are located on the X chromosome.

A genetic counsellor can help explain this information and discuss how this may impact your family planning decisions. If you need help finding a genetic counsellor, contact the CdLS Foundation at

# PREVALENCE OF INDIVIDUAL GENE CHANGES



70% NIPBL	2% RAD21
15% Undetermined	1% SMC3
1% BRD4	5% SMC1A
1% ANKRD11	5% HDAC8



# ABOUT THE CANADIAN CDLS FOUNDATION

The Canadian CdLS Foundation was founded in 2019 and is a not-for-profit registered Canadian charity (#707263885-RR001) that serves individuals with CdLS, their families, clinicians and educators.

The Foundation's mission is to:

- 1.Ensure every child born with CdLS in Canada thrives from childhood to old age.
2. Empower families to care and advocate for their family member with CdLS.
3. Provide support and education to families, care providers & educators of individuals with CdLS.
4. Raise awareness of CdLS in Canada to ensure early diagnosis.

# RESOURCES

Website:

[www.CanadianCdLSFoundation.com](http://www.CanadianCdLSFoundation.com)

Family, Clinician & Educator Support:

[support@canadiancdlsfoundation.com](mailto:support@canadiancdlsfoundation.com)



Brochure adapted with permission from the US CdLS Foundation.

# KNOW YOUR CdLS GENES





# WHAT IS CORNELIA de LANGE SYNDROME (CdLS)?

CdLS is a rare genetic spectrum disorder present from birth. This means that during conception a gene changed or mutated.

Most of the time, the gene change occurs spontaneously and has not been passed down from either parent. On occasion, the gene change can be inherited when parents who do not have CdLS have the variant in some of their egg or sperm cells.

CdLS can cause a broad range of potential physical, cognitive, and medical challenges. Physical features and medical conditions occur along a continuum with varying degrees of severity. **No two individuals with CdLS are the same.** It is estimated that CdLS occurs in 1 in 10,000 live births, affecting all races and genders equally. CdLS can remain undiagnosed because it presents in different forms and severity.









## MANAGEMENT GUIDELINES

Identifying concerns and initiating treatment to address medical and developmental issues are vital. The CdLS Management Guidelines outline necessary evaluations, and tests, recommended at different ages and intervals. To request a copy, contact the [support@CanadianCdLSFoundation.com](mailto:support@CanadianCdLSFoundation.com)

## BODY SYSTEMS MOST COMMONLY AFFECTED BY CdLS

Gene Variant/ Change	Cardiac/ Cardiovascular	Cognition and Behavior	Digestive System	Genitourinary	Head and Facial Features	Neurological	Skeletal System	Vision problems / Hearing Loss
NIPBL	✓	✓	✓	✓	✓	✓	✓	✓
SMC1A	✓	✓	✓	✓	✓	✓		✓
SMC3	✓	✓	✓	✓	✓	✓		✓
HDAC8	✓	✓	✓	✓	✓	✓	✓	✓
RAD21	✓	✓	✓		✓	✓		✓
ANKRD11		✓	✓	✓	✓		✓	✓
BRD4	✓	✓			✓		✓	✓

## FINDINGS YOU MAY SEE

 <b>Cardiac/Cardiovascular</b> <ul style="list-style-type: none"><li>• Congenital heart defects</li></ul>	 <b>Genitourinary</b> <ul style="list-style-type: none"><li>• Kidney abnormalities</li><li>• Irregular menstrual periods in females</li><li>• Undescended testicles in males</li><li>• Other male genitalia abnormalities</li><li>• Early prostate enlargement in males</li></ul>	 <b>Neurological System</b> <ul style="list-style-type: none"><li>• Seizures can occur</li></ul>
 <b>Cognition and Behavior</b> <ul style="list-style-type: none"><li>• Intellectual or learning disability (any degree)</li><li>• Autism spectrum disorder</li><li>• ADHD, Anxiety</li><li>• Self-injurious behavior</li><li>• Behavioral issues can escalate in response to discomfort from a medical issue.</li></ul>	 <b>Head and Facial Features</b> <ul style="list-style-type: none"><li>• Decreased head size</li><li>• Thick eyebrows that meet in the middle</li><li>• Long eyelashes</li><li>• Upturned nose</li><li>• Long smooth philtrum (the groove between the top of the lip and the bottom of the nose)</li><li>• Downturned corners of the mouth</li><li>• Small, recessed jaw, high, or cleft palate that can impact feeding</li></ul>	 <b>Skeletal System</b> <ul style="list-style-type: none"><li>• Short stature</li><li>• Smaller than average hands and feet</li><li>• Limb differences,</li><li>• Hip abnormalities</li><li>• Scoliosis</li></ul>
 <b>Digestive System</b> <ul style="list-style-type: none"><li>• Gastrointestinal reflux (GERD): at least 85% of those with CdLS experience some type of reflux</li><li>• Constipation, decreased bowel motility</li><li>• Chance of bowel obstruction</li></ul>		 <b>Vision Problems and Hearing Loss</b> <ul style="list-style-type: none"><li>• Hearing loss</li><li>• Multiple ear infections</li><li>• Visual impairment, often severe nearsightedness</li></ul>