Cornelia de Lange Syndrome
Precision Panel

Overview

Cornelia de Lange Syndrome (CdLS) is archetypical genetic syndrome characterized by intellectual disability, distinct facial features, upper limb anomalies, pernatal and postnatal growth retardation among other signs and symptoms. It is caused by mutations in genes that have a structural or regulatory function in the cohesion complex. Cohesin is a protein that plays a pivotal role in chromatid cohesion, gene expression and DNA repair. It is transmitted in an autosomal dominant and X-linked pattern.

The Igenomix Cornelia de Lange Syndrome Precision Panel can serve as a directed and accurate diagnostic tool ultimately leading to a better management and prognosis of the disease. It provides a comprehensive analysis of the genes involved in this disease using next-generation sequencing (NGS) to fully understand the spectrum of relevant genes.

Indications

The Igenomix Cornelia de Lange Syndrome Precision Panel is indicated in those cases where there is a clinical suspicion or diagnosis with or without the following manifestations:

- Intrauterine growth retardation
- Prematurity
- Facial features: arched eyebrows, short nose with depressed bridge, long philtrum, thin lips etc
- Low-pitched, weak cry in infancy
- Increased muscle tone
- Respiratory and feeding difficulties
- Developmental delay
- Intellectual disability
- Seizures

Clinical Utility

The clinical utility of this panel is:

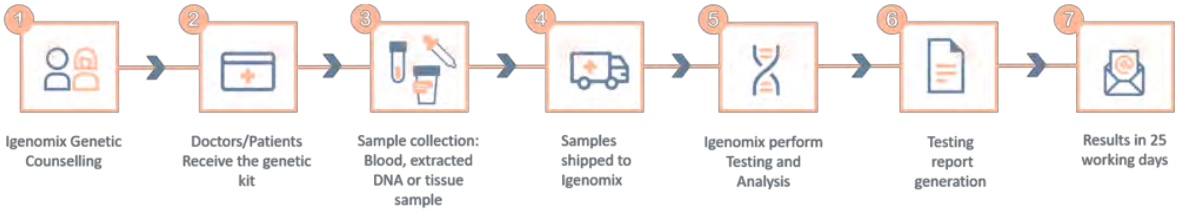
- The genetic and molecular diagnosis for an accurate clinical diagnosis and improve prognosis.
- Early initiation of treatment with a multidisciplinary team in the form nutritional rehabilitation, hearing and visual aids, surgical repair of congenital heart disease and urinary system abnormalities with pertinent consultations to specialists.
- Risk assessment and genetic counselling of asymptomatic family members to identify the individuals at risk.
- Improvement of delineation of genotype-phenotype correlation.

Genes & Diseases

GENE	OMIM DISEASES	INHERITANCE*	% GENE COVERAGE (20X)	HGMD**
HDAC8	Cornelia De Lange Syndrome, Wilson-Turner Syndrome	X,XD,G	99.78	-
KMT2A	Hairy Elbows, Short Stature, Facial Dysmorphism, And Developmental Delay, Cornelia De Lange Syndrome, Wiedemann-Steiner Syndrome	AD	98.14	144 of 149
NIPBL	Cornelia De Lange Syndrome	AD	99.32	409 of 426
RAD21	Cornelia De Lange Syndrome, Mungan Syndrome	AD,AR	99.8	16 of 17
SETD5	Autosomal Dominant Mental Retardation, Cornelia De Lange Syndrome	AD	99.77	37 of 37
SMC1A	Cornelia De Lange Syndrome, Semilobar Holoprosencephaly, Wiedemann-Steiner Syndrome	X,XR,XD,G	100	-
SMC3	Cornelia De Lange Syndrome	AD	100	30 of 30

* Inheritance: AD: Autosomal Dominant; AR: Autosomal Recessive; X: X linked; XLR: X linked Recessive; Mi: Mitochondrial; Mu: Multifactorial
** HGMD: Number of clinically relevant mutations according to HGMD

Methodology



Contact us

Call +34 963 905 310 or send an email to supportspain@igenomix.com for any of the following objectives:

- Get more information about the test.
- Request your kit.
- Request a pick up of the kit after collecting the sample.

References

1. Kline, A. D., Moss, J. F., Selicorni, A., Bisgaard, A. M., Deardorff, M. A., Gillett, P. M., Ishman, S. L., Kerr, L. M., Levin, A. V., Mulder, P. A., Ramos, F. J., Wierzba, J., Ajmone, P. F., Axtell, D., Blagowidow, N., Cereda, A., Costantino, A., Cormier-Daire, V., FitzPatrick, D., Grados, M., … Hennekam, R. C. (2018). Diagnosis and management of Cornelia de Lange syndrome: first international consensus statement. *Nature reviews. Genetics*, *19*(10), 649–666. https://doi.org/10.1038/s41576-018-0031-0
2. Boyle, M. I., Jespersgaard, C., Brøndum-Nielsen, K., Bisgaard, A. M., & Tümer, Z. (2015). Cornelia de Lange syndrome. *Clinical genetics*, *88*(1), 1–12. https://doi.org/10.1111/cge.12499
3. Sarogni, P., Pallotta, M. M., & Musio, A. (2020). Cornelia de Lange syndrome: from molecular diagnosis to therapeutic approach. *Journal of medical genetics*, *57*(5), 289–295. https://doi.org/10.1136/jmedgenet-2019-106277
4. Huisman, S., Redeker, E., Maas, S., Mannens, M., & Hennekam, R. (2013). High rate of mosaicism in individuals with Cornelia de Lange syndrome. *Journal Of Medical Genetics*, *50*(5), 339-344. doi: 10.1136/jmedgenet-2012-101477
5. Dowsett, L., Porras, A. R., Kruszka, P., Davis, B., Hu, T., Honey, E., Badoe, E., Thong, M. K., Leon, E., Girisha, K. M., Shukla, A., Nayak, S. S., Shotelersuk, V., Megarbane, A., Phadke, S., Sirisena, N. D., Dissanayake, V., Ferreira, C. R., Kisling, M. S., Tanpaiboon, P., … Krantz, I. D. (2019). Cornelia de Lange syndrome in diverse populations. *American journal of medical genetics. Part A*, *179*(2), 150–158. https://doi.org/10.1002/ajmg.a.61033