عنوان مقاله:

Diagnostic methods for Lysosomal Storage Disease

محل انتشار:

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خلاصه مقاله:

Lysosomal storage disorders (LSD) are a class of metabolic disturbance in which manifested by the accumulation of large molecules (complex lipids, glycoproteins, glycosaminoglycans, etc.) in lysosomes. LSDs have a wide range of clinical symptoms that may contain organ dysfunction, neurological and skeletal disorders. The first stage of diagnosis is clinically suspected by a physician. Next stage is enzyme activity assays including Fluorometry and MS/MS methods. These methods usually placed in newborn program screening. The second laboratory diagnostic stage is molecular examination (RFLP-PCR and ARMS-PCR, Mutations Scanning Methods, DNA sequencing, MLPA and NGS methods) that is confirmation of the enzyme assays. In this article, routine diagnostic methods for LSDs were discussed. The gold standard for enzyme activity assay and molecular diagnosis is TMS and NGS, respectively.

کلمات کلیدی:

Diagnostic methods, Enzyme activity, Lysosomal storage disease, Molecular assay.