

## We Already Have a CdLS Diagnosis...Why Follow Up With a Geneticist?

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Most families see the role of the geneticist or genetics clinic as helping to make a diagnosis or facilitating genetic testing. But there is value in returning to the genetics clinic for a follow-up visit or even seeing the geneticist on a regular basis.

First, geneticists and genetic counselors are considered experts in genetic conditions, not only in making a diagnosis, but also interpreting genetic testing results and management of the condition. For example, while other medical specialists focus on their specific fields, geneticists and genetic counselors focus on the genetic syndrome as a whole and provide an overview, such as: *Are all body systems being addressed? Are early intervention therapies appropriate? Is the family aware of specific recommendations or management guidelines at certain ages based on what we know about Cornelia de Lange Syndrome (CdLS)?*

Because of the expertise in the specific condition, the geneticist can provide anticipatory guidance regarding future concerns, referrals to appropriate subspecialists in the area who may have experience with CdLS, and discuss the benefits of a multidisciplinary clinic.

Additionally, questions regarding genetic testing results can be addressed with the genetic counselor or geneticist at a follow-up visit. These questions include: *What does this result mean? What are the chances of having another child with CdLS? Should additional testing be performed if a result is negative?*

Our knowledge and understanding of the gene changes and genes involved in CdLS are rapidly growing. It is the geneticist or genetic counselor's responsibility to re-investigate what we know about specific test results as researchers learn more about how gene changes cause CdLS. Therefore, questions about genetic testing results should be revisited periodically, ideally during a follow-up visit with a geneticist or genetic counselor.

In addition, geneticist or genetic counselor can assess how the family is doing overall and if they would benefit from support resources in the community. This may include facilitating contact with other local families or identifying ways in which the CdLS Foundation may be helpful, given the current issues faced by the family. The genetic counselor can address concerns about the chance of having a second affected child and share options for testing or monitoring any future pregnancies. This may also include questions about future pregnancies for other relatives, such as siblings.

Most importantly the geneticist is a resource and someone to ask the many questions that arise when caring for an individual with CdLS: *What is typical? What should we expect? How should we address this issue?*

To find a clinical geneticist in your area, go to [www.acmg.net](http://www.acmg.net), and search ‘find genetic services,’ or talk to your primary care physician.

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