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Published: 06 February 2024[1]

Congenital Disorders of Glycoslyation (CDG)
2024 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 2024 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_2024_Scheme**

2.4. Storage & stability:

2.4.1. The samples are not iron-saturated and were lyophilised in the presence of cryoprotectant

2.4.2. The lyophilised samples should be stored in the refrigerator at 2-8 °C and are stable until 31 December 2024 if stored under defined conditions.

2.4.3. The stability of the reconstituted product is 48 hours when handled appropriately: pure water, replacement of stopper and storage at 2-8°C.

[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.

2.5. **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 3.5. The reconstituted samples should then be stored at -20 °C.

2.6. **Safety:** Please 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.

3. Analysis and reporting

3.1. Analysis should be in two batches according to the time schedule given in section 4.5.

3.2. Analyse these samples in the normal way your laboratory would treat routinely tested patient samples.

3.3. 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.

3.4. Please note that samples from patients without any known IEM may be included in the scheme.

3.5. **Sample details**

Sample Number	Sample preparation	Clinical information			Material
		Phenotype	Sex	Age	
CDG-PP-2024-A	Add 25 µL water before use	Hepatomegaly, intellectual disability, epilepsy	M	8 years	plasma/serum
CDG-PP-2024-B	Add 25 µL water before use	Strabismus, axial hypotonia, deep venous thrombosis	F	10 years	plasma/serum
CDG-PP-2024-C	Add 25 µL water before use	Nephrotic syndrome, hypertrophic cardiomyopathy, osteoporosis	M	5 years	plasma/serum
CDG-PP-2024-D	Add 25 µL water before use	Delayed speech and language development, seizure, Intellectual disability	M	3 years	plasma/serum
CDG-PP-2024-E	Add 25 µL water before use	Hypoalbuminemia, elevated hepatic transaminases, ataxia	F	40 years	plasma/serum
CDG-PP-2024-F	Add 25 µL water before use	Hepatic fibrosis, kyphoscoliosis, peripheral neuropathy	F	15 years	plasma/serum

3.6. **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.

4. Result submission

4.1. 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 allowed in this scheme.

4.2. Online submission of all results is mandatory.

4.3. 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.

4.4. 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.

4.5. **2024 Scheme schedule**

Sample Dispatch Date	06 February 2024	
Circulation	**2024/1**	**2024/2**
Sample IDs	CDG-PP-2024-A CDG-PP-2024-B CDG-PP-2024-C	CDG-PP-2024-D CDG-PP-2024-E CDG-PP-2024-F
Analysis start and Website submission availability	15 April 2024	12 August 2024
Reminder for result submission	06 May 2024	02 September 2024
Results submission deadline	13 May 2024 (23:59 CEST[2])	09 September 2024 (23:59 CEST[2])
Annual Report 2024 available	December 2024	

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

4.6. 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.

4.7. It is not possible to make ANY changes to submitted results after the relevant report has been published.

4.8. **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.

5. Evaluation Criteria

5.1. 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

5.2. 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 4.8 for further details.**

5.3. 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.

5.4. In addition, ERNDIM applies the concept of 'critical error' in the scoring of results. 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. Appeals

6.1. During the scheme year queries about results evaluation should be sent to the ERNDIM Administration Office (admin@erndim.org).

6.2. 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].

7. Precautions and warnings

7.1. For *in vitro* diagnostic use only.

7.2. 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. Organisation

8.1. 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 sub-contracted activities include: aliquoting and labelling of samples, preparation and dispatch of the sample parcel and hosting of the results submission website.

9. Problems

9.1. If you have any issues with the samples or submitting your results please contact admin@erndim.org.

9.2. 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	06 February 2024	• 2024 scheme instructions published

END