
NOTICE DATE: September 28, 2016

EFFECTIVE DATE: September 23, 2016

TEST RESUMED

Pecan, IgE

Order Code: PCAN
Fee Code: 21402

Pecan, IgE allergy testing has resumed effective September 23, 2016.

EFFECTIVE DATE: September 22, 2016

NEW TESTS

GALC Gene Sequencing

Order Code: GALCS
CPT Code: 81406, G0452-26

GALC Deletion / Duplication Analysis

Order Code: GALCD
CPT Code: 81405, G0452-26

The MLabs Molecular Genetics Laboratory (MMGL) began offering full gene sequencing and deletion/duplication analysis of the GALC gene (OMIM: 606890) effective September 22, 2016.

The GALC gene is located on chromosome 14 at 14q31.3 and encodes a lysosomal enzyme (Galactosylceramidase or Galactocerebrosidase) which is essential for myelin turnover. Krabbe disease (also known as globoid cell leukodystrophy; OMIM 245200) is an autosomal recessive lysosomal storage disorder caused by deficiency of galactocerebrosidase (GALC) enzyme. Homozygous or compound heterozygous pathogenic variants in the GALC gene have been reported in patients with Krabbe disease. Infantile Krabbe disease is characterized by infantile-onset progressive neurologic deterioration and death before age two years (85%-90% of individuals), while Late-onset form is characterized by onset between age one year and the fifth decade with slower disease progression (10%-15%). Children with the infantile form appear to be normal for the first few months of life but show extreme irritability, spasticity, and developmental delay before age six months; psychomotor regression progresses to a decerebrate state with no voluntary movement. The onset and progression in the late-onset form can be quite variable. Individuals can be clinically normal until weakness, vision loss, and intellectual regression become evident. The onset of symptoms and clinical course can be variable even among siblings (GeneReviews, <http://www.ncbi.nlm.nih.gov/books/NBK1238/>). Full gene sequencing analysis of GALC will detect at least 50% of pathogenic variants, while the remaining pathogenic variants will be detected by deletion/duplication analysis.

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REMINDER: SPECIMENS REQUIRE 2 IDENTIFIERS

MLabs policy requires that ALL specimens, including slides, are labeled with both the patient's FIRST and LAST NAMES as well as a SECOND IDENTIFIER such as the patient's medical record number or date of birth. Acceptable second identifiers include, but are not limited to: date of birth, social security number, medical record number, accession number, requisition number, or specimen barcode label. Specimens without proper labeling including two person-specific identifiers will not be tested and MLabs will recommend that a new specimen be obtained if possible.