

Administration Office
c/o EMQN CIC Office,
Third Floor, ICE Building
3 Exchange Quay, Salford,
M5 3ED, United Kingdom
Tel: +44 161 757 4952
Fax: +44 161 850 1145
Email: admin@erndim.org

Scientific Advisor
Dr Dulce Quelhas
Laboratório de Bioquímica Genética
Serviço de Genética Laboratorial, Centro de
Genética Médica Jacinto de Magalhães,
Centro Hospitalar Universitário Santo
António EPE, Pr Pedro Nunes 88
Porto, 4099-028, Portugal

Deputy Scientific Advisor
Blai Morales Romero
Sección de Errores Congénitos del
Metabolismo, Servicio de Bioquímica y
Genética Molecular, Hospital Clínic de
Barcelona, C/Mejía Lequerica s/n, 08028,
Barcelona, Spain

Scheme Organisers
1. Sample dispatch
Dr. R.M. Schoeman
Streekziekenhuis Koningin Beatrix
MCA Laboratory
Beatrixpark 1
7101 BN Winterswijk
The Netherlands
Email: mca.office@skbwinterswijk.nl

2. Results Website
Alessandro Salemma; Rose Defossez
CSCQ, Swiss Center for Quality Control
2 chemin du Petit-Bel-Air
CH-1225 Chêne-Bourg
Switzerland
Email: erndim.survey@cscq.ch

Published: 10 February 2026¹

Congenital Disorders of Glycosylation (CDG) 2026 Scheme Instructions

1. General Information

- 1.1. Please read these instructions carefully.
- 1.2. A copy of this document and the covering letter are also available to download from the Participant Information tab of the ERNDiM Registration Website (www.eqa.erndim.org).
- 1.3. If any changes are made to these instructions or the covering letter, the named contacts for your laboratory will be sent an updated version by email. The latest version of these instructions and the covering letter will also be available to download from the ERNDiM Registration Website (see 1.2).
 - **If these scheme instructions are not Version 1 for this scheme year, go to APPENDIX 1 for details of the changes which have been made since the last version of this document.**
- 1.4. If you have any issues (e.g., if you are unable to test the samples, or have difficulty meeting deadlines etc.) please contact the ERNDiM Administration Office (admin@erndim.org) immediately.
- 1.5. **Complaints:** 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.

2. Participant information

- 2.1. Please find enclosed all six lyophilised plasma/serum samples for both rounds of the 2026 scheme.
- 2.2. Please record the date of sample receipt.
- 2.3. Please check all the samples to make sure they are all present, intact, have not been damaged in transit and that the sample numbers match those given in section 4.5. If you have any problems, please use the repeat sample request form [https://www.formdesk.com/erndim/ERNDiM Repeat Sample Requests 2026 Scheme](https://www.formdesk.com/erndim/ERNDiM_Repeat_Sample_Requests_2026_Scheme)

3. Storage & stability:

- 3.1. The samples are not iron-saturated and were lyophilised in the presence of cryoprotectant
- 3.2. The lyophilised samples should be stored in the refrigerator at 2-8 °C and are stable for the duration of the scheme's submission calendar if stored under the defined conditions.
- 3.3. The stability of the reconstituted product is 48 hours when handled appropriately: pure water, replacement of stopper and storage at 2-8°C.
- 3.4. **Reconstitution of the samples:** reconstitute by the addition of MilliQ water just before analysis. The volume of MilliQ water to be used per sample is given in the table under item 4.5.

¹ If these scheme instructions are not Version 1 for this scheme year, go to APPENDIX 1 for details of the changes which have been made since the last version of this document.

3.5. **Safety:** Please see item 8.

4. Analysis and reporting

- 4.1. Analysis should be in two batches according to the time schedule given in section 4.5.
- 4.2. Analyse these samples in the normal way your laboratory would treat routinely tested patient samples, including the use of routine measurement or test methods.
- 4.3. We would like to remind you that the quality of the samples, as well as the quality of clinical information, depends on the contributing participant.
- 4.4. Please note that samples from patients without any known IEM may be included in the scheme. As these samples are from real patients, each sample report should be aligned with real-world practice.
- 4.5. **Sample details**

Sample Number	Sample preparation	Clinical information				Material
		Clinical Information	Sex	Age at Diagnosis	Current Age	
CDG-PP-2026-A	Add 25 µL water before use	Microcephaly, feeding problems, hypotonia	M	5 months	2 years	plasma/serum
CDG-PP-2026-B	Add 25 µL water before use	Epilepsy, intellectual disability, nonverbal	F	12 years	15 years	plasma/serum
CDG-PP-2026-C	Add 25 µL water before use	<i>Retinitis pigmentosa</i> , marked gait imbalance, peripheral neuropathy	F	65 years	68 years	plasma/serum
CDG-PP-2026-D	Add 25 µL water before use	Hepatomegaly, hypoglycaemia, abdominal pain	F	6 months	19 years	plasma/serum
CDG-PP-2026-E	Add 25 µL water before use	Abnormal liver function, neuropathy	F	50 years	58 years	plasma/serum
CDG-PP-2026-F	Add 25 µL water before use	Mild neurological features and hypergonadotrophic hypogonadism	F	3 years	19 years	plasma/serum

- 4.6. **Report** as you would to a **non-specialist paediatrician** in a distant general hospital. If a specialist colleague (e.g., a clinical geneticist or metabolic paediatrician) normally gives **interpretation and advice on further investigation** please include their contribution in your report. Reports must be submitted in English.

5. Result submission

- 5.1. Each participating laboratory must carry out both the analysis and interpretation of the EQA samples. The use of subcontracted (or 'cluster' labs) laboratories is not permitted in this scheme.
- 5.2. Online submission of all results is mandatory.
- 5.3. The results are to be returned via the website: <https://cscq.hcuge.ch/cscq/ERNDiM/>. The named contact for your laboratory should already have a username and password for this website but please contact admin@erndim.org if you have any problems accessing the website.
- 5.4. It is highly recommended that you upload labelled copies of the scans/chromatograms to the CSCQ results website.

5.5. Advice on submitting results

User profile

Methods Specifications

- **Important:** Methods used routinely in your laboratory for diagnosis of patient specimens must be specified before result entry.

Results entry

- **Step 1: Selection of used analytes/methods**
 - Select the methods used for the survey and sample.
- **Step 2: Analytical results input: input of key metabolites to be evaluated**
 - Report each transferrin glycoform preferably as a percentage of total transferrin and provide the reference ranges used by your laboratory for each glycoform. For MS-based methodologies, alternative result outputs consistent with the analytical approach employed are also acceptable. In such cases, both the results and the corresponding reference ranges must be entered in the "Comments".
- **Step 3: Interpretation input:**
 - Specify the type of profile (e.g. normal or abnormal, profile type: suggestive for CDG-I, CDG-II or other).
 - Do not give advice for further investigation here.
- **Step 4: Further lab investigations:**
 - Scored together with the interpretative score.
 - Only advice for further investigation will be evaluated here.

- Advice regarding treatment is not scored.
 - **Step 5: File upload**
 - **Important:** File upload is size dependent.
 - **Step 6: Proof reading**
 - Please review your submitted results immediately after saving them and **print a pdf version** for your records, using the standard menu of your web browser.
- 5.6. A submission reminder will be sent to all scheme participants by CSCQ one week before each submission deadline, BUT please set your own reminders for each deadline in case of communication problems.
- 5.7. **2026 Scheme schedule**

Sample Dispatch Date	10 February 2026	
Circulation	2026/1	2026/2
Sample IDs	CDG-PP-2026-A CDG-PP-2026-B CDG-PP-2026-C	CDG-PP-2026-D CDG-PP-2026-E CDG-PP-2026-F
Analysis start and Website submission availability	13 April 2026	10 August 2026
Reminder for result submission	04 May 2026	31 August 2026
Results submission deadline	11 May 2026 (23:59 CEST ²)	07 September 2026 (23:59 CEST ²)
Annual Report 2026 available	January – March 2027	

- 5.8. Please review your submitted results immediately after saving them and print a pdf version for your records, using the standard menu of your web browser. If it is not possible to review your results and print a pdf version this may indicate a problem with the website which should be reported immediately to admin@erndim.org.
- 5.9. If you miss a submission deadline or realise after the deadline that you need to amend an already submitted result, please contact admin@erndim.org as soon as possible. Please note that result submission extensions will only be granted under exceptional circumstances and no late/amended results can be accepted if the relevant consensus results or diagnoses have already been published.
- 5.10. **IMPORTANT:** please make sure you submit results for all the samples in the scheme year. **If you submit results for 3 or less samples in the scheme year your laboratory will be classed as a partial submitter.** If you do not submit any results your laboratory will be classed as a non-submitter. As the number of participants in this scheme is limited due to the use of clinical samples as the EQA materials, persistent non- and partial submitters may be excluded from participation in future years.

6. Evaluation Criteria

- 6.1. Two criteria will be evaluated: analytical performance and interpretative proficiency. The recommendations pertaining to further investigations should be submitted as a part of interpretative proficiency. The summary of scoring criteria is given below.

A	Analytical performance	Correct interpretation of the profile as normal/abnormal and correctly assigning the abnormal profile type (if relevant)	2
		Correct interpretation of the profile as normal/abnormal, or Correctly assigning the abnormal profile type (if relevant)	1
		Unsatisfactory or misleading (in some instances will be evaluated also as a critical error), or No result submitted	0
I	Interpretative proficiency	Good (diagnosis was established and appropriate further tests were recommended)	2
		Helpful but incomplete	1
		Misleading/wrong diagnosis (will be most likely evaluated also as a critical error), or No result submitted	0

- 6.2. The maximum score achievable is twenty-four points (with six samples) and acceptable performance will generally be held to be a score of seventeen (71%) or more for full submission. **Please note that submission of results for only one out of the two submission rounds will be classified as partial submission, see item**

² CEST = Central Summer European Time, UTC +02:00

5.10 for further details.

- 6.3. Samples with a low overall proficiency (for example 50-60%) will be discussed by the Scientific Advisory Board (SAB) to review their suitability for evaluation. If the SAB agrees that a sample will be classed as an Educational Sample, the scores associated with the sample will be removed from performance evaluations.
- 6.4. In addition, the concept of 'critical error' is applied to the scoring of results in this scheme. In principle this is a category of error that would be unacceptable to the majority of labs and would have a serious adverse effect on patient management. When a critical error is established for one or more samples, performance is not acceptable in that year, regardless of the number of points assigned. All assigned critical errors are ratified by the ERNDiM Scientific Advisory Board.
- 6.5. Further information regarding the Framework for Assessment and Education in Qualitative schemes can be found under the Participant Information tab on the ERNDiM registration website (www.ega.erndim.org).

7. Appeals

- 7.1. During the scheme year queries about results evaluation should be sent to the ERNDiM Administration Office (admin@erndim.org).
- 7.2. After the end of the scheme year, appeals against classification as a poor performer can be made by completing an online 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].

8. Precautions and warnings

- 8.1. For *in vitro* diagnostic use only.
- 8.2. This product should be handled with care, as appropriate for biological materials. Outdated and left-over material should be discarded as potentially infectious material, according to the procedures in your institute.
- 8.3. Please also refer to the relevant MSDS for information on safe handling of the EQA samples in this scheme, which can be downloaded from the ERNDiM registration website under Participant Information.

9. Organisation

- 9.1. This scheme is designed, planned and co-ordinated by ERNDiM. Some activities are subcontracted to the scheme organisers (see page 1); however, ERNDiM remains responsible for this work. For this scheme the subcontracted activities include: aliquoting and labelling of samples, preparation and dispatch of the sample parcel and hosting of the results submission website.

10. Problems

- 10.1. If you have any issues with the samples or submitting your results, please contact admin@erndim.org.
- 10.2. If you believe there is an issue with sample quality or you would like to carry out a repeat analysis, in cases where your results do not match those given in the relevant cycle review, please request a repeat sample using the link to the form given in item 2.3.

APPENDIX 1. Change log (changes since the last version)

Version Number	Published	Amendments
1	10 February 2026	<ul style="list-style-type: none"> • 2026 scheme instructions published.

END

Administration Office

c/o EMQN CIC Office,
Third Floor, ICE Building
3 Exchange Quay, Salford,
M5 3ED, United Kingdom.
Tel: +44 161 757 4952
Fax: +44 161 850 1145
Email: admin@erndim.org

Scheme Organiser**(Sample dispatch & Results Website)**

Dr. R.M. Schoeman
Email: mca.office@skbwinterswijk.nl
Irene de Graaf, BSc (Website manager www.erndimqa.nl)
Email: I.deGraaf@skbwinterswijk.nl
Streekziekenhuis Koningin Beatrix
MCA Laboratory
Beatrixpark 1
7101 BN Winterswijk
The Netherlands

Receptionist's le

Published: 10 February 2026¹

Storage

See instructions!!

**Quantitative & Hybrid EQA Schemes
2026 Covering Letter**

03/03/26

AA

1. General Information

- 1.1. Please find enclosed the samples for the 2026 ERNDIM EQA schemes from the list below, for which you subscribed.
- 1.2. This is a combined shipment for the following schemes:
 - Acylcarnitines in Serum
 - Cystine in White Blood Cells
 - Lipids in Serum
 - Lysosomal Enzymes in Fibroblasts
 - Neurotransmitters in CSF
 - Pterins in Urine
 - Purines and Pyrimidines
 - Quantitative Amino Acids
 - Quantitative Organic Acids
 - Special Assays in Dried Blood Spots
 - Special assays in Serum
 - Special assays in Urine
 - CDG
 - Urine MPS
- 1.3. Also included are the scheme instructions for each of the schemes listed under item 1.2 that you have subscribed for. These include instructions for sample storage, sample handling and submission of results.
- 1.4. **Please read this covering letter and the individual scheme instructions carefully.**
- 1.5. A copy of this document and all the individual scheme instructions are also available to download from the Participant Information tab of the ERNDIM Registration Website (www.eqa.erndim.org).
- 1.6. If any changes are made to this letter or the scheme instructions the named contacts for your laboratory will be sent the updated version(s) by email. The latest version of these documents will also be available to download from the ERNDIM Registration Website (see 1.5).
 - **If these scheme instructions are not Version 1 for this scheme year, go to APPENDIX 1 for details of the changes which have been made since the last version of this document.**
- 1.7. If you have any issues (e.g. if you are unable to test the samples or have difficulty meeting deadlines etc.) please contact the ERNDIM Administration Office (admin@erndim.org) immediately.
- 1.8. **Complaints:** 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.

2. Participant Information

- 2.1. Please check all the samples to make sure they are all present, intact, have not been damaged in transit and that the sample numbers match those given in the individual scheme instructions. See the instructions for each scheme for the number of samples in each scheme. In the event of missing or damaged vials please contact the ERNDIM Administration Office at the earliest opportunity by email, admin@erndim.org and use the repeat sample request form: [https://www.formdesk.com/erndim/ERNDIM Repeat Sample Requests 2026 Scheme](https://www.formdesk.com/erndim/ERNDIM_Repeat_Sample_Requests_2026_Scheme)
- 2.2. **Storage & stability:** see the individual scheme instructions for details.
- 2.3. **Preparation for Use:** see the individual scheme instructions for details.
- 2.4. **Analytes:** see the individual scheme instructions for details.

¹ If this letter is not Version 1 for this scheme year, go to APPENDIX 1 for details of the changes made since the last version of this document.

2.5. Precautions and warnings:

- 2.5.1. See the individual scheme instructions for details.
- 2.5.2. Please also refer to the relevant MSDS for information on safe handling of the EQA samples in each scheme. All available MSDS for ERNDIM EQA samples can be downloaded from the ERNDIM registration website under Participant Information.

3. Result submission

- 3.1. Each participating laboratory must produce its own results and cannot send samples to a subcontracted laboratory.
- 3.2. Results have to be submitted before the relevant deadline via one of the ERNDIM results websites. See the individual scheme instructions for details.
- 3.3. **PLEASE NOTE: the list of analysis methods available for each scheme with results submission on www.erndimqa.nl has been harmonized. The list of available analysis methods for each scheme can be viewed when defining your 'methodset' and analytical methods at the beginning of the scheme year.** The complete list of available methods for each scheme, along with their abbreviations, can also be found on the registration website under 'Information for Participants' here: <https://eqa.erndim.org/information/view/1>. If you have any queries about defining your method, please contact the admin office as soon as possible at admin@erndim.org.

4. Report Publication

- 4.1. See the individual scheme instructions for the schedule for report publication.

5. Organisation

- 5.1. This scheme is designed, planned and co-ordinated by ERNDIM. Some activities are subcontracted to the scheme organiser; however, ERNDIM remains responsible for this work. For each scheme the information on the sub-contracted activities are given in the scheme instructions and include: aliquoting and labelling of samples, preparation and dispatch of the sample parcel and hosting of the results submission website.

6. Problems

- 6.1. If you have any issues with the samples or submitting your results, please contact admin@erndim.org.
- 6.2. If you believe there is an issue with sample quality or you would like to carry out a repeat analysis, in cases where your results do not match those given in the relevant cycle review, please request a repeat sample using the link to the form given in item 2.1.

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