

Annual Report 2025

Version Number¹: 1

Date of issue: 27th March 2026

Note: This annual report is intended for participants of the ERNDIM Lysosomal Enzymes in fibroblasts scheme. The contents should not be used for any publication without permission of the Scientific Advisor.

The fact that your laboratory participates in ERNDIM schemes is not confidential. However, the raw data and performance scores are confidential and will only be shared within ERNDIM for the purpose of evaluating performance of your laboratory, unless ERNDIM is required to disclose performance data by a relevant government agency. For details, please see the terms and conditions in the EQA Schemes Catalogue and Participant Guide and the ERNDIM Privacy Policy on www.erndim.org.

1. Scheme Design

The scheme has been designed, planned and coordinated by Dr Ed Jacobs (as Scientific Advisor) and Dr. R.M. Schoeman (as Scheme Organiser; sub-contractor on behalf of MCA Laboratory); both appointed by and according to procedures laid down by the ERNDIM Board.

1.1. Sub-contracted activities:

The fibroblasts used as the EQA materials were cultured by Centre de Biotechnologie Cellulaire, CHU de Lyon. The fibroblasts were prepared and aliquoted by MCA Laboratory, Netherlands, which also hosts and manages the results submission website (www.erndimqa.nl) on behalf of ERNDIM; however, ERNDIM remains responsible for this work.

2. Samples

All EQA materials are lyophilised samples of human fibroblasts. All samples were obtained following local ethical and consent guidelines.

Details regarding stability of samples were provided in the scheme instructions, which are available to download from the Participant Information tab of the ERNDIM Registration Website (www.eqa.erndim.org). Samples are stable for the duration of the scheme's submission calendar when stored under defined conditions.

Table 1: Samples included in the EQA scheme

Sample	Disorder	Enzyme Defect	Reporting deadline
LEFB2025.01	Control	All enzymes normal	30 May 2025
LEFB2025.02	MPS VI	Arylsulphatase B	
LEFB2025.03	Pompe disease	α -Glucosidase	
LEFB2025.04	MPS II	Iduronate sulphatase	29 August 2025
LEFB2025.05	CLN2	Tripeptidyl peptidase 1	
LEFB2025.06	MPS 3C	Acetyl-CoA-glucosamine acetyltransferase	

¹ If this Annual Report is not Version 1 for this scheme year, go to APPENDIX 3 (page 12) for details of the changes made since the last version of this document.

3. Shipment

One shipment of six samples was dispatched from MCA Laboratory in The Netherlands on 4th February 2025, to the 69 laboratories, from 28 countries, which registered for the scheme.

4. Receipt of results

There were two submission deadlines for the 2025 scheme: (LEFB2025.01, 02 & 03 on 30th May) and (LEFB2025.04, 05 & 06 on 29th August).

Laboratories were asked to submit results for each EQA sample by the relevant submission deadline using the results website www.erndimqa.nl. All submitted results are treated as confidential information and are only shared with ERNDiM approved persons for the purposes of evaluation and reporting.

Laboratories were asked to report the total protein in mg/vial, and the activities for 10 enzymes in absolute units (See Table 2 for details). In 2025, the activity as a percentage of sample LEFB 01 was automatically calculated by the results website.

Laboratories could submit results for as many, or as few, of these 10 enzymes as they wished and were asked to select an 'interpretation' of the results from a dropdown list on the results website.

Table 2: Parameters to be measured

Analyte	Parameter 1	Parameter 2 (calculated automatically)
Protein	mg/vial	-
Acetyl-CoA-glucosamine acetyltransferase	nmol/17h/mg protein	% of sample LEFB 01
Alpha-galactosidase	nmol/h/mg protein	% of sample LEFB 01
Alpha-glucosidase	nmol/h/mg protein	% of sample LEFB 01
Arylsulphatase A	nmol/h/mg protein	% of sample LEFB 01
Arylsulphatase B	nmol/h/mg protein	% of sample LEFB 01
Beta-galactosidase	nmol/h/mg protein	% of sample LEFB 01
Beta-glucosidase	nmol/h/mg protein	% of sample LEFB 01
Beta-mannosidase	nmol/h/mg protein	% of sample LEFB 01
Iduronate sulphatase	nmol/4h/mg protein	% of sample LEFB 01
Tripeptidyl peptidase 1	nmol/h/mg protein	% of sample LEFB 01

5. Reports

All data-transfer, the submission of data as well as request and viewing of reports is via the interactive website www.erndimqa.nl which can also be reached through the ERNDiM website (www.erndim.org). The results of each laboratory are confidential and only accessible by password protected laboratory accounts. The anonymised mean results of all labs are accessible to all participants. Statistics of the respective reports are explained in the general information section of the website.

Short-term reports on the six individual specimens are available three weeks after the submission deadline and provide up-to-date information on analytical performance. Although it is technically possible to produce reports immediately there is a delay of 21 days to enable the Scientific Advisor to inspect the results and add comments to the report when appropriate.

A second important characteristic of the website is the different levels of detail of results which allows individual laboratories the choice of fully detailed and/or summarised reports.

The "Analyte in Detail" is the most detailed report and shows the results of a specific analyte in a specific sample. Thus, for the 10 enzymes in the year 2025 cycle, 6 x 10 (60) such Analyte-in-Detail-reports can be requested.

The "Cycle Review" summarises the performance for all enzymes in a specific sample (6 such Cycle Reviews can be requested in 2025).

6. Scoring scheme and Poor performance policy

If the interpretation of a result is incorrect (such that a deficiency is missed) for a specific enzyme and this is designated as a critical error, a performance support letter will be issued. This is to initiate a dialogue between us, the EQA scheme advisor/organiser and you, the participating laboratory, to solve any particular analytical problems and to help you improve performance. If a participant scores less than 70% of the maximum number of points that can be obtained, they will be classed as a poor performer, and a letter will be sent to that participant.

Comments box: Participant comments may be taken into account by the Scientific Advisor. Please use this box to note any issues regarding the sample or assay, or to note further relevant information.

The **diagnostic proficiency** was scored for each enzyme: i.e., if the interpretation *and* measurement was correct it was awarded two points

For the protein value a maximum of 2 points could be scored depending on the % recovery. Note that the term recovery is now used instead of CV, as this is the appropriate terminology.

Table 3: Scoring criteria

	Criteria	Score	
Protein	Recovery	Recovery < 35%	2
		35% ≤ Recovery ≤ 60%	1
		Recovery > 60%	0
Enzymes	Diagnosis	Diagnosis correct	2
		Diagnosis incorrect	0

Laboratories could participate in as many of the ten enzymes offered in the scheme, plus the protein assay as required. Each enzyme is assessed individually, the emphasis being on the correct interpretation of the result. Making the correct interpretation / diagnosis for each enzyme/ sample is the priority: i.e., identifying a deficiency in an affected patient and reporting normal activity in unaffected samples.

Further information regarding the Framework for Assessment and Education in the LEFB scheme can be found under the Participant Information tab on the ERNDiM registration website (www.eqa.emdim.org).

6.1. Diagnosis

The participants must select an interpretation from the dropdown list on the results website.

Diagnosis correct: correct interpretation *and* correct measurement of enzyme activity level.

Diagnosis incorrect: incorrect interpretation and/or incorrect measurement of enzyme activity level.

6.2. Recovery

Only the recovery for protein contributes to scoring: this is calculated from the median results for all labs.

6.3. Appeals

If your laboratory has been sent a performance support letter for the 2025 scheme and you wish to appeal against this classification, please complete the online appeal form (see below) within one month of the date of the relevant Performance Support Letter. Full details of the reason for the appeal should be included. Initial appeals will be considered by the relevant Scientific Advisor and a decision sent within 21 days of receipt of the appeal.

Appeal form: https://www.formdesk.com/erndim/Poor_Performance_Appeals_Form [please note this form will only be accessible for one month after the performance support letters have been sent].

7. Results

Sixty-nine laboratories were registered in the 2025 scheme. Sixty-nine laboratories (100% of registered laboratories) submitted sufficient results for their performance to be assessed.

Table 4: Results returns for the 2025 scheme

	Submission Deadline					
	30 th May 2025			29 th August 2025		
Sample Numbers:	2025.01	2025.02	2025.03	2025.04	2025.05	2025.06
No. of labs that submitted results:						
By the submission deadline	69	69	69	69	69	69
Within 7 days of the submission deadline	0	0	0	0	0	0
Within 2 weeks of the submission deadline	0	0	0	0	0	0
Did not submit	0	0	0	0	0	0

The results for each sample were published on the results website 21 days after the relevant submission deadline.

Full details of the results for each participant's results (for labs that submitted results) are given in Appendix 1 but summaries are presented here:

- 87% of participating laboratories submitted results for 5 or more enzymes, see Table 5.
- The proficiency per analyte is given in
- Table 6.
- The majority of participants made the correct interpretation.

- 69.5% of participating laboratories achieved >90% of their maximum possible score (i.e., of enzymes plus proteins). See Table 7 which shows the percentage of the maximum possible score for the laboratories that submitted results.

Table 5: Number of enzymes for which laboratories submitted results (excluding non/partial submitters)

Number of Enzymes for which results were submitted	Number of laboratories
0	0
1	0
2	2
3	3
4	4
5	2
6	11
7	6
8	9
9	6
10	26
Total number of labs	69

Table 6: Proficiency per analyte

Analyte	No of returns	Correct interpretation* (diagnostic proficiency)
Protein	69	86.2%
Acetyl-CoA-glucosamine acetyltransferase	35	91.4%
Alpha-galactosidase	66	93.9%
Alpha-glucosidase	58	96.6%
Arylsulphatase A	60	94.2%
Arylsulphatase B	52	88.5%
Beta-galactosidase	65	96.9%
Beta-glucosidase	68	95.6%
Beta-mannosidase	46	89.1%
Iduronate sulphatase	47	91.5%
Tripeptidyl peptidase 1	36	95.8%

* = percentage of maximum possible score (for laboratories that submitted results)

Table 7: Percentage of maximum possible scores for laboratories that submitted results (excluding partial submitters)

%age of maximum possible score	No of submitting labs	%age of submitting labs
0% – 9%	0	0.0%
10% – 19%	1	1.4%
20% – 29%	0	0.0%
30% – 39%	0	0.0%
40% – 49%	0	0.0%
50% – 59%	1	1.4%
60% – 69%	3	4.3%
70% – 79%	2	2.9%
80% – 89%	14	20.3%
90% – 99%	11	15.9%
100%	37	53.6%
Total	69	100%

Table 8: Number of enzymes for which laboratories had satisfactory performance

Anon Lab No.	No of enzymes for which:	
	results were submitted by lab	lab had satisfactory performance
1	6	6
2	6	6
3	10	10
4	10	10
5	8	8
6	6	5
7	10	10
8	8	7
9	4	4
10	4	4
11	10	10
12	10	10
13	10	10
14	7	7
15	10	9
16	10	8
17	9	9
18	4	4
19	3	3
20	8	8
21	10	10
22	9	9
23	10	8
24	10	10
25	10	8
26	7	5
27	8	7
28	8	8
29	10	10
30	7	6
31	4	4
32	6	3
33	6	6
34	8	8
35	2	2
36	9	9

Anon Lab No.	No of enzymes for which:	
	results were submitted by lab	lab had satisfactory performance
37	6	6
38	2	2
39	8	5
40	10	8
41	10	10
42	7	7
43	10	10
44	6	6
45	10	10
46	6	5
47	9	9
48	6	0
49	5	3
50	3	3
51	6	5
52	3	3
53	10	9
54	10	10
55	10	10
56	7	6
57	10	10
58	9	9
59	10	10
60	10	10
61	8	8
62	10	10
63	5	5
64	9	9
65	10	10
66	6	5
67	7	7
68	8	7
69	10	10

8. Certificates of Participation

As for other ERNDiM schemes, the performance for this scheme is summarised in the annual Certificate of Participation. The certificate lists the total number of enzymes in the scheme, the number for which results have been submitted and the number for which satisfactory performance has been achieved. It is important to bear in mind that the certificate must be backed up by the laboratory's individual on-line reports in the case of internal or external auditing.

9. Comments on Overall Scheme Performance.

All ten enzymes included in the 2025 scheme were assayed in all six samples prior to distribution for validation.

- One cell line (LEFB 01) had no enzyme deficiency confirming no disorder in respect to the enzymes to be tested and was classified as Control.

- Five affected cell lines (LEFB 02 to LEFB 06) had clear enzyme deficiencies confirming the specific disorder in each case.
- The remaining enzymes in all six samples included in the scheme had confirmed normal or near-normal levels of enzyme activity.

The majority of participants made the correct interpretation: that is, the correct enzyme deficiency was observed in the samples from affected patients and normal activity was observed in the unaffected samples.

LEFB 01 was included as a control to enable an improved comparison of overall results from all participants, and to provide a control to participants that do not use fibroblasts.

LEFB 02 was a patient with MPS VI. The correct interpretation for this sample was arylsulphatase B deficiency. The activity measured by the reference lab, in two vials, was 39 and 37 nmol/h/mg (normal 320-1080 nmol/h/mg; patients 19-105 nmol/h/mg). Fifty-two participants submitted data for this enzyme. Proficiency for this enzyme was 88.5% and three CE were given. The historical proficiency since 2015 is 90.7% (2021; deficient).

LEFB 03 was a patient with Pompe disease. The correct interpretation for this sample was α -glucosidase deficiency. The activity measured by the reference lab, in two vials, was 6.3 and 5.5 nmol/h/mg (normal 45-180 nmol/h/mg; patients 0-20 nmol/h/mg). Fifty-eight participants submitted data for this enzyme. Proficiency for this enzyme was 96.6% and one CE was given. The historical proficiency since 2015 is 69% (2015; normal); 76% (2016; normal); 85% (2017; deficient); 90% (2018; normal); 88% (2019; deficient); 100% (2020; normal); 91.1% (2021; normal); 94% (2022; deficient); 88.9% (2023; deficient) and 100% (2024; normal).

LEFB 04 was a patient affected with MPS 2. The correct interpretation for this sample was iduronate sulphatase deficiency. The activity measured by the reference lab, in two vials, was 0.2 and 0.1 nmol/4h/mg (normal 26-109 nmol/4h/mg; patients 0-2.5 nmol/4h/mg). Forty-seven participants submitted data for this enzyme. Proficiency for this enzyme was 91.5% and two CE were given. The historical proficiency since 2015 is 90% (2019; normal) and 93.3% (2023; normal).

LEFB 05 was a patient affected with CLN2. The correct interpretation for this sample was tripeptidyl peptidase 1 deficiency. The activity measured by the reference lab, in two vials, was 0 and 0 nmol/h/mg (normal 136-454 nmol/h/mg; patients 0-7.8 nmol/h/mg). Thirty-six participants submitted data for this enzyme. Proficiency for this enzyme was 95.8% and no CE were given. The historical proficiency since 2015 is 92% (2019; deficient).

LEFB 06 was a patient with MPS 3C. The correct interpretation for this sample was acetyl-CoA-glucosamine acetyltransferase deficiency. The activity measured by the reference lab, in two vials, was 1.2 and 2.4 nmol/17h/mg (normal 50-225 nmol/17h/mg; patients 0.7-4 nmol/17h/mg). Thirty-five participants submitted data for this enzyme. Proficiency for this enzyme was 91.4% and two CE were given. This was the first time this enzyme was included in the LEFB scheme.

Additional information regarding sample LEFB 06 of the LEFB 2024 scheme.

In the LEFB 2024 annual report a paragraph was dedicated to the relatively high residual activity of β -mannosidase, which was far below the normal range, 16% of the activity in sample 01 but higher than the classical range of patients.

This cell line was chosen because enzymatically the diagnosis β -mannosidase deficiency was made in 1997 in leukocytes and fibroblasts. Because of the relatively high residual activity, CHU in Lyon have sequenced the *MANBA* gene and found a single heterozygous pathogenic variant. A second variant was not found. We therefore have to conclude that the high residual activity rather reflects heterozygosity than a true deficiency.

10. Preview of the scheme in 2026.

- a) There will be two submission deadlines for the 2026 scheme:
 - Samples 01, 02 & 03 to be submitted by 29th May 2026
 - Samples 04, 05 & 06 to be submitted by 28th August 2026
- b) Introduced in the 2024 LEFB Scheme, the following will be continued:
 - Age, gender and clinical symptoms will be provided to facilitate the interpretation of the enzyme testing results, especially in case of late onset disorders.
 - The CV has been renamed recovery, as this is the appropriate term.
 - All values as % activity in sample 2026.01 will be automatically calculated by the results website.
 - **An interpretation/diagnosis must be selected for each sample from the multiple-choice list.** If at least one of the offered interpretations/diagnoses is not selected for each sample, it is not possible to evaluate the results of any of the enzymes for which values are submitted. Therefore, if at least one interpretation/diagnosis is not selected for **every** sample, zero points will be given for **each** of the submitted enzymes. In case no deficiency is found in one or more of the measured enzymes, whether or not all 10 enzymes have been measured, "No obvious deficiency (according to the enzymes tested)" has to be selected. It has been made clearer in the 2026 scheme instructions that interpretations/diagnoses made in the comment box, do not replace the interpretation/diagnosis selected in the multiple choice list or the obligation to select at least one of the offered interpretations or diagnoses for each sample.
- c) Some changes have been made to the enzymes included in the 2026 LEFB scheme. See Table 9 for the list of enzymes in the 2026 scheme.
- d) For purposes of laboratory accreditation there is an increasing demand for the inclusion of further and different enzymes in the scheme. In order to address this requirement, it is intended that ERNDiM continue to provide regular rotation of the enzymes included each year. In addition, ERNDiM has investigated if the scheme can be extended to different enzymes as well as to a higher frequency of rotation. To this end, two surveys have been sent out to learn which enzymes should be in the LEFB scheme. ERNDiM is now working on the realisation of an extension of the scheme.
- e) Furthermore, ERNDiM is still working on investigating whether it is possible to initiate a Lysosomal Enzymes in Dried Blot Spot (LEDB) pilot scheme.

Table 9: Analytes to be measured in 2026

Analyte	2018	2019	2020	2021	2022	2023	2024	2025	2026
Protein	✓	✓	✓	✓	✓	✓	✓	✓	✓
Acetyl-CoA-glucosamine acetyltransferase	x	x	x	x	x	x	x	✓	x
Arylsulphatase A	x	✓	✓	x	✓	✓	x	✓	x
Arylsulphatase B	x	x	x	✓	x	x	x	✓	x
Aspartylglucosaminidase	x	x	x	x	✓	x	x	x	✓
Galactose-6-sulphate sulphatase	✓	x	x	x	x	✓	x	x	x
Galactosylceramidase	✓	✓	✓	x	✓	x	x	x	x
Heparan-N-sulphatase	x	x	x	x	x	✓	x	x	✓
Iduronate-sulphatase	x	✓	x	x	x	✓	x	✓	x
Lysosomal acid lipase (LAL/acid/esterase)	x	✓	✓	x	x	x	✓	x	x
N-acetylglucosamine-6-sulphatase	x	x	x	x	x	x	x	x	✓
Palmitoyl protein thioesterase	x	✓	✓	x	x	x	✓	x	x
Sphingomyelinase	✓	x	x	✓	✓	x	x	x	x
Tripeptidyl peptidase	x	✓	x	x	x	x	x	✓	x
α-Fucosidase	x	x	x	✓	x	x	x	x	✓
α-Galactosidase	✓	✓	✓	✓	✓	✓	✓	✓	✓
α-Glucosidase	✓	✓	✓	✓	✓	✓	✓	✓	✓
α-Iduronidase	✓	x	x	x	x	✓	x	x	x
α-Mannosidase	x	x	x	✓	x	x	✓	x	✓
α-N-Acetyl-glucosaminidase	x	x	✓	x	x	x	✓	x	x
β-Galactosidase	✓	✓	✓	✓	✓	✓	✓	✓	✓
β-Glucosidase	✓	✓	✓	✓	✓	✓	✓	✓	✓
β-Glucuronidase	x	x	✓	x	x	x	✓	x	✓
β-Hexosaminidase A	✓	x	x	✓	✓	x	x	x	x
β-Hexosaminidase A+B	✓	x	x	✓	✓	✓	x	x	x
β-Mannosidase	x	x	x	x	x	x	✓	✓	x

11. Questions, Comments and Suggestions

If you have any questions, comments or suggestions in addition to specific user comments please address these to the either the ERNDIM Administration Office (admin@erndim.org), the scientific advisor of the scheme, Dr Ed Jacobs, (admin@erndim.org) or the scheme organiser Dr. R.M. Schoeman (mca.office@skbwinterswijk.nl).

Most complaints received by ERNDIM consist of minor misunderstandings or problems with samples, which can usually be resolved via direct contact with the ERNDIM administrative staff. If you wish to file a formal complaint, please email your complaint with details of your issue to admin@erndim.org or contact us through our website at <https://www.erndim.org/contact-us/>

12. Confidentiality Statement

This annual report is intended for participants of the ERNDIM Lysosomal Enzymes in fibroblasts scheme. The contents should not be used for any publication without the permission of the Scientific Advisor and Administration Office.

27th March 2026



Ed Jacobs
Scientific Advisor

APPENDIX 1. Results per laboratory (part 1)

(see page 12 for key)

Anon Lab No.	Protein/vial	Score				
		Acetyl-CoA- glucosamine acetyltransferase	α -Galactosidase	α -Glucosidase	Arylsulphatase A	Arylsulphatase B
1	2		2	2	2	2
2	1		2	2	2	
3	0	2	2	2	2	2
4	2	2	2	2	2	2
5	2		2	2	2	2
6	2		2	2	2	0
7	1	2	2	2	2	2
8	2		2	2	2	2
9	2		2		2	2
10	2		2	2		
11	2	2	2	2	2	2
12	2	2	2	2	2	2
13	1	2	2	2	2	2
14	2		2	2		2
15	1	2	2	2	2	2
16	0	0	2	2	2	0
17	2	2	2	2	2	2
18	2		2		2	
19	2		2	2		
20	1	2	2	2		2
21	2	2	2	2	2	2
22	2		2	2	2	2
23	2	2	0	0	2	2
24	2	2	2	2	2	2
25	2	2	2	2	0	0
26	2		2	0	2	0
27	2		2	2	2	2
28	2	2	2	2	2	2
29	1	2	2	2	2	2
30	2		2	2	2	2
31	2		2		2	
32	1		2	2		0
33	2		2	2	2	2
34	2	2	2	2	2	
35	2			2		
36	2	2	2	2	2	2
37	2		2	2	2	2
38	0		2			
39	2		2	2	2	0
40	2	0	2	2	1	2
41	1	2	2	2	2	2

Anon Lab No.	Score					
	Protein/vial	Acetyl-CoA-glucosamine acetyltransferase	α -Galactosidase	α -Glucosidase	Arylsulphatase A	Arylsulphatase B
42	2		2		2	2
43	2	2	2	2	2	2
44	1		2	2	2	
45	2	2	2	2	2	2
46	2		2	2	2	
47	2	2	2	2	2	2
48	2		0		0	
49	2		0		2	
50	2		2			
51	2		2		0	2
52	1				2	2
53	2	0	2	2	2	2
54	2	2	2	2	2	2
55	1	2	2	2	2	2
56	2		2	2	2	
57	2	2	2	2	2	2
58	1	2	2	2	2	2
59	2	2	2	2	2	2
60	2	2	2	2	2	2
61	2	2			2	2
62	2	2	2	2	2	2
63	2		2	2		
64	2		2	2	2	2
65	2	2	2	2	2	2
66	2		0	2	2	
67	2	2	2	2	2	
68	1		2	2	2	2
69	2	2	2	2	2	2

APPENDIX 1. Results per laboratory (part 2)

(see page 12 for key)

Anon Lab No.	Score				
	β -Galactosidase	β -Glucosidase	β -Mannosidase	Iduronate sulphatas	Tripeptidyl peptidase 1
1	2	2			
2	2	2		2	
3	2	2	2	2	2
4	2	2	2	2	2
5	2	2	2	2	
6	2	2			
7	2	2	2	2	2
8	2	2		0	2
9	2				
10	2	2			
11	2	2	2	2	2
12	2	2	2	2	2
13	2	2	2	2	2
14	2	2	2	2	
15	2	2	2	0	2
16	2	2	2	2	2
17	2	2	2	2	
18	2	2			
19		2			
20	2	2	2	2	
21	2	2	2	2	2
22	2	2	2	2	2
23	2	2	2	2	2
24	2	2	2	2	2
25	2	2	2	2	2
26	2	2		2	
27	0	2		2	2
28	2	2	2		
29	2	2	2	2	2
30	2	0	2		
31	2	2			
32	2	0		0	
33	2	2			
34	2	2	2	2	
35		2			
36	2	2	2	2	
37	2	2			
38		2			
39	2	2	0	0	
40	2	2	2	2	2
41	2	2	2	2	2
42	2	2	2		2

Anon Lab No.	Score				
	β -Galactosidase	β -Glucosidase	β -Mannosidase	Iduronate sulphatas	Tripeptidyl peptidase 1
43	2	2	2	2	2
44	2	2		2	
45	2	2	2	2	2
46	2	2	0		
47	2	2		2	2
48	0	0	0		0
49	2	2	0		
50	2	2			
51	2	2	2		
52		2			
53	2	2	2	2	2
54	2	2	2	2	2
55	2	2	2	2	2
56	2	2	0		2
57	2	2	2	2	2
58	2	2	2	2	
59	2	2	2	2	2
60	2	2	2	2	2
61	2	2	2	2	2
62	2	2	2	2	2
63	2	2		2	
64	2	2	2	2	2
65	2	2	2	2	2
66	2	2	2		
67	2	2		2	
68	2	2		2	1
69	2	2	2	2	2

Key

	no data submitted for this enzyme
	correct interpretation and correct measurement
	incorrect interpretation and/or incorrect measurement: normal enzyme assigned as deficient (0 pts)
	incorrect interpretation and/or incorrect measurement: deficient enzyme assigned as normal (0 pts and CE)
	no diagnoses submitted
	not enough data submitted for this enzyme
	very deviating activities, either vs mean and/or between the 6 samples (0 pts)

APPENDIX 2. Overall scores per laboratory**Key**

N = Poor performer for score only

CE = Poor performer for critical error only

P = Partial submitter

D = Did not submit any results

Anon lab no.	Total points scored	Maximum points	% score	Performance code
1	14	14	100.0%	
2	13	14	92.9%	
3	20	22	90.9%	
4	22	22	100.0%	
5	18	18	100.0%	
6	12	14	85.7%	CE
7	21	22	95.5%	
8	16	18	88.9%	CE
9	10	10	100.0%	
10	10	10	100.0%	
11	22	22	100.0%	
12	22	22	100.0%	
13	21	22	95.5%	
14	16	16	100.0%	
15	19	22	86.4%	
16	16	22	72.7%	CE
17	20	20	100.0%	
18	10	10	100.0%	
19	8	8	100.0%	
20	17	18	94.4%	
21	22	22	100.0%	
22	20	20	100.0%	
23	18	22	81.8%	
24	22	22	100.0%	
25	18	22	81.8%	CE
26	12	16	75.0%	CE
27	16	18	88.9%	
28	18	18	100.0%	
29	21	22	95.5%	
30	14	16	87.5%	
31	10	10	100.0%	
32	7	14	50.0%	N&CE
33	14	14	100.0%	
34	18	18	100.0%	
35	6	6	100.0%	
36	20	20	100.0%	
37	14	14	100.0%	
38	4	6	66.7%	N

Anon lab no.	Total points scored	Maximum points	% score	Performance code
39	12	18	66.7%	N
40	19	22	86.4%	
41	21	22	95.5%	
42	16	16	100.0%	
43	22	22	100.0%	
44	13	14	92.9%	
45	22	22	100.0%	
46	12	14	85.7%	
47	20	20	100.0%	
48	2	14	14.3%	N
49	8	12	66.7%	N
50	8	8	100.0%	
51	12	14	85.7%	
52	7	8	87.5%	
53	20	22	90.9%	CE
54	22	22	100.0%	
55	21	22	95.5%	
56	14	16	87.5%	
57	22	22	100.0%	
58	19	20	95.0%	
59	22	22	100.0%	
60	22	22	100.0%	
61	18	18	100.0%	
62	22	22	100.0%	
63	12	12	100.0%	
64	20	20	100.0%	
65	22	22	100.0%	
66	12	14	85.7%	
67	16	16	100.0%	
68	16	18	88.9%	
69	22	22	100.0%	

APPENDIX 3. Change log (changes since the last version)

Version Number	Published	Amendments
1	27 March 2026	<ul style="list-style-type: none"> 2025 Annual Report published

END OF REPORT