

White Cell Enzymes

Synonyms

Lysosomal storage disease, lysosomal enzymes, hexosaminidase, mannosidase, lysosomal acid lipase

Clinical Indication

Lysosomes are cytoplasmic, single membrane-bound organelles that contain enzymes responsible for a variety of compounds including mucopolysaccharides, sphingolipids and glycoproteins. Deficient activity of specific enzymes leads to progressive accumulation of partially degraded material, leading to distention of the cell, disruption of cellular function and sometimes failure of active transport of small molecules from the lysosomes.

Lysosomal storage diseases can be subdivided according to the compound or pathway involved.

- Mucopolysaccharidoses (e.g. Hurler syndrome)
- Sphingolipidoses (e.g. GM1 gangliosidosis, Tay Sachs, Fabry, Gaucher)
- Glycoproteinoses (e.g. mannosidosis)
- Mucolipidoses
- Lysosomal membrane transport disorders (e.g. sialic acid storage disease)
- Other e.g. lysosomal acid lipase deficiency

The clinical manifestations vary depending upon the location and extent of storage that occurs but may include progressive hepatomegaly, splenomegaly, neurologic regression, short stature, coarsening of facial features, limitation/restriction of small and large joints, peripheral neuropathy, and/or ataxia.

Lysosomal storage diseases screened for: GM1 gangliosidosis, metachromatic leucodystrophy, Hrabble leucodystrophy, fucosidosis, mucopolysaccharidosis VII, beta-mannosidosis, GM2 gangliosidosis, alpha-mannosidosis, Schindlers disease, mucopolidosis II and III, Gaucher disease, Fabrys disease.

Great Ormond Street provide a specific request form to capture as much useful information as possible as an aid to testing:

[Laboratory specimen requirements | Great Ormond Street Hospital](#)

Part of Profile / See Also

Request Form

Combined Pathology manual blood form or ICE request

Full clinical details must be given so that the appropriate testing and interpretation can be provided.

Availability / Frequency of Analysis

Referred test: Analysed by Great Ormond Street Hospital if specific criteria met. (Lab registration 8692)

Turnaround Time

6 weeks

Patient Preparation

Blood transfusion within the last 4 weeks may invalidate the test.

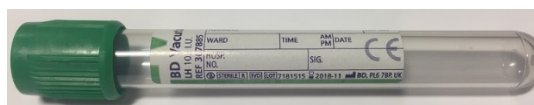
Samples must only be collected Monday to Thursday.

Sample Requirements

Specimen Type Lithium heparin whole blood

Volume At least 5 mL

Container



Green top lithium heparin tube



Paediatric lithium heparin (orange top Sarstedt)



Paediatric lithium heparin (green top BD microtainer)

Samples must be collected Monday to Thursday only, and must be transported to laboratory immediately.

Reference Range & Units

Provided on the report

Interferences

Blood transfusion within the last 4 weeks may invalidate the test.

Interpretation & Clinical

Decision Value (if applicable)

Full interpretation will be provided by the referral laboratory.

The very rare forms of these disorders due to an activator protein deficiency cannot be excluded by these tests. Synthetic substrates are used to assay the enzymes and very rarely, with certain mutations which cause low activity with the natural substrate, normal activity may be seen with the synthetic substrate. This screen does not exclude all lysosomal enzyme disorders.

References

Up to date – Inborn Errors of Metabolism – searched February 2019

[GOSH test database \(individual enzymes listed\) - Laboratory tests database |](#)

[Great Ormond Street Hospital](#)

Test code

WENZ

Lab Handling

DO NOT SEPARATE SAMPLE. Store as whole blood at 4°C in the referrals rack. Sent daily by courier to Great Ormond Street, London. Sample must reach the referral laboratory within 72 hours.



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