



TECHNICAL NOTICE

SOUTH BEND MEDICAL FOUNDATION

February 2009

Angiotensin Converting Enzyme (ACE) Genotyping by RT-PCR

Effective Date: February 1, 2009

Performing Department: Molecular Pathology

Method: Real -Time Polymerase Chain Reaction (RT-PCR)

Use: Genotyping (insertion/deletion polymorphism) of ACE gene located in intron 16 on chromosome 17

Clinical Significance and Intended Use:

Angiotensin-converting enzyme (ACE) is a peptidyl dipeptide hydrolase that plays a role in the conversion of angiotensin I to angiotensin II and inactivates the vasodilator bradykinin in the renin-angiotensin system. The levels of plasma and cellular ACE are strongly determined genetically. Several studies have shown that 50% of the variability of plasma ACE among individuals is the result of an insertion/deletion (I/D) polymorphism in intron 16 of the ACE gene on chromosome 17. The presence or absence of a 287 bp Alu sequence DNA fragment results in I or D polymorphism, respectively. Presence of D allele has been reported to be associated with higher circulating ACE levels and several human disorders, such as hypertension, coronary heart disease and type II diabetes mellitus.

Reference Range: ACE genotype: I/I*

*Patient has 2 copies of insertion variants (I/I) in intron 16 of the ACE gene on chromosome 17

Specimen Requirements and Collection:

Preferred Specimen: • Whole blood in lavender top (EDTA) tube

Requested Volume: • 5.0 mL

Minimum Volume: • 1.0 mL

Processing: • Do not centrifuge • Do not remove plasma from cells • Do not freeze

Stability: • 1 week refrigerated (2-8°C)

Storage/Transport: • Refrigerated

Causes For Rejection: • Clotted, severely hemolyzed or frozen sample

Testing Schedule: • Samples received by Monday are reported by Friday • Occasional reports may take up to 2 weeks

Order: • Angiotensin Converting Enzyme Genotyping, by RT-PCR Test #: **36151**

CPT: • 83891 • 83896 x 2 • 83898

Additional Information:

This test was developed and its performance was established and confirmed by the SBMF. This test is not cleared or approved by the U.S. FDA. This test is used for clinical purposes and should not be regarded as investigational or for research. The South Bend Medical Foundation is authorized under Clinical Laboratory Improvement Amendments (CLIA) to perform high-complexity testing. This test is performed pursuant to an agreement with Roche Molecular Systems, Inc.

Please direct questions or comments regarding this notice to Bobbie C. Sutton, M.D., Deborah H. Sun, Ph.D., or Sally Cornwall of South Bend Medical Foundation, (574) 234-4176 or (800) 544-0925.

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