# White Cell Enzymes

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| **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.<br><br>Lysosomal storage diseases can be subdivided according to the compound or pathway involved.<br>• Mucopolysaccharidoses (e.g. Hurler syndrome)<br>• Sphingolipidoses (e.g. GM1 gangliosidosis, Tay Sachs, Fabry, Gaucher)<br>• Glycoproteinoses (e.g. mannosidosis)<br>• Mucolipidoses<br>• Lysosomal membrane transport disorders (e.g. sialic acid storage disease)<br>• Other e.g. lysosomal acid lipase deficiency<br><br>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.<br><br>Lysosomal storage diseases screened for: GM1 gangliosidosis, metachromatic leucodystrophy, Hrabble leucodystrophy, fucosidosis, mucopolysaccaridosis VII, beta-mannosidosis, GM2 gangliosidosis, alpha-mannosidosis, Schindlers disease, mucolipidosis II and III, Gaucher disease, Fabrys disease.<br><br>Great Ormond Street provide a specific request form to capture as much useful information as possible as an aid to testing:<br><br>http://www.labs.gosh.nhs.uk/laboratory-services/chemical-pathology/enzyme-laboratory/sending-us-a-sample |
| **Part of Profile / See Also** | |
| **Request Form** | Combined Pathology manual blood form or ICE request<br><br>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.<br><br>Samples must only be collected Monday to Thursday. |
| **Sample Requirements** | |

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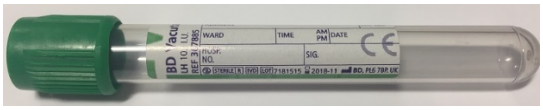
| **Specimen Type** | Lithium heparin whole blood |
| **Volume** | At least 5 mL |
| **Container** |  Green top lithium heparin tube<br>  Paediatric lithium heparin (orange top Sarstedt)<br>  Paediatric lithium heparin (green top BD microtainer)<br>Samples must be collected Monday to Thursday only, and 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.<br>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<br>GOSH enzyme laboratory - http://www.labs.gosh.nhs.uk/laboratory-services/chemical-pathology/enzyme-laboratory |
| **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. |

UKAS MEDICAL
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