

## PATIENT INFORMED CONSENT AND AUTHORIZATION FORM FOR MOLECULAR GENETIC TESTING - OSTEOGENESIS IMPERFECTA

I \_\_\_\_\_ (patient name) \_\_\_\_\_ (Date of Birth: MM-DD-YYYY) request DNA testing for \_\_\_\_\_ (disease).

I request and authorize Connective Tissue Gene Tests (CTGT) to conduct DNA testing for my (or my child or fetus) sample for the above mentioned condition(s) as ordered by my physician / health care provider.

### Description of the Disorder:

Osteogenesis imperfecta (OI), also known as brittle bone disease is a rare inherited bone fragility disorder. It is a genetically and clinically heterogeneous disease. Defects in multiple different genes have been shown to cause OI. Clinical findings include low bone mass, fractures of the long bones following minimal trauma, vertebral compression fractures, blue sclera, hearing loss, and abnormal teeth. The findings vary from mild to severe. Patients with milder forms may have only a few fractures in a lifetime. Severe forms are characterized by multiple fractures, bone deformities, or death in the perinatal period. OI genetic test is useful for confirming a clinical diagnosis of inherited brittle bone disease.

### Description and Principle of the Test:

A biological specimen will be collected from you (or your child/fetus) for DNA isolation. DNA will be used for molecular genetic testing. The purpose of the genetic test is to look for changes in your DNA that will cause the disease you are being tested for.

### Test Results:

- The test is positive if a mutation(s) (a disease causing change) is identified in your DNA. A positive result may indicate that you have a genetic disease or you are at increased risk to develop the disease later in life.
- The test is negative if a mutation is not found. However, mutations are not always detected (see Limitations and Risks below). DNA based testing is confirmatory. A negative test result reduces but does not eliminate the likelihood of a disease.
- The test may also identify a variant that has an uncertain clinical significance. This means there is not enough information to reliably interpret if the variant is associated with a genetic disease.

### Limitations and Risks:

- Although DNA testing is highly sensitive, mutation detection may not be 100 % due to limitations in technology and incomplete knowledge of genes. Some other reasons for not detecting a mutation may include mislabeling a sample, only a small proportion of cells in your body have the mutation (mosaicism), a bone marrow transplantation, a recent blood transfusion, or a small possibility of technical errors.
- An error in a patient's clinical diagnosis can lead to incorrect interpretation of the test result.
- It is possible that DNA testing identifies a variant that is suggestive of a different disease than was initially considered.
- It is possible that the test may reveal non-paternity (the father of an individual is not the biological father) or consanguinity (the parents of an individual are related by blood).
- New information, when available in the future, may add to or change the interpretation of the results. CTGT does not and it is not obligated to routinely re-analyze test results. Physicians / health care providers may monitor the availability of new information relevant to the disease.

### Reporting Results and Confidentiality:

Because of the complexity of DNA testing and the possible impact of the test results, results will be reported directly to the physician / health care provider, who ordered the test. Patient results and information are confidential and they may only be released to the ordering physician / health care provider. Results are only released to other parties with your written consent or as allowed by law.

### Genetic Counseling:

Genetic counseling is recommended before and after DNA testing. Further DNA testing or additional physician consults may be warranted.

### Sample Retention:

- Connective Tissue Gene Tests does not return DNA samples. Connective Tissue Gene Tests is not a DNA banking facility and the sample may not be available for future clinical testing. Any requests for additional DNA testing must be ordered by your physician / health care provider. After completion of a DNA test, de-identified sample and test result may be used for internal quality assurance, educational, test validation, and research purposes. Such samples are not available for any future clinical studies. It is possible that research leads to new discoveries that may potentially have commercial value. Such discoveries are owned by Connective Tissue Gene Tests. Test sample donors do not have any rights to the discoveries and will not gain any financial benefits from the discoveries.
- Participation in a research study is voluntary. Please initial here if you do not want to participate in any research studies: \_\_\_\_\_
- New York residents: All biological specimens and isolated DNA samples from New York will be retained for 60 days.

