Metabolic Biochemistry Network Best Practice Guideline
Reporting results of Lysosomal Enzyme Analysis

Background

This Guideline has been produced to enable consistent reporting of results of lysosomal enzyme analysis between laboratories, to ensure reports are clear, concise, accurate, fully interpretive and that they answer the clinical question. The aim is to ensure best practice and thus provide a high quality service to requesters, which will benefit patients and their families.

The Guideline was written following a meeting of Clinical Scientists and Biomedical Scientists from Metabolic Biochemistry Network (MetBioNet) stakeholder laboratories (see below for contributors) which provide a service for the analysis of lysosomal enzymes on 1st June 2015 in Bristol. (See below for contributors). It has been based on the Genetic Services Quality Committee of the European Society of Human Genetics recommendations for reporting of results of diagnostic genetic testing [1].

The guidelines were reviewed and updated in June 2021 by Katie Harvey (Great Ormond Street Hospital for Children, London)and Victoria Warburton (University Hospitals Bristol NHS Foundation Trust).

Best practice guidelines for a report of lysosomal enzymes analysis

Administrative

The report should include the full name, date of birth and gender of the patient and any additional patient identification provided (e.g. NHS number, referral laboratory number).

The sample type, the date and time it was taken and the date and time it arrived in the enzyme laboratory, if known should be stated on the report.

If there are any problems with the sample e.g. haemolysis, small sample volume, this should also be stated.

The address and telephone number of the laboratory performing the analysis and the name of scientist(s) who validated the report and the date the report is issued should be included.
The report should fulfil the requirements of ISO 15189 [2].

Clinical information

The clinical information supplied with the request should be included on the report.

For analysis of complex metabolic investigations, provision of clinical information is important. If clinical details are not provided, the laboratory should attempt to obtain information regarding the reason for the request. This is less important for single enzyme requests where the requesting clinician is asking for a specific disorder to be tested. If no information is available, the report should state that no clinical information was provided. Clinical information enables the reporting scientist to ensure the appropriate tests are done on the sample, to interpret the

results in the context of the clinical picture and offer advice on additional testing which may be appropriate.
A request form designed for use for requesting analysis of lysosomal enzymes and other metabolic investigations may be helpful in ensuring appropriate information required for interpretation of results is provided.

Results

Results should be brief and unambiguous. The terms 'positive' and 'negative' should not be used.

For quantitative results, include a reference range where appropriate. For less frequently monitored assays, where reference ranges are less robust or not established, it may be more appropriate to report test patient results alongside within-assay controls. Where reference ranges are used these should be regularly monitored to take in to account any method changes.

If samples from controls (affected or unaffected) have been analysed at the same time, results of these samples should be included on the report.

Interpretation of the results

As far as possible, the clinical question should be answered concisely and clearly.

The conclusion should be highlighted e.g. by using bold text or putting the text in a box.

Normal findings

The name of each enzyme tested and the name of the disorder caused by the deficiency of the enzyme should be provided on the report. When a lysosomal enzyme panel is performed an appropriate caveat stating that not all lysosomal storage disorders have been excluded by the analysis should be included on the report.

Non-specific findings

If there is evidence of sample degradation or other artefacts interfering with the result, this should be stated on the report and advice given on suitable repeat sample requirements.

Results of uncertain significance

Intermediate results should be reported. The significance of such results may become clearer in the future with additional information about the patient or disease. Discuss the results with the requesting clinician to determine the significance of the findings in the context of the clinical history. Published scientific reports may describe similar results and be useful in assisting with interpretation.
Repeat analysis on another sample should be recommended.

Pathogonomic (disease-specific) findings

For results which are associated with disease, the analysis should be repeated on that sample, provided there is sufficient viable sample remaining. If repeat analysis is in agreement the results should be communicated quickly and directly to the requestor.

Disease specific ranges should be supplied if available.

The report should state if further tests are required to confirm the diagnosis in question e.g. repeat enzyme analysis, genetic testing. Advice may be given about alternative specialist laboratories where samples can be sent for these investigations.
Advise referral of the patient to a Metabolic Consultant.

Test Limitations

The report should state any known limitations which may affect test performance particularly in relation to disease detection. This may include known assay limitations, for example, enzyme assays that utilise synthetic substrates may not detect disorders caused by some mutations which would be detected by assays utilising the natural substrate. It may also include other test limitations such as the detection of activator protein deficiencies.

Disclaimer

These guidelines represent best practice in the opinion of the contributors listed below and have been peer reviewed. They are not evidence based but reflect expert opinion. MetBioNet cannot accept any responsibility for any errors or omissions.

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References

[1] Clausters M, Kožich V, Dequeker E *et al*. Recommendations for reporting results of diagnostic genetic testing (biochemical, cytogenetic and molecular genetic). *European Journal of Human Genetics* (2014) 22, 160 – 170.
[2] Medical Laboratories – Requirements for quality and competence (ISO 15189:2012). The British Standards Institution (2012)