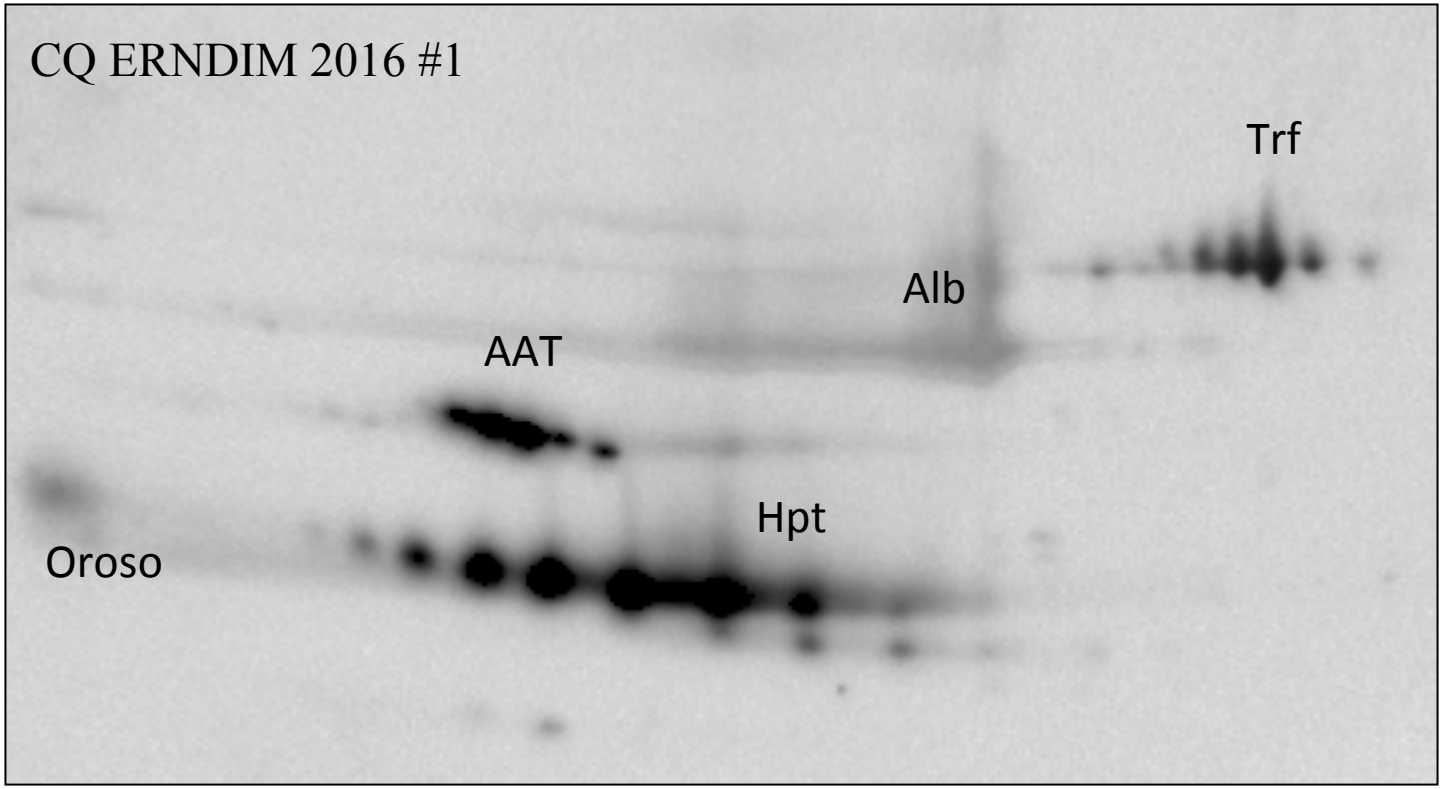
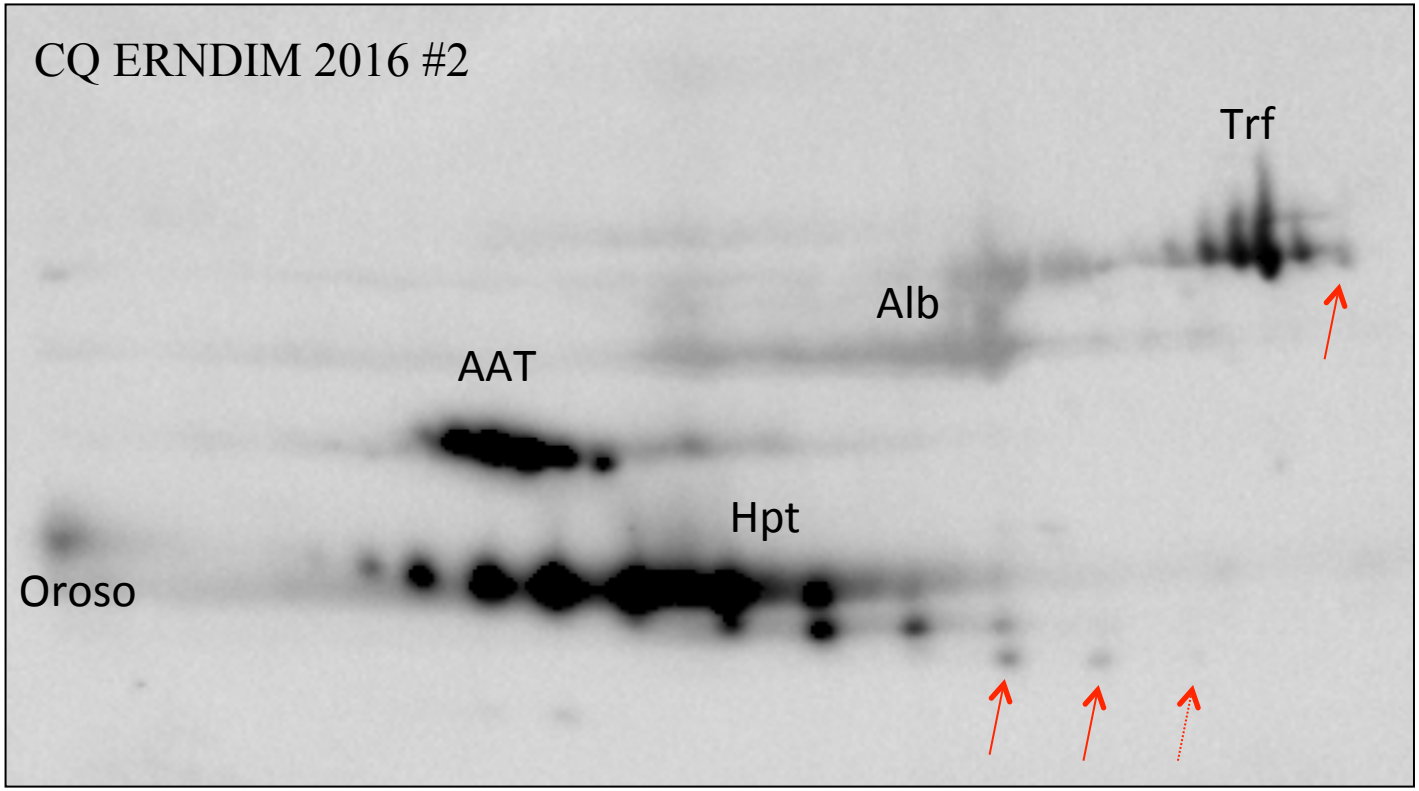


<u>CQ ERNDIM 2016 – n°1</u>

Normal 2D pattern.

The screening tests do not suggest a CDG I or CDG II

Do not propose anything in the field of CDG

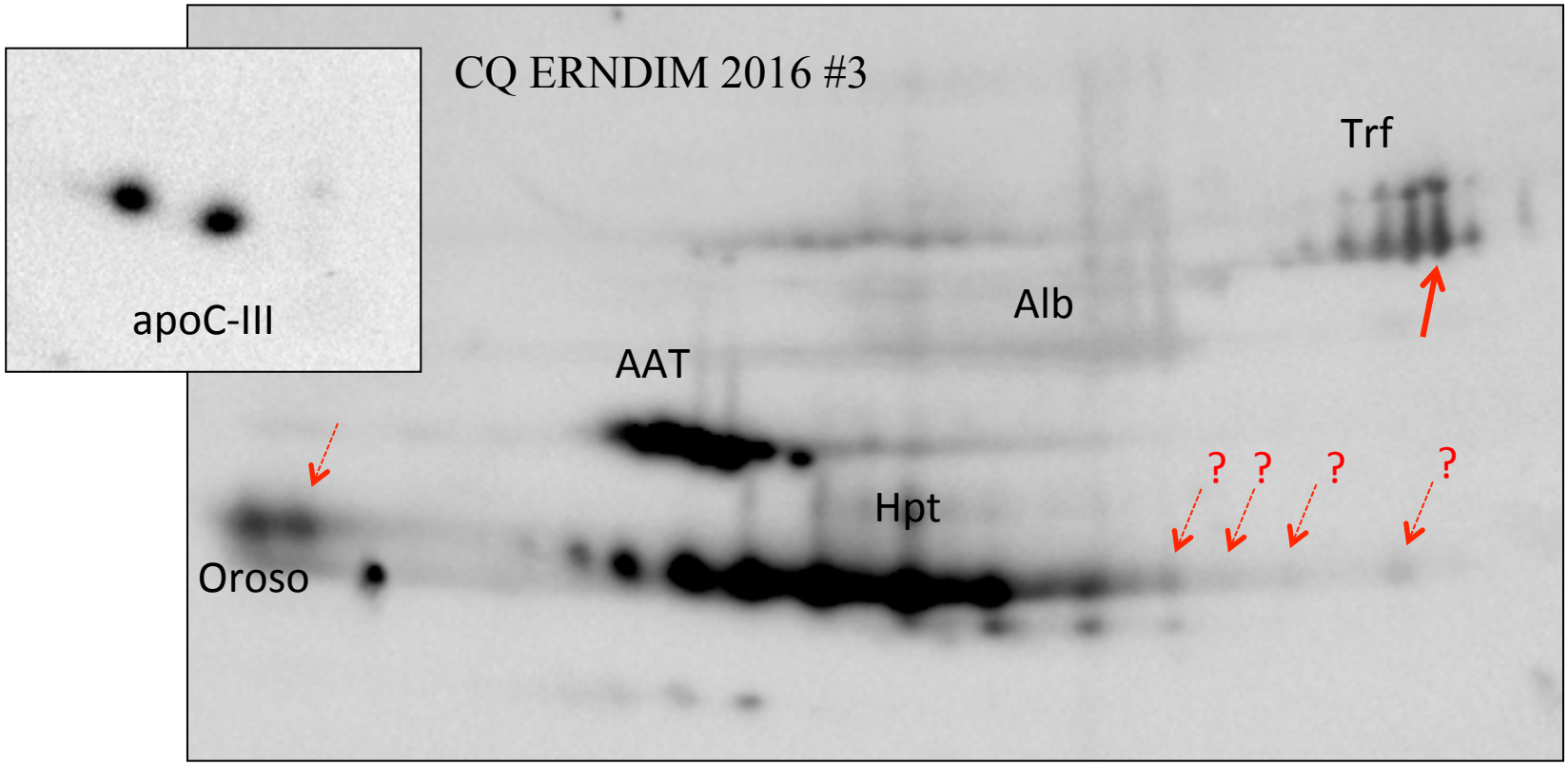


CQ ERNDIM 2016 – n°2

Glycosylation abnormalities for Trf (discrete) **and Hpt**

The screening tests suggest a CDG I

Secondary causes of CDG I should be excluded (galactosemia, hereditary fructose intolerance, alcoholism, liver failure). Ask for EDTA blood sample to test PMM and PMI activities. Ask for a skin biopsy in order to be able to test other enzymatic activities. EDTA and skin biopsy will also be used for molecular analysis (targeted CDG1-related genes sequencing). Ask for an informed consent for genetic study.



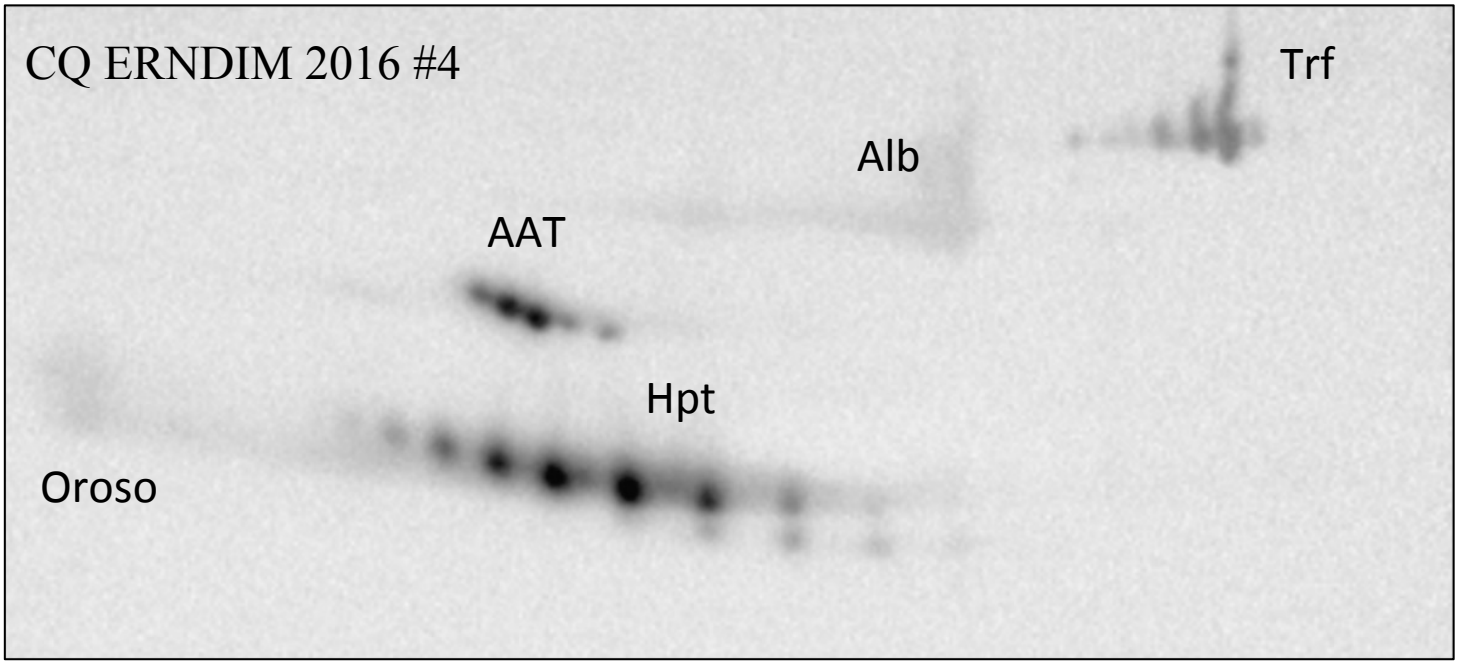
CQ ERNDIM 2016 – n°3

Glycosylation abnormalities or potential variant for Trf

Doubtful 2D pattern for Hpt and oroso

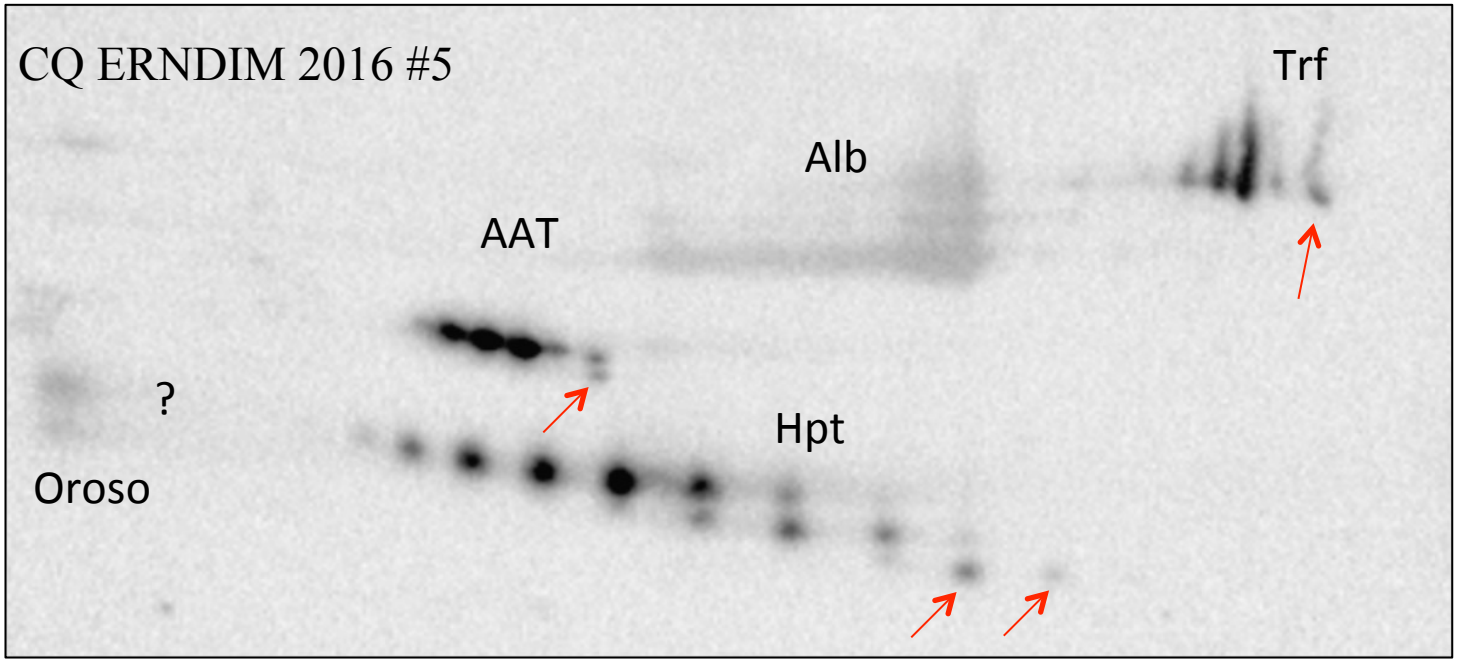
The screening test suggest a CDG II or a potential Trf variant

The transferrin CDG II profile and clinical context (*Cutis laxa*) suggest a potential ATP6V0A2-CDG. But **normal apoC-III pattern** and unclear 2D pattern of others N-glycoproteins (normal AAT and doubtful haptoglobin and orosomucoïd) also suggest a potential Trf variant. To check for this later hypothesis, ask for additional patient's serum sample for (*i*), CE of transferrin with/without neuraminidase and (*ii*), for MS analysis of N-glycans. Once excluded potential Trf variant, ask for additional EDTA blood sample (and informed consent) for targeted CDG2-related genes sequencing.



<u>CQ ERNDIM 2016 – n°4</u>: F, 8 yrs, frequent infections, liver fibrosis

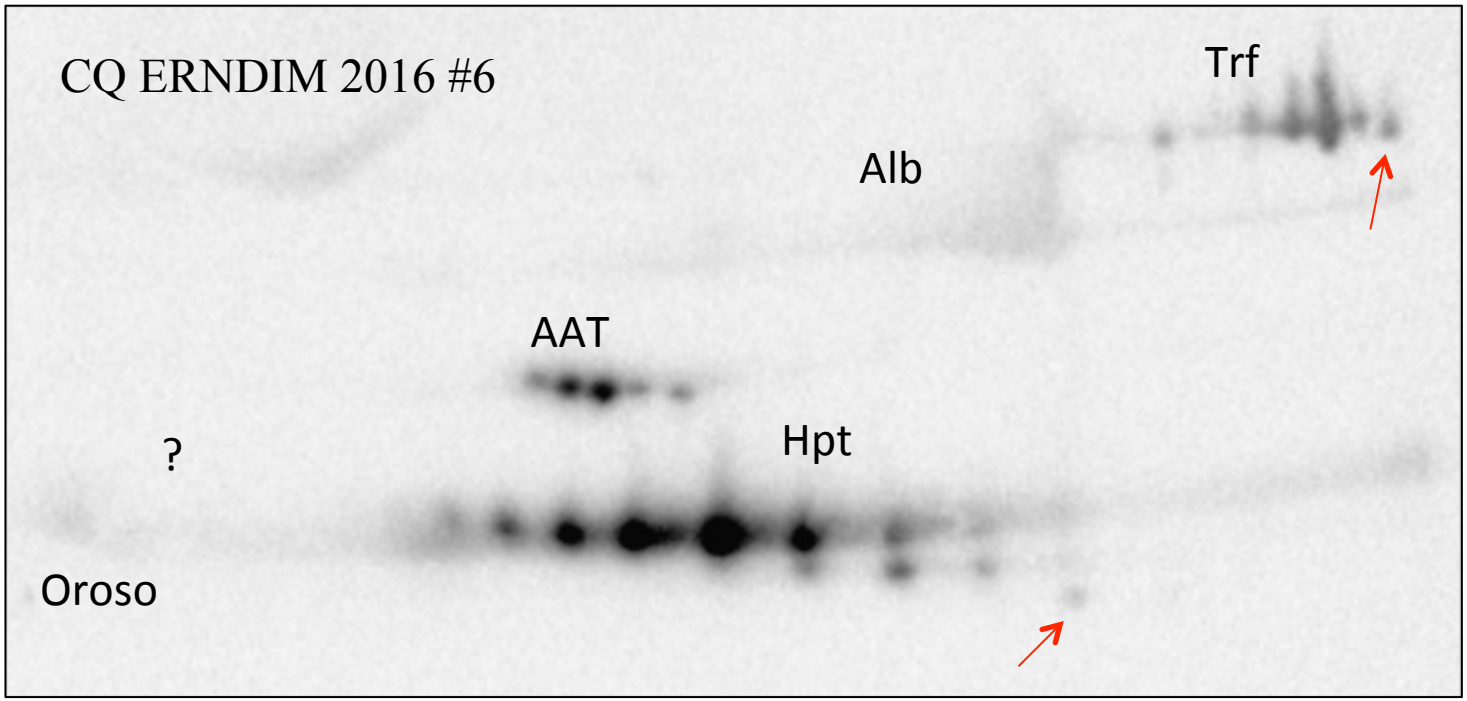
Normal 2D pattern.
The screening tests do not suggest a CDG I or CDG II
Do not propose anything in the field of CDG



<u>CQ ERNDIM 2016 – n°5</u>: F, 1 year, inverted nipples, hypotonia, strabismus, coagulopathy, psychomotor retardation

Glycosylation abnormalities for Trf, AAT and Hpt
The screening tests suggest a CDG I

Secondary causes of CDG I should be excluded (galactosemia, hereditary fructose intolerance, alcoholism, liver failure). Ask for EDTA blood sample to test PMM and PMI activities. Ask for a skin biopsy in order to be able to test other enzymatic activities. EDTA and skin biopsy will also be used for molecular analysis (targeted CDG1-related genes sequencing). Ask for an informed consent for genetic study.



<u>CQ ERNDIM 2016 – n°6</u>: M, 30 years, liver disease

Glycosylation abnormalities for Trf and Hpt

The screening tests suggest a CDG I

In this case, glycosylation abnormalities can be related to liver disease. If possible, ask for a new sample far from liver disease. Other secondary causes of CDG I should be excluded (galactosemia, hereditary fructose intolerance, alcoholism). If abnormal screening test is confirmed, ask for EDTA blood sample to test PMM and PMI activities. Ask for a skin biopsy in order to be able to test other enzymatic activities. EDTA and skin biopsy will also be used for molecular analysis (targeted CDG1-related genes sequencing). Ask for an informed consent for genetic study.