



## Microbiology vs. Molecular Testing

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Microbiology testing is important in diagnosing and treating disease in patients based upon the presence of a, or multiple, micro-organism(s).

Molecular testing is important in predicting, diagnosing and treating diseases or disabilities related to changes in cells at the molecular level.

What happens when the two cross? When and how do you handle testing and diagnosis when they overlap?

### **Focus on Microbiology testing**

Microbiology testing focuses on micro-organisms such as pathogens, molds, bacteria and yeast. Each of these micro-organisms is a foundation for serious health issues in humans. Some are allergens, others cause severe deterioration or an attack of major organs and can lead to death.

Microscopic organisms can be single cell, a cell colony or acellular. Testing in this field can focus on virology, mycology, parasitology and bacteriology. Pathogens can be from external sources such as the environment, other people, or animals or they can be from internal sources (host-based).

The diagnosis of these types of illnesses requires testing. Many clinical symptoms will present themselves in ways that cannot be distinguished from a variety of viruses or infections. Clinical information about the patient is necessary for the lab to determine which types of diagnostic studies should be performed. Clinical information provided by

the physician that will aid the lab in determining the path that should be taken includes demographic information about the patient, clinical symptoms, onset of symptoms, any treatments already tried, suspected exposures and progression of the symptoms. Different types of specimens also have requirements for their collection, storage and transportation – the lab will inform the physician of these requirements at the time of order.

In short, the Microbiology lab performs tests on specimens that include a microorganism (of varying kinds) from a patient in order to diagnose the type of illness and to provide an interpretation of the results.

### **Focus on Molecular testing**

Molecular testing is used to diagnose, monitor and treat an existing disease, detect risk and map specific treatments to the individual patient based upon their unique biological markers for developing disease and for the risk of passing on any abnormalities to unborn children. In short, Molecular testing is relevant to any diseases or malformations that relate to cells.

There are several sub focuses of molecular and they include:

Genetic testing – identifying changes in genes, chromosomes and/or proteins. Genetic testing or genetic counseling is done to either confirm or rule out a condition whether it is currently present, a person's likelihood of developing it (to include the related risk percentage) and whether a couple may pass it on to a child.

Infectious Disease –many types of infectious disease are diagnosed using molecular testing assays. Examples include: sexually transmitted diseases, healthcare associated diseases, hepatitis, respiratory and central nervous system. Benefits of utilizing molecular testing methodologies for infectious disease diagnosis include rapid detection, new strains, sensitivity, potential outbreaks as well as severity.

Oncology – Testing for gene alterations will diagnose cancer. There are four types of gene alterations these include: Single nucleotide variants, small duplications of nucleotides, whether consecutive, one or multiple or complex mutations, structural variants, or Exon copy number changes. The reason tumor or cell testing is critical is because each type of tumor responds differently to treatment and drug therapies. It is important to identify the specific genetic makeup of the tumor in each individual patient.

Hematology – this type of testing is performed on whole blood, bone marrow and flow separated cells. Testing will provide diagnosis, treatment monitoring, efficacy of treatment and disease progression and prognosis for diseases such as hemophilia, leukemia, von Willebrand disease, thrombosis, and monitoring of graft, bone marrow and stem cell transplantation.

Pharmacology – Molecular testing as it relates to pharmacology helps to determine and support specific treatment plans for disease and individuals. Just as individuals will respond differently to treatment and different drugs, so will tumors. Molecular testing of the individual and how their genes express proteins, and mutations as well as specific tumors and malignant cells will determine everything from drug resistance to drug and treatment efficacy.

### **Both Molecular and Microbiology Testing can Apply to Precision Medicine and Personalized Medicine**

Precision medicine – This discipline is focused on an individual patient and includes all aspects of their life. Areas that are taken into consideration are their specific genetic makeup, environment, behaviors and lifestyle. This is all combined to provide treatment, monitoring and in some cases, prevention of diseases that will have the best outcome for one individual.

Personalized medicine – This discipline puts individuals into categories or groups based upon the above factors. The idea is that a group of similar individuals with similar diseases or illnesses will respond to a set of treatments or drug therapies the same way. Pharmacogenomics falls into this category of medicine. Things such as patient demographics (age, sex, lifestyle, diagnosis) can all predict how any given medication or drug therapy might work for you or may have adverse side effects for you vs. how it would respond in other individuals with a different demographic.

Pharmacogenomics can provide information for each group to help determine things such the risk for side effects, dosage amounts for optimal treatment, if there would be no benefit, the likelihood of benefit from treatment, optimal length of time for the treatment.

### **Do They Cross?**

They do actually. The microbiology lab should be performing molecular testing on infectious diseases.

Many infectious diseases are best diagnosed using molecular testing methodologies to determine the progression of the disease, management of the disease and stage of the disease. The current industry thought and trend on best practices is that the best patient care and outcomes at the most reasonable cost is to follow this model. Many of the assays used in microbiology testing for infectious disease are highly complex in both their development and their application. This is amplified when there is an unusual pathogen, a new pathogen or a mixed infection present.

### **Overlap of Molecular and Microbiology Testing**

Overlapping areas of testing revolve mainly around infectious diseases. With the emergence and growth of Molecular testing and labs who are expanding and incorporating this type of testing into traditional clinical practice areas – there comes the age old question of the chicken and the egg.

Most facilities have to entertain the question of centralization or decentralization of the multidisciplinary test menus. Most of the answer boils down to the ability to achieve efficiencies based upon equipment, expertise, personnel, reagents and location. It comes down to cost, versatility and efficiency – just as any business decision ultimately does.