

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

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Scheme Organisers

1. Sample dispatch

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Dear Participant,

Please find below the diagnoses for the 2026 CDG EQA scheme 1st 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

With regards,
ERNDIM Administration Team

On behalf of
Dr Dulce Quelhas
Scientific Advisor, CDG Scheme