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Informed Consent for DNA Testing

I, _____, request DNA-based testing for [circle] MYSELF and/or MY CHILD for the **Hyper-IgE Syndromes (HIES) Panel / DOCK8, SPINK5, STAT3 and TYK2 genes or one of the stand-alone tests for analysis of the DOCK8, SPINK5 or STAT3 genes.** I understand that more information about HIES is available from my health care provider and can also be found in the HIES Information Sheet on the GeneDx website (www.genedx.com).

I understand that biological samples will be removed using standard techniques that 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 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. I hereby give permission to collect biological samples for this test from the minor children listed below:

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

I understand that:

1. The methods used by GeneDx are expected to be greater than 99% sensitive in detecting mutations identifiable by sequencing. The DNA test may be unable to identify an abnormality that is present 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.

   The likelihood of finding a mutation in a person with a clinical diagnosis of one of the Hyper-IgE Syndromes is included below:

   - **~60% of individuals with a diagnosis of an autosomal dominant form of HIES harbor a mutation in STAT3**
   - **~75% of individuals with a diagnosis of an autosomal recessive form of HIES harbor mutations in DOCK8**
   - **~66-75% of individuals with a diagnosis of Netherton syndrome harbor mutations in SPINK5**
   - **<<1% of individuals with a diagnosis of HIES harbor mutations in TYK2**

   The testing performed by GeneDx will detect most of the previously reported mutations in the genes listed above. The test is not designed to detect established or novel mutations in non-coding regions of the genes (deep in introns or in promoter regions). It also will not detect heterozygous deletions or duplications involving one or more exons of these genes, with the exception of DOCK8.

   I have been informed of the likelihood of finding a mutation in the gene for which I am being tested and have received test-specific information. _____ (Initials)

2. 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.

3. 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.

4. 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.

5. I understand that my sample is not being banked at GeneDx. DNA samples are not returned to individuals or to referring physicians. However, in some cases if further diagnostic tests are needed, a referring physician may request in writing that additional tests be performed on an existing DNA sample (additional costs apply). Additional testing will not be performed unless requested by an authorized healthcare professional.

6. DNA samples may be kept by the laboratory for up to 25 years. DNA samples may be available for additional diagnostic testing as indicated above and may be used for internal laboratory quality assurance purposes. In some cases, anonymized DNA may be used by the laboratory for new test development after all identifiers have been removed. _____ (Initials)

   NEW YORK STATE ONLY:
   I consent to having my DNA sample retained for greater than 60 days after the completion of the testing.
   PLEASE INITIAL: _____

7. 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, certified genetics professional or other professionals as entitled by law. 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.

8. I will receive a copy of this consent form.

By my signature below, I attest to the following:

1. I have read and understand the information provided on this form.
2. I understand that I may request genetic counseling PRIOR to having this test, and genetic counseling is also available after the test has been completed..

Patient's Signature: _____Date: _____

Signature of Parent/Guardian if patient is a minor: _____

Print name of Parent/Guardian: _____

Health Care Provider's Statement: By my signature below, I indicate that I am the referring physician or authorized health care provider. I have explained the purpose of the test described above. The patient has been given the opportunity to ask questions and/or seek genetic counseling. The patient has voluntarily decided to have the test performed by GeneDx.

Health Care Provider's Signature: _____ Date: _____

