

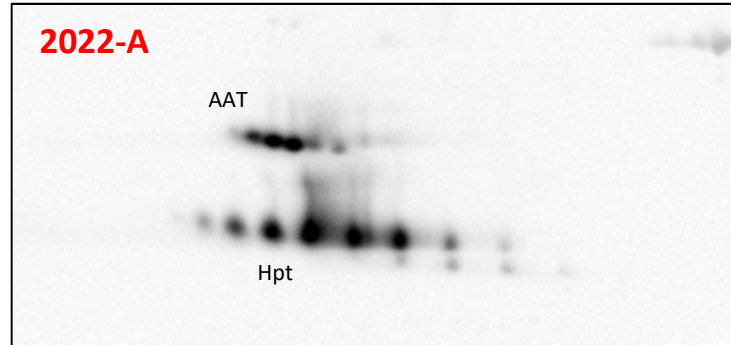
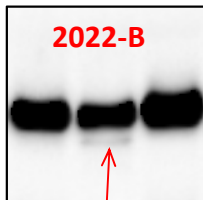
ERNDIM CDG-PP-2022 A/B/C

Lab412 (Bichat hospital, Paris, France)

AAT= alpha-1-antitrypsin
Hpt= haptoglobin

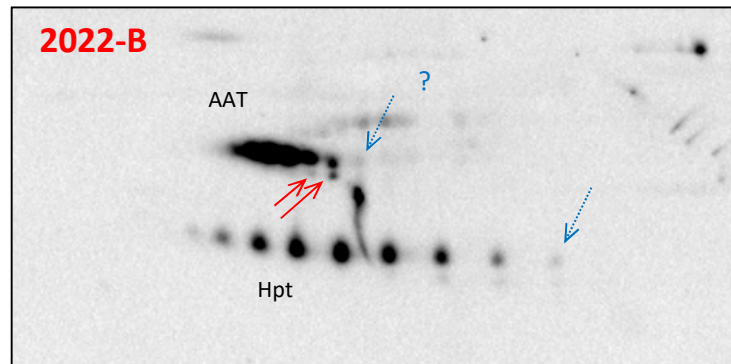
→ Type1
→ Type2

Western-blot of AAT



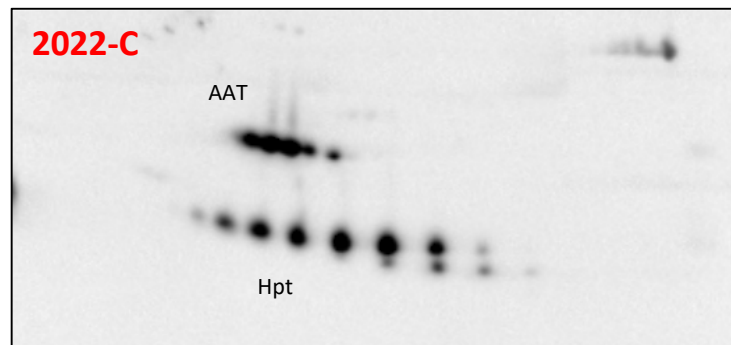
F, 10 years, Epileptic encephalopathy, facial dysmorphic features.

Normal 2D pattern
Not suggestive for CDG



M, 1 year, Bilateral congenital hip dislocation, facial dysmorphic features, microcephaly.

CDG type 1 abnormalities on AAT.
Perhaps discrete associated CDG type 2 abnormalities on Hpt (and AAT?)
Suggestive for CDG-1 (or CDG-1+2?).



M, 3 years, Autistic spectrum behavior, seizures, deafness

Normal 2D pattern
Not suggestive for CDG