

Informed Consent for *CASR* Gene Sequencing

Patient's Name: _____

Patient's ID Number: _____

Patient's Date of Birth: _____

Ordering Provider's Name: _____

Note to the Ordering Health Care Provider: Some states require that patients (or their authorized representatives) provide their written informed consent prior to receiving genetic testing, and that the ordering health care provider maintain documentation of the informed consent in the patient's medical record. This form is intended to assist you with obtaining the patient's informed consent in accordance with applicable law.

- **Reasons for Medical Referral** — The calcium-sensing receptor, CaSR, regulates parathyroid hormone secretion and calcium release from bone and excretion from the kidney to maintain serum calcium at an optimal level. Individuals with variants (mutations) in the *CASR* gene can have dysregulated serum calcium levels, with symptoms ranging from benign to severe. Testing for *CASR* gene mutations can be useful to diagnose CaSR defects, and testing other appropriate members of your family will help determine those individuals' *CASR* status. The identification of *CASR* mutation(s) may be helpful in predicting the need for parathyroid surgery, the risk of kidney stones, and the response to treatment with calcium-related therapies.
 - **General description and purpose of the test** — A blood sample is taken from you. This may cause you some pain and discomfort. The blood sample is sent to the laboratory and DNA is purified from your blood sample. Your DNA is analyzed to look for mutations in selected regions of the *CASR* gene.
 - **Limitations of the test** — This test analyzes selected regions of the *CASR* gene where the vast majority of activating and inactivating mutations are located. Because all regions are not tested, the rare possibility cannot be ruled out that you have a *CASR* gene mutation in a region not analyzed. You could also have a rare variant that may interfere with the detection of a *CASR* gene mutation. Donor DNA from transplants and recent transfusions can lead to inaccurate results. For familial mutation studies, accurate interpretation of test results will depend upon the accuracy of the diagnosis of CaSR functional deficiency or CaSR activation in your family and of the family relationships; cases of adoption or non-paternity, if not specified, can result in erroneous interpretations.
 - **Availability of genetic counseling before and after testing** — I have been provided with information about obtaining genetic counseling prior to giving my consent for this testing. I further understand that my health care provider may recommend consultation with a medical geneticist, genetic counselor, and/or a physician after the testing is completed.
 - **Meaning of a positive test result** — A positive test result indicates the presence of a clinically important, or possibly clinically important, variation in your *CASR* gene. If available, medical literature publications are cited to provide clinical significance of the variation with respect to CaSR function. If literature citations are not available, clinical significance may be unknown. The presence of a *CASR* gene mutation is not completely deterministic; some individuals with a *CASR* gene mutation may not develop clinically significant hypercalcemia or hypocalcemia.
 - **Meaning of a negative test result** — A negative test result indicates that no clinically important variation was detected in the targeted regions of the *CASR* gene. A negative test result does not rule out the possibility of an undetected variant in a region of the *CASR* gene not analyzed by this test. It is also possible that a rare false negative result can be obtained due to the presence of a variant in the *CASR* gene that interferes with this assay. Also, hyperparathyroidism or hypoparathyroidism can occur due to mutations in other genes. Thus, a negative result does not mean that you will not develop any symptoms associated with CaSR alterations. You will at least have the risk of the general population for developing these conditions.
 - **Disclosure of test results** — Stringent laboratory processes are in place to keep your personal information and your *CASR* gene test results strictly confidential. Only the *CASR* Gene Sequencing test will be performed on your specimen. The *CASR* Gene Sequencing test results will be released only to your physician or to the referring institution.
 - **Retention of specimens** — Endocrine Sciences may retain your blood and/or DNA specimens for up to 60 days after completion of testing.
- I have read the information provided above and I have discussed the *CASR* Gene Sequencing test with my physician/healthcare provider. I have had the opportunity to ask any questions regarding this test, and all questions have been answered to my satisfaction. In no way does this waive my legal rights or release anyone from their legal and professional responsibilities.
- | | |
|---|---------------|
| _____
Signature of Patient or
Patient's Authorized Representative | _____
Date |
| _____
Relationship to Patient
(If the Patient's Authorized Representative) | _____
Date |
| I have counseled Mr/Ms _____
regarding genetic testing and have obtained his/her or the legal
guardian's informed consent for the <i>CASR</i> Gene Sequencing test. | |
| _____
Signature of Ordering Health Care Provider | _____
Date |