Congenital Disorders of Glycosylation (CDG)
2022 EQA Sample Diagnoses

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Dear Participant,

Please find below the diagnoses for 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
2022-D	Liver fibrosis, increased transaminases	F	19 years	Normal
2022-E	Hypoglycaemia, hepatomegaly, proximal tubulopathy	M	4 months	Hereditary fructose intolerance (HFI)
2022-F	Intellectual disability, ataxia, low factor XI	M	16 years	PMM2-CDG

With regards,
ERNDIM Administration Team

On behalf of
Dr Dulce Quelhas

Scientific Advisor, CDG Scheme