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Sanofi Genzyme, PerkinElmer offer free Dx testing for LSD

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Sanofi's Genzyme subsidiary and PerkinElmer Genomics, the molecular genetics laboratory of PerkinElmer, have launched a program called the Lantern Project to provide no-cost diagnostic testing to patients in the US who suffer from certain lysosomal storage disorders (LSDs).

Through the project physicians will be able to arrange for screening of certain suspected LSDs as well as confirmatory DNA testing and phlebotomy services for their patients. Physicians will be able to refer patients who may be suffering from Gaucher disease, Fabry disease, Pompe disease, mucopolysaccharidosis type I (MPS I), or acid sphingomyelinase deficiency (ASMD), also known as Niemann-Pick disease types A and B. Additionally, patients have the option of undergoing testing with an enzyme panel for seven mucopolysaccharidoses and a 105-gene panel for limb-girdle muscular dystrophies (LGMD) and other myopathies.

LSDs comprise a group of more than 40 genetic diseases that are frequently characterized by a range of symptoms. Disease progression and symptom severity can vary widely. One of the benefits of the Lantern Project is the LGMD panel, which is able to sequence 105 genes known to be associated with LGMDs and other myopathies, a heterogeneous group of muscular weakness disorders that vary in severity and age of onset. The panel uses NGS technology to sequence multiple genes simultaneously, testing for multiple LGMD disorders at once in addition to other diseases that may cause similar symptoms, including Pompe disease and spinal muscular atrophy.