

Congenital Disorders of Glycosylation (CDG) 2022 EQA Sample Diagnoses

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Dear Participant,

Please find below the diagnoses for the first round of the 2022 CDG EQA scheme.

Sample ID	Clinical information (sex, age, phenotype)	Sex	Patient Age	Diagnoses
2022-A	Epileptic encephalopathy, facial dysmorphic features	F	10 years	Normal
2022-B	Bilateral congenital hip dislocation, facial dysmorphic features, microcephaly	M	1 year	ATP6V0A2-CDG
2022-C	Autistic spectrum behaviour, seizures, deafness	M	3 years	Transferrin polymorphic variant

With regards,
ERNDIM Administration Team

On behalf of
Dr Dulce Quelhas
Scientific Advisor, CDG Scheme