



Frequently Asked Questions: *CHD7* Testing

Why have *CHD7* testing?

- **To confirm the clinical diagnosis**
 - Fewer doctor appointments searching for a confirmed diagnosis.
 - Qualification for state programs, services, and insurance.
 - Confidence regarding directions of therapies.
- **To have important genetic information**
 - Even though more than 98% of CHARGE syndrome cases occur spontaneously and are not likely to be hereditary, other family members may wish to be tested at child-bearing age, especially siblings. Knowing the mutation allows for targeted blood test analysis in other family members. This is a gift you can give them.
 - Sense of identity and “belonging” to a group with similar mutation.
 - Provides answers to questions about causes for the disorder.
 - Alleviate “what ifs.” “What if I had done *this*,” or “what if I had done *that*” (*or not*). Mutations are almost always spontaneous with no known environmental causes.
- **To learn all you can about the disorder**
 - It may eventually help complete our understanding of CHARGE syndrome - specific mutation types may reveal areas of strengths or concerns.
- **To be eligible for clinical trials and natural history studies**
 - Do it now and be ready
- **To contribute to the expansion of scientific knowledge about *CHD7* mutations**

CHD7 TESTS

- **Sequence Analysis** (gene sequencing, sequencing):
 - This is the most common form of *CHD7* testing. Involves analysis of the entire coding region.
- **Deletion/Duplication Analysis** (MLPA, PCR, Southern blot):
 - If the sequencing analysis is normal, deletion/duplication analysis must be performed next for full *CHD7* testing to have occurred.
- **How do we go about getting the blood drawn for the test?**
 - A local collecting lab will require an order from your child’s physician that states “*CHD7* full sequencing of all coding exons and if negative, duplication/deletion analysis.”

- **The lab collecting the blood has a contract with a commercial lab to perform the tests. Can any commercial lab perform full *CHD7* testing?**
 - No. Be aware that some laboratories only perform sequencing of exons and do not perform deletion/duplication analysis (i.e. not “full” testing).
- **Will my insurance pay for testing and how much does it cost?**
 - Private insurance will sometimes pay for *CHD7* testing, but will often require copays or have deductibles. State Medicaid programs vary from state to state.
 - Check with your insurance company for specific coverage information. Out of pocket costs vary depending on your plan deductibles and copays, whether the test is “covered” and whether the lab is in-network or out-of-network for your plan. Check with both your insurance and the individual lab for prices and billing issues. A letter of medical necessity (LMN) may be required for your insurance to consider covering genetic testing.
- **My child had the sequence test and it was negative. What test should be done now and does she need to have blood collected again?**
 - If only sequencing was originally ordered and was negative, the test for deletion/duplication needs to be performed next. Contact the lab that performed sequencing to determine if a sample is stored and can be used for further analysis, avoiding the need for another blood collection. Check with your insurance about coverage.
- **Full *CHD7* testing was negative. Are there other blood tests that need to be done?**
 - If *CHD7* sequencing and deletion/duplication tests were negative, and you are confident that full testing was performed, speak with your child’s geneticist about other possible testing.
- **All of the tests were negative. Does that mean my child does not have CHARGE?**
 - No. CHARGE is a clinical diagnosis. In about 10-20% of children who have a definite clinical diagnosis, no genetic cause can be identified. Knowing the genetic change in a child is helpful, but not required for a diagnosis. Check with your geneticist in a couple of years – new tests are being developed at a rapid rate.