Courtagen
DIAGNOSTICS LABORATORY

Patient Informed Consent For Genetic Testing

***REQUIRED**

*Name of person being tested**:_____

*Date of Birth of person being tested**:_____

*Gender of person being tested**: _____

I understand that I/my child am/is submitting a sample for the purpose of genetic testing to Courtagen Diagnostics Laboratory ("Courtagen") as ordered by my physician. I have discussed the benefits, risks, and limitations of this testing with my healthcare provider, and have had my questions answered. By signing below, I give consent to have my/my child's DNA sample and relevant clinical information sent to Courtagen for testing. I also consent to relevant follow-up testing ordered by my physician and understand that this consent form will expire one year after its receipt.

FOR PARENTAL / FAMILIAL TESTING ONLY

If the individual above is being tested as part of additional parental/familial testing, does he/she experience (or has he/she previously experienced) similar symptoms to the original patient (proband)?

☐ No, individual is unaffected ☐ Yes (If yes, please explain)

_____

if additional relatives are similarly affected, please attach a pedigree/family history

SIGNATURE

My signature below acknowledges that I give my consent for genetic testing and I understand the medical and financial outcomes, including the benefits, risks, and limitations, as described above.

*Patient signature or parent/guardian (Print and sign)**

Email

Patient phone number

Address

City/State/ZIP

Date of collection

Physician name

Where to send completed forms:
If you have received this form with your sample collection kit, you MUST return this form with your sample in the box provided. A photocopy of the signed form is acceptable.
Via secure fax: 617-892-7192
In order to expedite the insurance claims process please include a photocopy of both sides of your insurance card. If you have questions, please feel free to contact us at 877-395-7608 or 617-714-0315

ABOUT COURTAGEN'S NEXT-GENERATION GENETIC TEST

The purpose of this test is to identify the underlying genetic basis of my/my child's symptoms. Next-Generation Sequencing is a method of detecting changes in an individual's DNA sequence that cause or are related to his/her medical concerns. This test sequences or "proof-reads" genes, and looks for variants or "misspellings". Because genes are the instructions for how our bodies function and how the proteins in our body are made, variants in the genes can lead to proteins that do not work correctly and cause disease. Variants detected by Next Generation Sequencing may also be confirmed using a second method of genetic testing, such as Sanger sequencing (variants on reports for patients in New York State will be confirmed with Sanger sequencing). This test is not the only way to look for genetic changes, and my healthcare provider may recommend this test before or after other genetic or laboratory tests.

POTENTIAL TEST RESULTS

Results are examined by a team of PhD and MD geneticists, who determine if any variants are likely related to my/my child's symptoms. A written report that discusses each potentially disease-related variant will be issued to my ordering physician. Possible test results are as follows:

1. Negative – No variants identified of suspected disease association
2. Likely negative – Variants identified are likely not associated with disease
3. Uncertain – Variants identified are of uncertain significance
4. Likely positive – Variants identified are likely associated with disease
5. Candidate – Variants identified are plausible candidates for disease
6. Positive – Variants identified are associated with disease

In addition to the results above, the results of this test may be suggestive of, or reveal a predisposition to, a condition other than the diagnosis in question. For example, a variant may be identified in a gene that, while related to the condition being tested for, also increases an individual's cancer risk or is associated with a neurological disease. These findings are known as "incidental findings" or "secondary findings". Please see "INCIDENTAL FINDINGS AND OPT-OUT" section for additional information on these findings and how to opt-out of receiving this information.

LIMITATIONS OF THIS TEST

A negative test result does not rule out all genetic causes of disease. It is still possible that my/my child's symptoms have an underlying genetic cause that this technology is unable to detect or that they are caused by one or more genes that are not part of this specific gene panel.

Some results are of uncertain clinical significance meaning that while they may be related to disease, they may also be harmless changes or unrelated to disease.

A positive result will not necessarily predict the prognosis or severity of disease. Likewise, while identifying a variant of clinical significance may help direct management and treatment, it is also possible that a positive result will not affect management or treatment.

This test is not meant to evaluate for carrier status related to reproduction. Most individuals are apparent carriers for more than one of the genes sequenced on this panel, and with rare exceptions we do not pursue or confirm this data. Furthermore, this Next-Generation sequencing assay as used has poor sensitivity to detect large gene deletions or duplications

(i.e. copy number variation or "CNV").

GENETIC COUNSELING AND TESTING IN OTHER FAMILY MEMBERS

Genetic counseling is the process of providing individuals and families with information on the nature, inheritance, and implications of genetic disorders to help them make informed medical and personal decisions. Courtagen is available to offer suggestions for such providers in your area should you wish to see one. The interpretation of a genetic test result may change depending on whether or not the genetic change(s) identified are also present in parents and/or other family members. For this reason, Courtagen may recommend genetic testing in other family members. Courtagen may provide parental testing for select variants to aid in variant classification. If other family members have the same test, the results may suggest previously unrecognized or unknown biological relationships, such as non-paternity. In the event of a family member receiving a variant confirmation test, Courtagen will amend the proband's (first tested patient) report using the relationship of those family members tested as identifiers. We will note any changes made to the report based on these results for the physician. The amended report will be delivered to the ordering physician of the proband. Individual reports will not be issued for family members tested.

RISKS OF DISCRIMINATION

The Genetic Information Nondiscrimination Act (GINA) of 2008 prohibits health insurance plans and employers from discrimination based on genetic information, including the results of genetic testing. However, such genetic testing may result in life insurance, disability insurance and/or long-term care insurance discrimination that is not prohibited by law.

DNA SAMPLE AND RESULTS DISCLOSURE

I authorize Courtagen to release the test results to the ordering physician and any other health care provider that I designate in writing. Test results may be released to any entity that, by statute of law, has the legal authority to request and receive genetic test results.

I understand that DNA samples will be used for testing that is authorized by an ordering physician and any subsequent related or follow-up testing ordered by the physician. Any leftover sample of genomic DNA will stored for two (2) years at which point it will be discarded. Samples from New York residents will be discarded 60 days after testing unless the individual opts in by signing below. Some samples may be maintained indefinitely after all testing has been completed for use in clinical tests and/or for research purposes to advance scientific knowledge. In such a case, all personal identifiers will first be removed, and I will not be informed of any results, as Courtagen will have no means to determine the sample origin. In addition, I understand that Courtagen may discuss summaries of genetic test results in scientific presentations, publications, or marketing pieces given that no names or personal identifiers are revealed. Unless opted in below, residents of the state of New York will be excluded from sample retention beyond 60 days and from allowing Courtagen to use their genomic data for research purposes.

OPT-IN REQUIRED FOR NY RESIDENTS (SIGN HERE)

_____

FINANCIAL AND INSURANCE CONSENTS

I consent to an insurance pre-authorization. I authorize Courtagen or their agent to perform this insurance pre-authorization on my behalf, with the insurance carrier(s) on file with my ordering physician, or for any insurance carrier(s) given to Courtagen separately but specifically for this testing. If my insurance information has recently changed, I will immediately contact Courtagen to ensure they have the correct information on file. I understand Courtagen will not begin processing of my DNA sample without completion of a pre-authorization.

I consent to a release of information to pay benefits. I authorize Courtagen to provide my insurance carrier with all appropriate information concerning my laboratory test(s). I understand I am responsible for all charges that result due to this genetic testing. I will pay to Courtagen any amounts due within 30 days. In the event that my insurance carrier issues a payment directly to me for this genetic testing, I will remit those funds directly to Courtagen within 30 days. Cancellations of this test must be made in writing by the ordering physician and are subject to Courtagen's financial policy, which can be found at www.courtagen.com.

I consent to appeals of insurance claims denials when necessary. In the event of a significant underpayment or denial of coverage by my insurance carrier, I authorize Courtagen or their agent to submit an appeal to my insurance carrier on my behalf. I also authorize Courtagen to perform the actions or provide the information necessary to overturn a denial or receive reimbursement on the insurance claim. This authorization shall remain valid until the charges for this genetic testing are paid in full or otherwise settled.

INCIDENTAL FINDINGS AND OPT-OUT

Many genes have multiple functions and mutations in these genes may be associated with more than one disease or medical condition. While the genes on our panels are selected based on function and disease association, there is a possibility that these Next-Generation (Next-Gen) Sequencing multi-gene panels will identify a variant that is associated with a disease not directly related to the reason for ordering the test. These variants are called "incidental findings" or "secondary findings" and may reveal information that is not expected.

The American College of Medical Genetics (ACMG) recommends that incidental findings in 57 genes associated with inherited disease be reported for patients undergoing genetic testing. Courtagen's Next-Gen Sequencing panels include18 of the ACMG recommended 57 genes. These genes, in addition to having possible clinical implications for the disease I am/my child is seeking testing for, may also be associated with additional medical conditions that may affect medical management. For example: *RYR1* and *CACNA1S* are associated with mitochondrial function, but additionally predispose towards a potentially fatal reaction to anesthesia, malignant hyperthermia, for which precautions at the time of anesthesia administration are indicated.

REQUEST TO OPT-OUT OF RECEIVING INCIDENTAL FINDINGS:

I do NOT wish to receive results that are not directly related to the reason for which my physician is ordering this Next-Gen Sequencing panel.

Print Name: _____

Signature:_____ Date:_____

REQUEST TO OPT-OUT OF BEING CONTACTED BY OUR CLINICAL TEAM:

Courtagen may need to obtain additional clinical information about you or your child to provide the best possible genetic interpretation. Sign here if you do NOT want to be contacted directly by our clinical team and would prefer that Courtagen contact your physician instead.

Print Name: _____

Signature:_____ Date:_____