ERNDIM CDG-PP-2022 D/E/F

Lab412 (Bichat hospital, Paris, France)

2D electrophoresis

2022-E

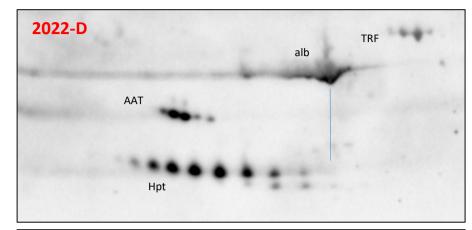
AAT= alpha-1-antitrypsin Hpt = haptoglobin Alb = albumin (non-glycosylated).

Type1

Western-blot of AAT

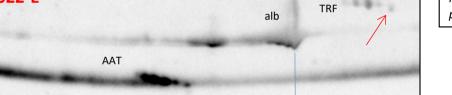
) E





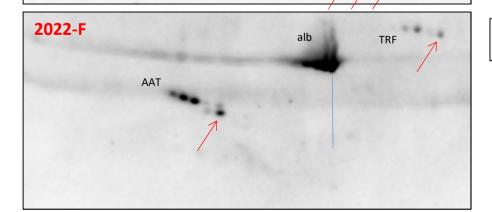
F, 19 years, Liver fibrosis, increased transaminases

Normal 2D pattern
Not suggestive for CDG



M, 4 months, hypoglycaemia, hepatomegaly, proximal tubulopathy.

CDG type 1 abnormalities on AAT, Hpt and Trf. Suggestive for CDG-1



M, 16 years, intellectual disability, ataxia, low factor XI.

CDG type 1 abnormalities on AAT, and Trf. Undetectable Hpt.

Suggestive for CDG-1