

TOP 5 REASONS TO HAVE *MECP2* TESTING
even if the clinical diagnosis was made years ago.....

1. To confirm the clinical diagnosis

- Fewer doctor appointments searching for a confirmed diagnosis
- Qualification for state programs, services and insurances
- Confidence regarding directions of therapies

2. To have important genetic information

- Even though more than 99% of RTT 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 if’s”. “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.

3. To learn all you can about the disorder

- It may help complete an individual picture of Rett syndrome - specific mutation types may reveal areas of strengths or concerns

4. To be eligible for clinical trials and natural history studies

- Do it now and be ready

5. To contribute to the expansion of scientific knowledge about *MECP2* mutations

***MECP2* TESTS**

Sequence Analysis (gene sequencing, sequencing):

This is the most common form of *MECP2* testing. Involves analysis of the entire coding region including exons 1-4.

Deletion/Duplication Analysis (MLPA, PCR, Southern blot):

If the sequencing analysis is normal, deletion/duplication analysis must be performed next for full *MECP2* testing to have occurred.



4600 Devitt Drive
Cincinnati, Ohio 45246
+1 513 874 3020 phone
+ 513 874 2520 fax
+1 800 818 RETT (7388) toll free
www.rettsyndrome.org

RECOMMENDED U.S. LABS FOR FULL *MECP2* TESTING

On the next page is a list of recommended laboratories for Rett testing. However, there are several other domestic laboratories that perform the full or selected exons sequencing analysis. Please contact the lab for specific testing information, including sample requirements, costs and billing options.

For non-US residents seeking to have *MECP2* testing in the US, it is recommended that you contact the lab of choice regarding international sample shipping instructions and the shipping courier in your country regarding US Customs regulations. You may need to consult with a local physician regarding sample collection.

Alfred I DuPont Hospital for Children (Delaware)

(302) 651-6039

Email contact: Susan Kirwin, PhD at skirwin@nemours.org

Baylor College of Medicine (Texas)

1-800-411-GENE

<http://www.bcm.edu/geneticlabs/>

Boston University School of Medicine (Massachusetts)

(617) 638-7083

<http://www.bumc.bu.edu/Dept/Content.aspx>

Greenwood Genetic Center (South Carolina)

1-888-GGC-GENE

www.ggc.org

University of Chicago (Illinois)

1-888- UC-GENES

<http://genes.uchicago.edu/>

FAQ'S ABOUT *MECP2* TESTING

How do we go about getting the blood drawn for the test?

- A local collecting lab will require an order from your daughter's physician that states "*MECP2* full sequencing of exon 1-4 and if negative, duplication/deletion analysis".



4600 Devitt Drive
Cincinnati, Ohio 45246
+1 513 874 3020 phone
+ 513 874 2520 fax
+1 800 818 RETT (7388) toll free
www.rett syndrome.org

The lab collecting the blood has a contract with a commercial lab to perform the tests. Can any commercial lab perform full *MECP2* testing?

- No. Be aware that some laboratories only perform sequencing of exons 2-4, some only look for certain point mutations, and some 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 *MECP2* testing. State Medicaid programs will only pay for testing that is performed by an in-state lab. Check with your insurance company for specific coverage information. Costs may vary slightly between labs. Check with the individual lab for prices and billing issues.

My daughter had the sequence test that 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 which performed sequencing to determine if a sample is stored and can be used for further analysis, avoiding the need of another blood collection.

Full *MECP2* testing was negative. Are there other blood tests that need to be done?

- If *MECP2* sequencing and deletion/duplication tests were negative, and you are confident that full testing was performed, speak with your daughter's physician about whether other tests such as *CDKL5* and *FOXG1* are indicated. The decision to test for these should be based on her clinical history and presentation.