

Congenital Disorders of Glycosylation (CDG) 2026 EQA Sample Diagnoses 1st and 2nd round

ERNDIM 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
Unidade Bioquímica Genética
Centro de Genética Médica Jacinto de Magalhães,
Centro Hospitalar do Porto, EPE, Pr Pedro Nunes 88
Porto, 4099-028, Portugal

Email: admin@erndim.org

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

Email: admin@erndim.org

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

Dear Participant,

Please find below the diagnoses for the 2026 CDG EQA scheme 1st and 2nd round.

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	<i>Retinitis pigmentosa</i> , marked gait imbalance, peripheral neuropathy	F	65 years	68 years	PMM2-CDG
CDG-PP-2026-D	Hepatomegaly, hypoglycaemia, abdominal pain	F	6 months	19 years	Hereditary Fructose Intolerance
CDG-PP-2026-E	Abnormal liver function, neuropathy	F	50 years	58 years	Alcohol abuse
CDG-PP-2026-F	Mild neurological features and hypergonadotrophic hypogonadism	F	3 years	19 years	PMM2-CDG

With regards,
ERNDIM Administration Team

On behalf of
Dr Dulce Quelhas
Scientific Advisor, CDG Scheme