Congenital Disorders of Glycosylation (CDG)

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2026 First Round Interim Report

Version Number[1]: 01
Date of issue: 11 August 2026

Please Note:

- This interim report is intended for participants of the ERNDIM CDG scheme. The contents should not be used for any publication without permission of the Scientific Advisor.
- This is an interim report, and it includes provisional scores only. All scores are subject to change following moderation at the Scientific Advisory Board meeting in autumn of this year. For final scores and performance data the ERNDIM CDG Annual Report should be referred to.
- The fact that your laboratory participates in this scheme is not confidential, however, the raw data and performance scores are confidential and will only be shared within ERNDIM for the purpose of evaluating your laboratories performance, unless ERNDIM is required to disclose performance data by a relevant government agency. For details, please see the ERNDIM Privacy Policy on www.erndim.org.

1. Results Submission

Results were submitted to the online results website (cscq.hcuge.ch/cscq/ERNDIM/) which is hosted and maintained by CSCQ. The submission deadline for the first round (samples CDG-PP-2026-A, -B and -C) was 11th May 2026.

52 laboratories registered for the 2026 CDG scheme; of these, 48 labs (92%) submitted results for the first round.

2. Scoring scheme

In agreement with ERNDIM rules, we applied a scoring system of 2+2:

Technical aspects: 1 point for identification of an abnormal profile and 1 point for correct identification of the profile as type I or II.

Diagnostic suggestions: This section should be filled for scoring. Just referring to a specialised lab is insufficient. If required, advice can be obtained from a reference laboratory or in collaboration with a clinical colleague. For normal profiles 2 points are scored. For abnormal profiles, comments should be made on the possibility of the presence of a secondary cause in light of the clinical indication. In addition, the right suggestions should be made for the next step in the diagnostic process that eventually will lead to the genetic defect. Scoring for this part is not so straightforward, but we tried to keep it as consistent as possible. The maximum score achievable with full submission for all samples is 24, while a maximum of 12 points are available for labs that only submitted results for the first or second round. The level for satisfactory performance is 17 points.

For the 2022 scheme onwards labs that only submit results for 3 or fewer samples in a scheme year will be classed as partial submitters and their performance will not be evaluated. This information is included in the CDG

[1] **If this Annual Report is not Version 1 for this scheme year, go to APPENDIX 2 (page 4) for details of the changes made since the last version of this document.**

scheme instructions for 2022 onwards. Partial submitters receive a formal Non-submitter letter notifying them of this status and their certificate of participation shows them as not submitting results for the relevant scheme. As the number of participants in the CDG scheme are limited due to the nature of the EQA samples, ERNDIM reserves the right to exclude participants that are classed as partial/non-submitters for 2 out of 3 registered years (i.e., persistent partial and non-submitters) from the scheme.

Another criterion for satisfactory performance is the absence of any "critical error", which is defined as an error resulting from seriously misleading analytical findings and/or interpretations with serious clinical consequences for the patient. For the 2026 CDG scheme, any critical errors will be agreed at the meeting of the Scientific Advisory Board on 3rd and 4th December 2026, and details of these will be included in the 2026 CDG Annual Report.

a. Appeals

If your laboratory is classed as having poor performance at the end of the 2026 scheme and you wish to appeal against this classification, please use the link given in the Performance Support letter you will be sent, to submit your appeal request. The online form should be completed with full details of the reason for your appeal and submitted within one month of receiving your Performance Support Letter. Please note that only appeals submitted using the online response form will be considered.

3. Results of samples and evaluation of reporting

The shipped samples were from (CDG) patients and from controls. The final results of the three first round samples with respect to CDG are summarised in Table 1 below.

Table 1: Samples in the first-round of the 2026 scheme

Sample ID	Clinical information (sex, age, phenotype)	Sex	Age at Diagnosis	Current Age	Diagnoses
CDG-PP-2026-**A**	Microcephaly, feeding problems, hypotonia	M	5 months	2 years	Transferrin polymorphic variant
CDG-PP-2026-**B**	Epilepsy, intellectual disability, nonverbal	F	12 years	15 years	Normal
CDG-PP-2026-**C**	*Retinitis pigmentosa*, marked gait imbalance, peripheral neuropathy	F	65 years	68 years	PMM2-CDG

All submitted results are treated as confidential information and are only shared with ERNDIM approved persons for evaluation and reporting purposes.

Among the laboratories that reported their analytical method (49/52), isoelectric focusing remained the most frequently used technique (15/49), closely followed by HPLC (14/49), capillary electrophoresis (10/49), mass spectrometry (8/49), and other methods (2/49).

Table 2: Scoring of the first-round samples in the 2026 scheme

Sample	No of returns	Technical Aspects (%)	Diagnostic Suggestions (%)	Total (%)
CDG-PP-2026-A	48	91.7	91.7	91.7
CDG-PP-2026-B	48	95.8	95.8	95.8
CDG-PP-2026-C	48	97.9	93.8	95.8

The full anonymised results for all labs that submitted results are given in APPENDIX 1 on page 4 of this report.

3.1. CDG-PP-2026-A: Transferrin polymorphic variant

Most laboratories reported an abnormal transferrin glycoform profile and suggested a protein variant. The transferrin variant, which migrated within the pentasialotransferrin fraction, was detectable by IEF, CE and HPLC and was confirmed by neuraminidase digestion by several labs.

Six laboratories using CE, four using IEF, and six using HPLC did not suggest the presence of a transferrin variant, with ten laboratories ultimately reporting the profile as normal. It is noteworthy that, in this sample, as can be inferred from the relative intensities of the two tetrasialotransferrin peaks/bands, the variant protein appears to account for less than 50% of the total tetrasialotransferrin. Although this is not the typical appearance of the more common transferrin polymorphisms, in which the two tetrasialotransferrin peaks or bands are usually present in approximately equal amounts, some transferrin variants may be underrepresented at the protein level because of reduced protein stability or lower expression of the variant allele compared with the wild-type allele. As a result, these variants may be more difficult to recognise. In routine diagnostics, neuraminidase treatment followed by repeat analysis may therefore be necessary, if required for correct interpretation, to confirm the

presence of a transferrin variant in such cases. It should be noted that, given the underrepresentation of the variant protein in this sample and the fact that the variant migrated within the pentasialotransferrin fraction, laboratories reporting the profile as normal were not penalised for analytical performance, as the sample was correctly classified as normal with respect to CDG.

As mentioned in previous reports, although the presence of a transferrin polymorphism is clinically benign, it can complicate the interpretation of the glycoform profile and should always be ruled out before considering a pathological cause.

3.2. CDG-PP-2026-B: Normal sample

A normal transferrin glycoform profile was identified and interpreted as normal by almost all laboratories, resulting in a total proficiency score of 95.8%.

3.3. CDG-PP-2026-C: PMM2-CDG

A type I abnormal transferrin glycoform profile was reported by almost all laboratories, resulting in a total proficiency score of 95.8%. The profile was clearly abnormal, and no relevant differences were observed between the different analytical methods. The high total score reflects not only the correct recognition of the abnormal profile but also the inclusion of appropriate diagnostic recommendations, which were mainly focused on genetic studies. Nevertheless, one participant reported the sample as normal, resulting in a missed diagnosis.

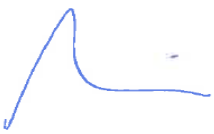
The clinical presentation was highly suggestive of PMM2-CDG, and accordingly around 90% (44/49) of participants suggested this diagnosis. The patient's age, together with the neurological symptoms, led some participants to consider alcohol abuse as a possible secondary cause. Although this is a reasonable differential diagnosis in adults with abnormal transferrin glycoform profiles, the overall clinical presentation was more consistent with PMM2-CDG. Correct identification of the profile as abnormal and the suggestion of PMM2-CDG as a possible diagnosis were required for full scoring.

4. Questions, Comments and Suggestions

If you have any questions, comments or suggestions in addition to specific user comments please contact the ERNDIM Administration Office (admin@erndim.org).

5. Confidentiality Statement

This interim report is intended for participants of the ERNDIM Congenital Disorders of Glycosylation scheme. The contents of this report or data derived from the use or analysis of ERNDIM EQA materials must not be used in written publications or oral presentations unless the explicit prior consent of ERNDIM has been granted.



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APPENDIX 1. **Detailed scores for submitting laboratories**

Sample ID	Technical				Advice				Total score (Max 12)
	A	B	C		A	B	C		
Average score / Anon Lab ID	1.83	1.92	1.96	Total	1.83	1.92	1.88	Total	
1	2	2	2	6	2	2	2	6	**12**
2	2	2	2	6	2	2	2	6	**12**
3	2	2	2	6	2	2	2	6	**12**
4	2	2	2	6	2	2	2	6	**12**
5	2	2	2	6	2	2	2	6	**12**
6	0	2	2	4	0	2	2	4	**8**
7	2	2	2	6	2	2	2	6	**12**
8									**Non-submitter**
9	2	2	2	6	2	2	2	6	**12**
10	0	2	2	4	0	2	2	4	**8**
11	2	2	2	6	2	2	2	6	**12**
12	2	2	2	6	2	2	1	5	**11**
13	0	0	2	2	0	0	2	2	**4**
14									**Non-submitter**
15	2	2	2	6	2	2	2	6	**12**
16	2	2	2	6	2	2	2	6	**12**
17	2	2	2	6	2	2	1	5	**11**
18	2	2	2	6	2	2	2	6	**12**
19	2	2	2	6	2	2	2	6	**12**
20	2	2	2	6	2	2	2	6	**12**
21	2	2	2	6	2	2	2	6	**12**
22	2	2	2	6	2	2	2	6	**12**
23	2	2	2	6	2	2	2	6	**12**
24	2	2	2	6	2	2	2	6	**12**
25	2	2	2	6	2	2	2	6	**12**
26	2	2	2	6	2	2	2	6	**12**
27	2	2	2	6	2	2	2	6	**12**
28	2	2	2	6	2	2	2	6	**12**
29	2	2	2	6	2	2	1	5	**11**
30	2	2	2	6	2	2	2	6	**12**
31	2	2	2	6	2	2	2	6	**12**
32	2	2	2	6	2	2	2	6	**12**
33	2	2	2	6	2	2	2	6	**12**
34	2	0	2	4	2	0	2	4	**8**
35	2	2	2	6	2	2	2	6	**12**
36	2	2	2	6	2	2	2	6	**12**
37	2	2	2	6	2	2	2	6	**12**
38	2	2	2	6	2	2	2	6	**12**
39	2	2	2	6	2	2	2	6	**12**
40	2	2	2	6	2	2	2	6	**12**

Sample ID	Technical				Advice				Total score (Max 12)
	A	B	C		A	B	C		
Average score / Anon Lab ID	1.83	1.92	1.96	Total	1.83	1.92	1.88	Total	
41	2	2	2	6	2	2	2	6	**12**
42	2	2	2	6	2	2	2	6	**12**
43									**Non-submitter**
44	2	2	2	6	2	2	2	6	**12**
45	2	2	2	6	2	2	2	6	**12**
46	2	2	2	6	2	2	2	6	**12**
47	2	2	2	6	2	2	2	6	**12**
48									**Non-submitter**
49	2	2	2	6	2	2	2	6	**12**
50	2	2	2	6	2	2	1	5	**11**
51	0	2	0	2	0	2	0	2	**4**
52	2	2	2	6	2	2	2	6	**12**

APPENDIX 2. Change log (changes since the last version)

Version Number	Published	Amendments
1	11 August 2026	• 2026 First round interim report published

END OF REPORT