Genetic Testing Informed Consent

A. I am interested in obtaining a genetic test, by submitting a biological sample of my own blood, tissue or other body fluids. I may be pregnant at the time of this genetic test or I may not be pregnant. I understand that the genetic test is for:

Check appropriate test □ Cystic Fibrosis □ HealthCheck | NB (see Exhibit A for complete list of disorders tested)

OR

B. I am the parent or legal guardian of the person who is interested in obtaining a genetic test, by submitting a biological sample of that person’s blood, tissue or other body fluids. The person for whom the sample is submitted may be pregnant at the time of this genetic test or may not be pregnant. In the event that I am completing this form for a person other than myself, I understand that the terms “I”, “me”, “my” and “mine”, as used below, refer to the person for whom the test will be performed. I understand that the genetic test is for:

Check appropriate test □ Cystic Fibrosis □ HealthCheck | NB (see Exhibit A for complete list of disorders tested)

1. The genetic test is being performed to identify whether or not I may be predisposed to contract this disease and/or whether I have this disease or condition.

2. This test may also identify whether I may be a carrier for this disease or condition so that any biological child of mine may be predisposed to contract this disease or have this disease or condition.

3. While results obtained from this type of genetic test are highly accurate, errors such as false positives or false negatives may occur. I understand that this test in no way guarantees my health or the health of my biological child.

4. I understand that genetic testing is a complex subject and that I may wish to obtain professional genetic counseling concerning the risks and benefits of such testing, either before or after genetic testing. My physician has provided me with written information identifying a genetic counselor or medical geneticist from whom I may obtain such counseling. By my signature on this informed consent, I confirm that I have had an opportunity to obtain professional genetic counseling prior to signing this informed consent.

5. It is difficult to be certain that a positive test result for any particular disease or condition will serve as a predictor of such disease or condition. I understand that I must confer with my physician, following my physician’s receipt of any genetic test result, to determine the predictive value of the test result and the advisability of further tests and/or genetic counseling.

6. In the event that my genetic test results are positive for this disease or condition, I understand that I may wish to consider the following:
   A. further independent testing
   B. consultation with my physician or other specialist physician(s)
   C. initial genetic counseling or further genetic counseling

7. I understand that, unless I direct otherwise, I will receive the results of the genetic test(s) performed. A. In the case of HealthCheck | NB, these results will be returned directly to my pediatrician.

8. My test results may be disclosed to the following persons or organizations:
   A. Ordering physician
   B. Other physicians and/or genetic counselors to whom the ordering physician has requested that test results be provided
   C. Please note any other individuals or organizations who may receive genetic test results. A biological mother must specifically authorize adoptive parents to obtain genetic test results for the biological mother’s sample.
9. I specifically authorize genetic test results be provided to my health insurer or health maintenance organization to the extent that they are reasonably required for purposes of claims administration. Further distribution within the insurer or to other recipients shall require my informed consent in each case.

10. I understand that my state may permit certain other persons or organizations that are not listed above access to my test results and that it is my right to delay testing until I have ascertained the laws of my state in this regard.

11. I understand that the results of this test may become part of my permanent medical record and that the results may be material to my ability to obtain certain insurance benefits.

12. I understand that no genetic tests other than those that I have authorized shall be performed on my biological sample and that the sample will be destroyed at the end of the testing process or not more than sixty days after the sample was taken. In the event that this sample is intended to be used for other non-genetic tests, that portion of the sample that is to be used for such tests may be retained for any necessary period of time that is required in connection with such non-genetic tests.

13. I understand that my biological sample may be disclosed or retained for use in anonymous research or coded research unless I request that my biological sample not be used for such research.

14. All records, findings and results of any genetic test performed on my biological sample will be treated as confidential and, except as authorized by me or by law, will not be disclosed without my written informed consent.

15. I represent that I am authorized to provide this consent on my own behalf. If the patient who has submitted the biological sample lacks the capacity to consent, I represent that I have the authority to provide informed consent on the part of such patient.

I have read and understand this consent form. I give my consent for genetic testing on the biological sample.

Signature: ____________________________________________

(Self)

Date: ________________

Print Name: ________________________________

If signing on behalf of a patient who lacks capacity:

Signature: ____________________________________________

Date: ________________

Print Name: ________________________________

Relationship to Person: ________________________________

DO NOT RETURN THIS CONSENT FORM TO NTD LABS, ViaCord or PerkinElmer Genetics. This consent form is provided as a convenience for the ordering physician. It is the physician's responsibility to obtain the proper form of consent from the patient or the person who is authorized to act for the patient when the patient lacks capacity. Please note that Nebraska has a model form of informed consent for genetic testing.
## Disorders Included in the Newborn Screening Panel

### Acylcarnitine Profile
- Fatty Acid Oxidation Disorders
  - Carnitine/Acylcarnitine Translocase Deficiency
  - Carnitine Palmitoyltransferase Deficiency Type I
  - 3-Hydroxy Long Chain Acyl-CoA Dehydrogenase Deficiency
  - 2,4-Dienoyl-CoA Reductase Deficiency
  - Medium Chain Acyl-CoA Dehydrogenase Deficiency
  - Multiple Acyl-CoA Dehydrogenase Deficiency
  - Neonatal Carnitine Palmitoyltransferase Deficiency Type II
  - Short Chain Acyl-CoA Dehydrogenase Deficiency
  - Short Chain Hydroxy Acyl-CoA Dehydrogenase Deficiency
  - Tri functional Protein Deficiency
  - Very Long Chain Acyl-CoA Dehydrogenase Deficiency

### Organic Acid Disorders
- 3-Hydroxy-3-Methylglutaryl-CoA Lyase Deficiency
- Glutaric Acidemia Type I
- Isobutyryl-CoA Dehydrogenase Deficiency
- Isovaleric Acidemia
- 2-Methylbutyl-CoA Dehydrogenase Deficiency
- 3-Methylcrotonyl-CoA Carboxylase Deficiency
- 3-Methylglutaconyl-CoA Hydratase Deficiency
- Methylmalonic Acidemia
  - Methylmalonyl-CoA Mutase Deficiency
  - Some Adenosylhomocystein Synthesis Defects
  - Maternal Vitamin B12 Deficiency
- Mitochondrial Acetoacetyl-CoA Thiolase Deficiency
- Propanoic Acidemia
- Multiple CoA Carboxylase Deficiency
- Malonic Aciduria

### Amino Acid Profile
- Amino Acid Disorders
  - Arginemia
  - Argininosuccinic Aciduria
  - 5-Oxoprolinuria
  - Carnitine/Phosphate Translocase Deficiency
  - Citrullinemia
  - Homocystinuria
  - Hyperornithinemia
  - Hyperornithinemia, Homocitrullinuria Syndrome
  - Hyperornithinemia with Gyril Atrophy
  - Maple Syrup Urine Disease
  - Phenylketonuria
    - Classical
    - Hyperphenylalaninemia
    - Biopterin Cofactor Deficiencies
  - Tyrosinemia
    - Transient Neonatal Tyrosinemia
    - Tyrosinemia Type I
    - Tyrosinemia Type II
    - Tyrosinemia Type III

### Other Observations
- Hyperalimentation
- Liver Disease
- Medium Chain Triglyceride Oil Administration
- Presence of EDTA Anticoagulants in blood specimen
- Treatment with Benzoate, Pyruvic Acid, or Valproic Acid
- Carnitine Uptake Deficiency

### Disorders Detected by Other Technologies
- Biotinidase Deficiency
  - Complete Deficiency
  - Partial Deficiency
- Glucose-6-Phosphate Dehydrogenase Deficiency
- Congenital Adrenal Hyperplasia
  - Salt Wasting 21-hydroxylase Deficiency
  - Simple Virilizing 21-hydroxylase Deficiency
  - Cystic Fibrosis (not valid after 3 months of age)*

### New Additions to the Panel
1. Genetic Hearing Loss (Connexin 26, Connexin 30, certain Mitochondrial mutations, Pendred syndrome mutations, congenital CMV)
2. Severe Combined Immunodeficiency (SCID)

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*The analyses conducted by PerkinElmer Genetics produce results that can be used by qualified physicians in the diagnosis of disorders described herein. Evidence of these conditions can be detected in the vast majority of affected individuals; however, due to genetic variability, age of patient at time of specimen collection, quality of specimen, health status of the patient, and other variables, such conditions may not be detected in all affected patients.

There is a lower probability of detection of this condition during the immediate newborn period.

*For information on DNA Carrier Testing for children over 3 months of age, please call 866.403.8430.

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