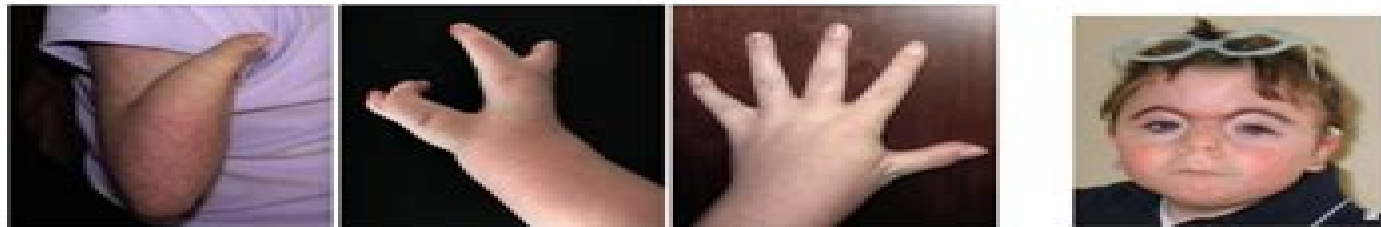


Síndrome de Cornelia de Lange

¿QUÉ ES?

El SCdL puede afectar a diversas partes del cuerpo y las pacientes con SCdL pueden mostrar rasgos físicos, cognitivos y de conducta. Los rasgos cognitivos son procesos cerebrales tales como la memoria y el pensamiento. Las características de comportamiento se refieren a ciertos comportamientos que los pacientes con SCdL es más probable que tengan. Estas características pueden variar. Esto se debe a que las pacientes con este suelen presentar rasgos faciales característicos, patrones de crecimiento alterados y malformaciones en extremidades. Estas características configuran el fenotipo SCdL clásico, cuyas características físicas, cognitivas y conductuales se asocian con el síndrome.



ETIOLOGÍA

- Desconocida. Aunque en su mayoría tienen una mutación en el gen NIPBL. En aproximadamente el 15% de los casos, se desconoce la causa del síndrome de Cornelia de Lange.

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