

External Quality Assurance Scheme for Lysosomal Enzymes

ERNDIM have run a pilot Lysosomal Enzyme scheme using lyophilized leucocytes or transformed lymphocytes from 2006. In 2010 this became a full ERNDIM scheme.

Currently it covers 10 enzymes with 8 samples distributed per year and there are 57 registered participants, although not all have submitted results.

Transformed lymphocytes are circulated from subjects with and without proven lysosomal storage disorders. Reports are then submitted in pre-defined units, and also as a percentage of the mean of normal samples run in parallel. This goes some way towards addressing the very different levels of activity between transformed lymphocytes and leucocytes for some enzymes and also variations in methodology.

One challenge is analysing the small amount of material provided for many enzymes within a short time frame. However the scheme is becoming more useful as it matures.

The report is in a format that will be familiar to users of the ERNDIM amino acids scheme.

See <http://cms.erndimga.nl/> for full details.

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