

Congenital Disorders of Glycosylation (CDG) 2024 EQA Sample Diagnoses 1st round

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Dear Participant,

Please find below the diagnoses for the 2024 CDG EQA scheme 1st round.

Sample ID	Clinical information (sex, age, phenotype)	Sex	Patient Age	Diagnoses
CDG-PP-2024-A	Hepatomegaly, intellectual disability, epilepsy	M	8 years	Control sample - Normal profile
CDG-PP-2024-B	Strabismus, axial hypotonia, deep venous thrombosis	F	10 years	Abnormal profile - Type 1 - PMM2-CDG
CDG-PP-2024-C	Nephrotic syndrome, hypertrophic cardiomyopathy, osteoporosis	M	5 years	Abnormal profile - Transferrin variant

With regards,
ERNDiM Administration Team

On behalf of
Dr Dulce Quelhas
Scientific Advisor, CDG Scheme