HUMAN HEALTH

ENVIRONMENTAL HEALTH

SCREENING FOR LYSOSOMAL STORAGE DISORDERS (LSD)

PerkinElmer Genetics, a technology leader in newborn screening services, now offers screening for six Lysosomal Storage Disorders (LSD). Early detection leading to early intervention has been shown to provide the best chance for a positive outcome. PerkinElmer Genetics uses tandem mass spectrometry (MS/MS) methodology[1] to measure enzyme activity in order to detect infants at risk for having a lysosomal storage disorder. Our Genetic Counseling team will communicate abnormal screening results to the pediatrician or health care professional, provide clinical information about the disorder, and provide recommendations for confirmatory testing through a metabolic treatment center. Assistance is also provided to connect the pediatrician or health care professional with metabolic or LSD specialists for follow-up care.

Overview of Lysosomal Storage Disorders

Lysosomal storage disorders develop as a result of an enzyme deficiency or malfunction that causes cell waste to build up within the cell instead of being excreted. There are approximately 50 known LSDs, each caused by a unique gene mutation which leads to a disease-specific enzyme that is deficient or malfunctioning. The stored cellular waste is also disease-specific, which helps in diagnosing the particular LSD. Almost all LSDs are inherited in an autosomal recessive fashion; one exception is Fabry Disease, which follows X-linked recessive inheritance. Each LSD has its own incidence rate, but as a group, LSDs occur in 1:5,000 to 1:10,000 births in the United States. Early detection and diagnosis are paramount to ensure timely intervention before irreversible symptoms occur.

Lysosomal Storage Disorders screened by PerkinElmer Genetics:

Fabry Disease
(α-galactosidase deficiency)

Gaucher Disease
(glucocerebrosidase deficiency)

Pompe Disease
(glycogen storage disease type II)

Krabbe Disease
(galactocerebrosidase deficiency)

Hurler Syndrome
(mucopolysaccharidosis I, MPS-I)

Niemann-Pick A/B Disease
(acid sphingomyelinase deficiency)

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Common Clinical Symptoms:

- Coarse Facial Features
- Bone Abnormalities
- Joint Stiffness
- Burning Sensations
- Corneal Clouding
- Enlarged Liver or Spleen
- Developmental Delay
- Ataxia
- Seizures
- Loss of Learned Skills

Clinical Features

There are a wide range of neurologic and non-neurologic clinical symptoms. The onset of symptoms can occur from infancy to adulthood. Some disorders are not known to have neurologic features, while others include a wide range of neurologic symptoms, which may or may not include other visible physical characteristics. Pre-symptomatic treatment of LSDs known to have neurological symptoms is very important, as damage from these symptoms is irreversible.

Treatment Options

Treatment for LSDs mainly involves treating the symptoms, not the disorder. There is no known cure for LSDs at this time, but emerging therapies are constantly being developed. Enzyme replacement therapy (ERT) is most effective in treating non-neurologic symptoms in diseases such as Gaucher, Fabry, MPS-I, and Pompe Disease. Hematopoietic stem cell transplantation (HSCT), using either bone marrow or cord blood to provide healthy stem cells that can produce the needed enzyme, is useful in treating diseases with neurologic symptoms, such as Krabbe, Gaucher Type II and MPS-I. Gene therapy has limited availability for some disorders, though mostly available as a part of clinical trials.

Specimen Requirements

Dried blood spots with a minimum of 25 µl of blood on 903,226 or other FDA cleared filter paper.

Turn-Around Time

72 hours from receipt of specimen.

For More Information

Please contact PerkinElmer Genetics at 866-463-6436 or visit **www.perkinelmergenetics.com**

1 - Li, Y., Scott, C.R., Chamoles, N.A., Ghavami, A., Pinto, B.M., Turecek, F., Gelb, M.H. (2004) "Direct Multiplex Assay of Lysosomal Enzymes in Dried Blood Spots for Newborn Screening," Clin. Chem., 50:1785-96.

PerkinElmer, Inc.
940 Winter Street
Waltham, MA 02451 USA
P: (800) 762-4000 or
(+1) 203-925-4602
www.perkinelmer.com

PerkinElmer®

For a complete listing of our global offices, visit www.perkinelmer.com/ContactUs

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