

Lab Updates

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April 2010

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Genetic Testing

Human genetics is often referred to as the “basic science” of medicine, and questions about inherited traits go back to the beginnings of civilization. The central theme to many questions about genetics are reflected in the fact that offspring of a species are always very similar, yet not identical to their parents—there always being some variation. Importantly, it is this variation that comprises the mechanisms leading to human individuality. Modern genetics is focused on identifying those processes that both normally and in disease lead to individual characteristics.

Diagnostic test – a test performed to determine the presence or absence of a specific medical condition. Molecular tests are used currently as an aid in evaluation of patients suspected of/with infectious diseases, genetic disorders and other disorders where are established known genetic risk factors. Also in the last few years pharmacogenetic testing evolved creating personalized approached to drug choices and dosing based on individual’s variants.

Genetic disorders – inherited conditions caused by an absent or defective gene (molecular genetic chapter) or by a chromosomal aberration. Genetic disorders can be tested on the level of DNA, RNA or protein. Testing for genetic risk factors and testing for infectious diseases in appropriate chapters.



Photo: Kevin Vance

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Detection and Monitoring Infectious Diseases

Sample should be collected before starting treatment.

Qualitative – **detection** of the presence of viral particles or **confirmation** of positive viral antibody test; reported as “positive” or “negative”; highly sensitive low limit of detection.

Quantitative – measurement of the amount of virus to **monitor** the effectiveness of a treatment (copies/ml, IU/ml, log).

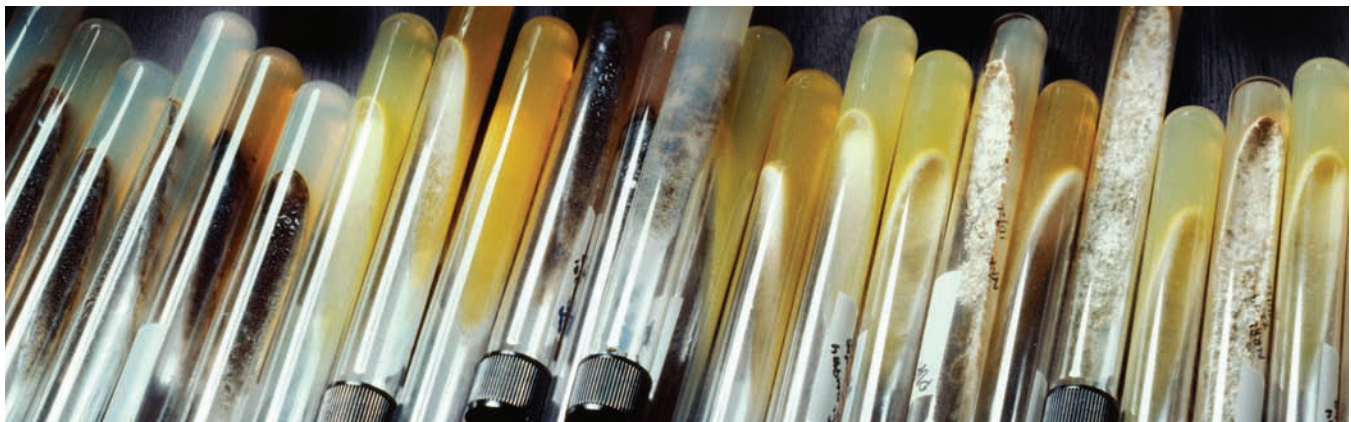
Genotyping – determination of the viral type or subtype when considering antiviral therapy. Genotype testing is available and is useful in treatment planning and for determining length and possible responses to treatment. Genotype testing should be done as part of the patient’s initial evaluation once infection has been confirmed. May aid in identifying source of infection.

High sensitivity of molecular assays allows on early detection of infection when other markers are negative, detection of infection in immunocompromise patients (antibodies negative) and in addition to monitoring patient’s response to therapy, molecular test will be negative before antibodies negative.

Molecular tests allow for high specificity of the tests by using conserve regions of genomic sequence of organisms species and subspecies.

Genetic Disorders

A genetic test is the analysis of human DNA, RNA, chromosomes, proteins, or certain metabolites in order to detect alterations related to a heritable disorder. This can be accomplished by directly examining the DNA or RNA that makes up a gene (direct testing), looking at markers co-inherited with a disease-causing gene (linkage testing), assaying certain metabolites (biochemical testing), or examining the chromosomes (cytogenetic testing) (www.genetests.org). The results of a genetic test can confirm or rule out a suspected genetic condition, determine person’s risk of developing disorder, identify carriers or assess variants influencing individual’s rate in drug metabolism. Several hundred genetic tests are currently in use, and more are being developed.



Genetic testing may be undertaken as part of the process of treating or advising an individual patient.

Forms of genetic testing:

Diagnostic Genetic Testing – confirmatory test for symptomatic individual.

Presymptomatic Genetic Testing – carried out in healthy people without symptoms for estimating risk of developing disease (e.g. Huntington's disease, Fabry Disease in women).

Carrier Testing – carrier testing is performed to determine whether an individual carries one copy of an altered gene for a particular recessive disease. Recessive diseases occur only if an individual receives two copies of a gene that have a disease-associated mutation; thus, each child born to two carriers of a mutation in the same gene has a 25-percent risk of being affected with the disorder.

Risk Factor Testing (susceptibility tests) – gene variants have been discovered which are associated with common diseases such as Alzheimer's disease and diabetes.

Pharmacogenetic Testing – determining differences in individual reactions to drugs

Pre-implantation Testing – pre-implantation diagnosis is used following in vitro fertilization to diagnose a genetic disease or condition in a preimplantation embryo.

Pre-natal Testing is used to diagnose a genetic disease or condition in a developing fetus.

Newborn Screening is performed in newborns in state public health programs to detect certain genetic diseases for which early diagnosis and treatment are available.

If you have questions, comments or suggestions, please contact:

Dr. Marzena Galdzicka at 508-856-4384 or via email at Marzena.Galdzicka@umassmed.edu



Changes in Specimen Labeling Requirements:

Two Person-Specific Identifiers are Required on Primary Specimen Containers

In our continuing effort to improve quality and in compliance with the College of American Pathologists (CAP) and the Joint Commission's 2009 National Patient Safety goals (1A), UMass Memorial Laboratories policy states that all specimens received for testing must be correctly and adequately labeled to assure positive identification. **Primary specimen containers must be labeled with two person-specific identifiers.** Person-specific identifiers may include patient's first and last names, date of birth, or unique identifying number (e.g., medical record number or accession number).

UMass Memorial Laboratories uses multiple patient identifiers to verify the correct patient is matched with the correct specimen and the correct order for testing services. As a specimen is received at UMass Memorial Laboratories, patient's first and last names and date of birth (alternatively medical record number or accession number may be used as person-specific identifiers) are verified by comparing the labels on the specimen tube or container with the electronic order and any paperwork (requisition form or manifest) that may accompany the



specimen to be tested. In order to use the patient's name as one form of identification, the patient's full first and last names must be indicated. Specimens are considered mislabeled when there is a discrepancy between the person-specific identifiers on the specimen and information accompanying the specimen (e.g., computer system, requisition form, additional paperwork). When discrepancies are identified, the UMass Memorial Medical Center customer service will telephone the client to verify discrepant information to assure UMass Memorial Laboratories are performing the correct testing for the correct patient.

If you have questions, comments or suggestions, please contact:

Dr. Nichole Korpi-Steiner at 774-442-9634 or via email at Nichole.Korpi-Steiner@umassmemorial.org

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- Frozen
- Refrigerate
- Room Temp

Fisherbrand® SPECIMEN BAG

STAT

BIOHAZARD

- Frozen
- Refrigerate
- Room Temp

SPECIMEN BAG

STAT

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BIOHAZARD

- ROOM TEMPERATURE
- REFRIGERATE
- FROZEN

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FROZEN

- 1) Insert Specimen in Longer Pouch.
- 2) Pull Off Liner, Press to Close.
- 3) Insert Requisition into Outside Pocket.
- 4) Tuck Top of Requisition Under Flap.

Changes in BNP and ACTH Specimen Collection Requirements

Clinically, BNP measurements are useful as an aid to diagnose acute heart failure in symptomatic patients presenting to an emergency department, stage severity and prognosis of congestive heart failure patients and to risk stratify patients with acute coronary syndromes. ACTH is very important in the diagnosis of Addison's disease, congenital adrenal hyperplasia, and Cushing's syndrome.

There is sparse and conflicting data regarding the stability of BNP and ACTH in the specimen container following blood collection. Proteolytic degradation of these molecules appears to occur as soon as blood is collected.

Based on internal validation studies as well as available published literature, the following changes will be made to the test specimen collection requirements to ensure accurate test results.

For both assays, EDTA plasma is the only suitable specimen. Following specimen collection, immediate centrifugation of the specimen with transfer of the plasma into a plastic pour off tube is required. Frozen transport of the specimen to the laboratory is also required. Separate samples must be submitted when multiple tests are ordered such as CBC and Hemoglobin A1c along with BNP or ACTH. Do not combine the test requests to one tube. BNP and ACTH test requests will be canceled if the specimen collection requirements and transport conditions are not followed.

If you have questions, comments or suggestions, please contact:
Dr. L.V. Rao, Director of Core Laboratories at 774-442-9615 or via email at Lokinendi.Rao@umassmemorial.org

B-Natriuretic Peptide (BNPX)

Methodology: Chemiluminescent Immunoassay
Performed: Daily
Reported: Same Day
CPT: 83880

Collect: Lavender (EDTA)
Transport: Frozen
Min. Volume: 1.0 mL Frozen plasma

Notes: Centrifuge specimen immediately, transfer plasma to a plastic pour-off tube using a plastic pipette and label "Plasma". Separate samples must be submitted when multiple tests are ordered.



Adrenocorticotrophic Hormone (ACTH)

Methodology: Chemiluminescent Immunoassay
Performed: Sun-Fri
Reported: Same Day
CPT: 82024

Collect: Lavender (EDTA)
Transport: Frozen
Min. Volume: 1.0 mL Frozen plasma

Notes: Centrifuge specimen immediately, transfer plasma to a plastic pour-off tube using a plastic pipette and label "Plasma". Separate samples must be submitted when multiple tests are ordered.



National Medical Laboratory Professionals Week

April 18 – April 24, 2010

The week of April 18 through April 24, 2010
has been designated as
National Laboratory Professionals Week.

The management team at
UMass Memorial Laboratories
wishes to recognize and thank
the more than 600 laboratory professionals
who work for our laboratories
and whose dedicated efforts enable us
to provide high-quality laboratory testing
and lab-support services to meet the needs
of over 5,000 health care providers
and their patients throughout New England.

We also wish to thank our laboratory clients
for the opportunity to provide
laboratory services for your organization,
your clinical staff, and your patients.



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We are one of the largest laboratory providers in New England

UMass Memorial Laboratories has opened a Patient Service Center (phlebotomy draw station) at 109 Beechwood Avenue, Pawtucket, Rhode Island.

The vision of UMass Memorial Laboratories is:

- To be a leading provider of laboratory services throughout New England, meeting the needs of patients and providers in the region, and
- To be one of the top ten academic medical center-based laboratories in the United States



Pawtucket PSC **109 Beechwood Avenue** **Pawtucket, Rhode Island**

Pawtucket PSC is located at 109 Beechwood Ave., Pawtucket, Rhode Island. The hours are Monday through Friday 8:30am-5:00pm, closed 12:00pm-1:00pm. The phone number at Pawtucket PSC is 401-729-7550.