



MEDICAL DIAGNOSTIC LABORATORIES, L.L.C.

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OB/GYN UPDATE, JUNE 2007

As part of a continual effort to enhance our testing for the OB/GYN market, MDL is delighted to offer the following testing that can now be ordered and performed from the **OneSwab™**:

Test #	New Test
1201	Cystic Fibrosis Gene Carrier Screening by Bio-Plex Analysis
215	Varicella Zoster Virus by Real-Time PCR

Specimen Requirements & Collection Procedures

- Specimen requirements - **OneSwab™** transport vial, stable at room temperature for up to five (5) days.
- The proper specimen collection technique is very important in identifying pathogens from DNA. Therefore, please refer to MDL's collection procedures to assist you in obtaining the best results.

As a premier infectious disease laboratory, MDL continually strives to improve our services by providing the most current, accurate, and informative diagnostic tools available.

If you should have any questions, please contact your sales representative or call MDL at:



Toll Free: (877) 269-0090

Cystic Fibrosis Carrier Screening

Cystic Fibrosis (CF) is an autosomal recessive inheritable disease that afflicts approximately 30,000 people within the United States and 70,000 worldwide, with 1,000 new cases diagnosed each year. Due to its recessive inheritable pattern, people may be carriers of the disease, having inherited a defective gene but not exhibiting symptoms. It is estimated that an additional ten million, or one in every thirty-one Americans, are carriers. Carrier status occurs more frequently within Ashkenazi Jewish and Caucasians of European descent populations, each of which has a one in twenty-nine carrier risk rate. The defective gene responsible for CF was identified in 1989 as the Cystic Fibrosis Transmembrane Conductance Regulator (CFTR) gene. The CFTR protein serves as a chloride channel within epithelial cells; disruption of its function induces an electrolyte imbalance that results in excess sodium chloride levels in sweat, a hallmark and diagnostic indicator of disease, and is believed to cause the thickening of fluids in the lungs and digestive tract. Since its discovery, more than 1,500 mutations have been defined within the CFTR gene. Both the American College of Obstetricians and Gynecologists (ACOG) and the American College of Medical Genetics (ACMG) have developed guidelines for genetic testing to include twenty-three of the most common CFTR mutations. The $\Delta F508$ mutation accounts for approximately two-thirds of all mutant CF alleles worldwide and 70% of the CF cases within the United States, while the W1282X mutation predominates within the Ashkenazi Jewish population. The MDL Cystic Fibrosis Gene Carrier Screening by Bio-Plex Analysis evaluates thirty-two possible point mutations. Reflexive testing for six additional mutations is automatically initiated following positive identification of either the $\Delta F508$ or R117H mutations.

Varicella Zoster Virus

Varicella Zoster Virus (VZV), also known as HHV3, is a member of the neurotrophic alpha herpesvirus family, which is considered to be the most infectious of the human herpes viruses. The alpha herpesviruses possess factors that increase their infectivity, including short reproductive cycles, the ability to replicate in multiple cell types, and the ability to induce high levels of host cell tissue destruction quite rapidly. Humans serve as the alpha herpesvirus only natural reservoir, which means transmission is person-to-person via an airborne route that involves the aerosolization of virus in nasopharyngeal secretions or more directly by contact with vesicle fluids or respiratory secretions. Primary infections result in chickenpox and 95% occur during childhood. Presenting symptoms include rash, low grade fever, headaches and malaise. Patients with chickenpox remain infective until the last skin lesion has dried and crusted over. Those who are infected during adulthood experience a greater number of complications and account for nearly half of all chickenpox-related deaths. Neonates and pregnant women are particularly susceptible to severe primary VZV infections. Complications associated with VZV infection include bacterial superinfections of the skin and lower respiratory tract. Diagnosis is typically based on clinical presentation, but in some instances, particularly immunocompromised individuals, clinical evaluation is necessary. Because VZV is capable of establishing a latent state within the sensory ganglia, infection is life-long and viral reactivation in the form of shingles or Ramsay Hunt Syndrome is possible at any age. Shingles occur in approximately 20% of the adult population at least once in their lives, with 1% experiencing multiple reactivations. Vaccination with the live attenuated Oka strain of VZV, Varivax, is available and recommended for adults over the age of sixty for the prevention of shingles.