

Genetic Carrier Screening

Now Available



- Cervicovaginal or buccal swab
- No refrigeration required before or after collection
- Rapid turnaround time of only 24-48 hours
- Test additions available for up to 30 days
- Specimen viability up to five (5) days

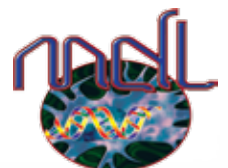
MDL is pleased to announce the availability of genetic carrier testing on the **OneSwab**[®] platform. **OneSwab**[®] technology provides a minimally invasive collection method that provides sufficient sample quantities obtained from a cervicovaginal or buccal swab with a rapid turnaround time of only 24 – 48 hours. Testing is now available for nine commonly inherited disorders included in the 2004 American College of Obstetricians and Gynecologists (ACOG) recommendations for all couples of Ashkenazi Jewish ancestry.

Tests currently available on the **OneSwab**[®]

- 1213 Ashkenazi Jewish Carrier Screening Panel by Bio-Plex Analysis (Canavan Disease, Cystic Fibrosis, Familial Dysautonomia, Tay-Sachs Disease)
- 1214 Ashkenazi Jewish Carrier Screening Expanded Panel by Bio-Plex Analysis (Bloom Syndrome, Canavan Disease, Cystic Fibrosis, Familial Dysautonomia, Fanconi Anemia Type C, Gaucher Disease, Mucopolipidosis Type IV, Niemann-Pick Disease Type A, Tay-Sachs Disease)
- 1207 Bloom Syndrome by Bio-Plex Analysis
- 1209 Canavan Disease by Bio-Plex Analysis
- 1201 Cystic Fibrosis Gene Carrier Screening by Bio-Plex Analysis
- 1210 Familial Dysautonomia by Bio-Plex Analysis
- 1205 Fanconi Anemia Type C by Bio-Plex Analysis
- 1211 Gaucher Disease by Bio-Plex Analysis
- 1203 Huntington's Disease by PCR
- 1212 Mucopolipidosis Type IV by Bio-Plex Analysis
- 1206 Niemann-Pick Disease Type A by Bio-Plex Analysis
- 1208 Tay-Sachs Disease by Bio-Plex Analysis



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1207 Bloom Syndrome by Bio-Plex Analysis

Clinical significance: Bloom syndrome is an extremely rare genetic disorder resulting from various mutations within the Bloom (BLM) gene, a DNA helicase. These mutations, though not lethal, increase the rate of chromosomal instability and the risk of early onset cancers; this predisposition often results in premature death of affected individuals in their twenties or thirties. Chronic lung problems, diabetes mellitus, male sterility and immune deficiencies are also associated with this syndrome.

1209 Canavan Disease by Bio-Plex Analysis

Clinical significance: Canavan Disease is a genetic disorder that occurs with greatest frequency among Jewish individuals of Ashkenazi descent. The disease, a result of various mutations within the aspartoacylase (ASPA) gene, leads to a host of neurological disorders and motor skill deficiencies within affected individuals. The aspartoacylase enzyme is critical to the maintenance and growth of myelin sheaths within the brain. Symptoms begin to emerge within the first nine months of life and are typified by increased head circumference and the lack of head control and visual responsiveness. There is no standard measure of treatment, but a lack of any medical intervention usually results in death by the age of four.

1201 Cystic Fibrosis Gene Carrier Screening by Bio-Plex Analysis

Clinical significance: 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. 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 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.

1210 Familial Dysautonomia by Bio-Plex Analysis

Clinical significance: Familial Dysautonomia is a neurological disorder primarily affecting the autonomic nervous system and, to a lesser extent, the sensory system. Also known as Riley-Day syndrome, neurological abnormalities arise due to mutations within the gene that encodes the IKB kinase complex associated protein (IKBKAP). This protein is critical to the transcriptional process and its defect decreases the number of non-myelinated neurons and also the diameter of those neurons that are myelinated. Over the course of the individual's lifetime, these neuropathies accumulate to affect multiple organs and systems, often dramatically shortening the affected individual's lifespan.

1205 Fanconi Anemia Type C by Bio-Plex Analysis

Clinical significance: Fanconi Anemia Type C is a rare, inheritable blood disorder in which the individual's bone marrow inefficiently produces the three main components of blood: platelets, red blood cells and white blood cells. Multiple organs and systems are affected within these individuals. Affected individuals are also at higher risk of genetic defects and early-onset cancers, about 10% of such individuals develop Acute Myelogenous Leukemias (AML). The major cause of death is bone marrow failure that is typified by thrombocytopenia and neutropenia that transcend into fatal pancytopenia and aplastic anemia.

1211 Gaucher Disease by Bio-Plex Analysis

Clinical significance: Gaucher Disease is the most common lipid storage disease resulting from a deficiency in the glucocerebrosidase enzyme. The inability to breakdown the fatty substance glucocerebroside, or glucosylceramide, allows for its accumulation within many organs and joints, disrupting normal function. There are three forms of the disease, types 1 through 3. Type 1, the most common form, is limited to liver and spleen enlargement, anemia and fatigue with no neurological involvement. Type 3 has a similar symptomology but neurological defects arise over time in the form of seizures. Type 2 is typified by extensive and progressive neurological damage that often leads to death by the age of 2.

1203 Huntington's Disease by Bio-Plex Analysis

Clinical significance: Huntington's disease (HD) is an inherited neurodegenerative disorder characterized by clinically progressive motor impairment, reduction in cognitive abilities and psychopathologic deficits. It is estimated that 1 in every 10,000 people in the United States has HD. Although HD is found in every country of the world, it is more prevalent in people of Western European descent affecting 1 per 10,000 people whereas it affects 1 per 1,000,000 people of Asian or African descent. The MDL Huntington's disease test provides a simple, non-invasive diagnostic test for the trinucleotide repeat expansion status in the Huntington's disease gene *IT-15*.

1212 Mucopolidosis Type IV by Bio-Plex Analysis

Clinical significance: Mucopolidosis Type IV is a lysosomal storage disorder resulting from mutations within the cation channel TRPML1 (Mucopolipin-1), which primarily localizes to endosomes. Mutations within the MCOLN1 gene, which encodes the TRPML1 channel protein, block normal endocytic transport and prevent the formation of lysosomes. Through an as-of-yet unknown mechanism, these mutations lead to severe neurologic and ophthalmologic abnormalities. Affected children display symptoms within the first two years of life and are expected to survive into adulthood but their physical and mental development are typically stunted, advancing only to the level of a one to two year old.

1206 Niemann-Pick Disease Type A by Bio-Plex Analysis

Clinical significance: Niemann-Pick Disease Type A is a lysosomal storage disease that results in a neurologic disorder resulting from the abnormal and toxic accumulation of the cholesterol, sphingomyelin, within cells. The disease can occur as one of four types, A, B, C or D, dependent upon the inheritable mutation. Types A and B occur due to a deficiency of the enzyme acid sphingomyelinase which is necessary for the removal of sphingomyelin, a fat found in all cell types. Type C occurs as a result of the inability to process cholesterol and other lipids leading to their toxic build-up, while Type D occurs due to an inability to move cholesterol among brain cells. Types A, C, and D adversely affect the brain and result in a progressive loss of motor and sensory function.

1208 Tay-Sachs Disease by Bio-Plex Analysis

Clinical significance: Tay-Sachs Disease is a fatal lipid storage disease resulting from the accumulation of toxic levels of the ganglioside lipid within the brain. This accumulation occurs due to the presence of mutations either within the ganglioside protein itself, resulting in its misfolding, or within the critical lysosomal enzyme beta-hexosaminidase A required for its degradation. Development appears normal initially, but mental and physical deterioration occur quickly within the first few months of life as the fat accumulates within nerves; deafness, blindness and an inability to swallow then ensue. There is no effective treatment and death by the age of 4 years is generally a certainty.