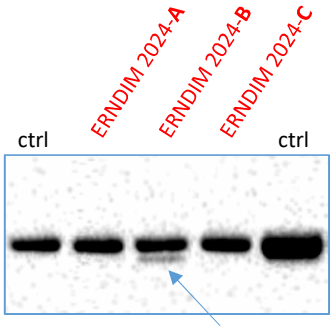
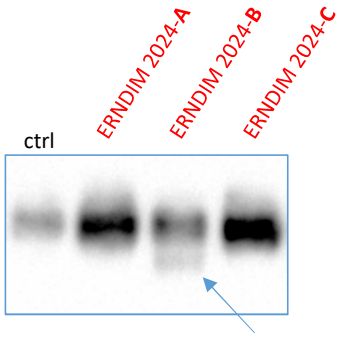
ERNDIM-CDG-PP-2024-A/B/C

CDG-PP-2024-A: *M, 8 y.o; Hepatomegaly, intellectual disability, epilepsy*
CDG-PP-2024-B: *F, 10 y.o; Strabismus, axial hypotonia, deep venous thrombosis*
CDG-PP-2024-C: *M, 5 y.o; Nephrotic syndrome, hypertrophic cardiomyopathy, osteoporosis*

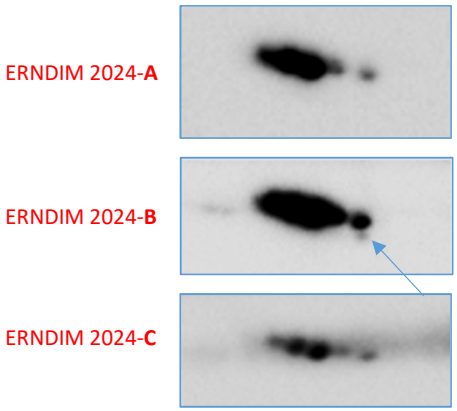
Western-blot of transferrin



Western-blot of α1GPA



2D of AAT



CDG-PP-2024-A:
Normal profile(s). Not suggestive for CDG.
Normal profiles. Do not propose anything in the field of CDG. Nevertheless, since falsely normal profiles have been described in some CDG cases, the diagnosis cannot be totally excluded.

CDG-PP-2024-B:
CDG type1 abnormalities on transferrin, alpha-1 glycoprotein acid (α1GPA) and α1 antitrypsin (AAT). Suggestive for CDG-I.
CDG-I profile(s). Secondary causes of CDG should be excluded (hereditary fructose intolerance, galactosemia, liver disease). Eventually, ask for EDTA blood sample to test PMM2 (PMM2-CDG) and MPI (MPI-CDG) enzymatic activities. Ask for skin fibroblasts to possibly test other enzymatic activities. EDTA blood sample and/or fibroblasts will also be used for molecular sequencing (CDG gene panels, WES...). Ask for an informed consent for genetic studies.

CDG-PP-2024-C:
Normal profile(s). Not suggestive for CDG.
Normal profiles. Do not propose anything in the field of CDG. Nevertheless, since falsely normal profiles have been described in some CDG cases, the diagnosis cannot be totally excluded.