



Informed Consent for DNA Testing

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I, _____, request DNA-based testing for [circle] MYSELF and/or MY CHILD or CHILDREN for **Multiple Endocrine Neoplasia Type 1 (MEN1) / MEN1 gene** (name of disease/gene). I understand that biological samples (blood, cheek cells, or skin) will be removed using standard techniques which carry very little risk. In addition, if prenatal diagnosis is being performed, fetal cells obtained by chorionic villus sampling or amniocentesis will be used. I understand that the blood, cheek cells, skin, or fetal samples will be used for the purpose of attempting to determine if I and/or members of my family are carriers of the disease gene, or are affected with, or at increased risk to someday be affected with this genetic disease. The minor children for which I hereby give permission to collect biological samples for this test are named below:

Child's Name	Date of Birth	Gender (M/F)
_____	_____	_____
_____	_____	_____
_____	_____	_____

I understand that:

1. In some cases the DNA test directly detects an abnormality, called a mutation, in the gene, and the test is better than 99% accurate.

In other cases, the DNA test is unable to identify an abnormality although the abnormality may still exist. This event may be due to our current lack of knowledge of the complete gene structure or an inability of the current technology to identify certain types of changes (mutations) in the gene.

GeneDx, Inc. will sequence the coding region of the MEN1 gene, where 75-90% of individuals with Multiple Endocrine Neoplasia Type 1 have been shown to have mutations. Mutations outside of the coding region or in another gene will not be identified. Bi-directional sequence analysis as performed by GeneDx is expected to identify >95% of existing small intragenic mutations; however gross deletions, insertions and rearrangements are not readily detectable by the sequencing approach.

I have been informed of the likelihood of finding a mutation in the gene for which I am being tested.
_____ (Initial)

2. In rare cases, GeneDx may use an indirect method called linkage analysis. If linkage analysis is being used, naturally occurring rearrangements in the DNA (known as "recombination") may produce an uncertainty in predicting carrier status or diagnosis. Rare variations in the DNA of individuals can also cause uncertainty in predicting carrier status or diagnosis. Thus, linkage analysis is not 100% accurate, and the results will be reported as a probability. In some families, the markers used for the linkage analysis may not be informative. In these cases, the DNA test will not be useful for that family or for some family members.
3. An error in the diagnosis of disease status may occur if the true biological relationships of the family members being tested are not as I have stated. For example, non-paternity means that the stated father

of an individual is not the true biological father. This test may detect non-paternity, and it may be necessary to report this finding to the individual who requested testing. Any erroneous diagnosis in a family member can lead to an incorrect diagnosis for other related individuals who are being tested.

4. I understand that the DNA analysis performed by GeneDx is specific for this disease and in no way guarantees my health or the health of my living or unborn children. The accuracy of DNA analysis is entirely dependent on the clinical diagnosis made elsewhere, and GeneDx cannot be responsible for an erroneous clinical diagnosis made elsewhere.
5. In order to perform accurate prenatal diagnosis on a fetal sample, biological samples are also required from the affected individual in the family, the mother, and in some cases the father.
6. These tests are relatively new and are being improved and expanded continuously. The tests are not considered research, but are considered to be the best and newest laboratory service that can be offered. This testing is complex and utilizes specialized materials so that there is always some very small possibility that the test will not work properly or that an error will occur. There is a low error rate (perhaps 1 in 1000 samples) even in the best laboratories. My signature below acknowledges my voluntary participation in this test, but in no way releases the laboratory and staff of GeneDx from their professional and ethical responsibility to me.
7. I understand that my sample is not being banked. GeneDx does not return DNA samples to individuals or physicians. However, in some cases it may be possible for GeneDx to reanalyze the remaining DNA upon request. The request for additional testing must be ordered and there will be an additional fee.
8. Because of the complexity of DNA based testing and the important implications of the test results, results will only be reported to me through a physician, genetic counselor, or certified genetics professional. The result reports are confidential and will only be released to other medical professionals or other parties with my express written consent. All laboratory data is confidential and will not be released from GeneDx. Participation in DNA testing is completely voluntary.
9. I will receive a copy of this consent form.

Signature: _____ Date: _____

Witnessed by: _____

Physician's/Counselor's Statement: I have explained DNA testing to this individual. I have addressed the limitation outlined above, and I have answered this person's questions.

Signature: _____ Date: _____