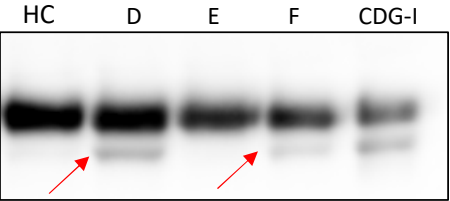


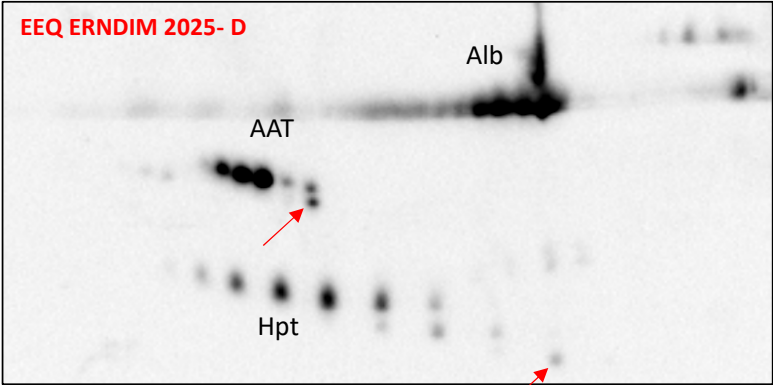
|               |                            |                                                        |   |          |              |
|---------------|----------------------------|--------------------------------------------------------|---|----------|--------------|
| CDG-PP-2025-D | Add 25 µL water before use | Cerebellar hypoplasia, dysmorphic features, nystagmus  | F | 4 years  | plasma/serum |
| CDG-PP-2025-E | Add 25 µL water before use | Autistic behaviour spectrum                            | M | 7 years  | plasma/serum |
| CDG-PP-2025-F | Add 25 µL water before use | Muscular hypotonia, intellectual disability, scoliosis | F | 11 years | plasma/serum |

Western blot of AAT

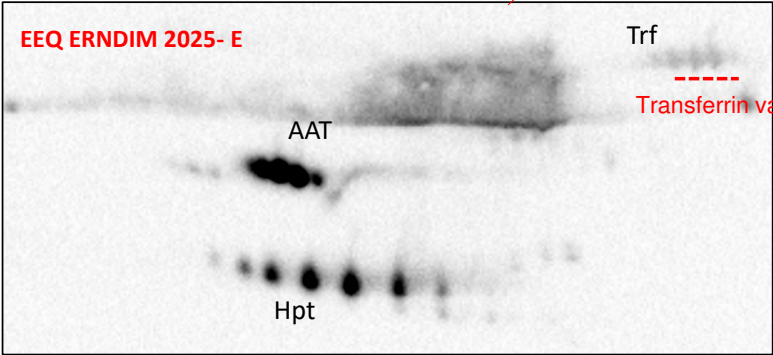


HC: healthy control  
D: EEQ ERNDIM 2025- D → suggestive of CDG-I  
E: EEQ ERNDIM 2025- E → not suggestive of CDG  
F: EEQ ERNDIM 2025- F → suggestive of CDG-I  
CDG-I: CDG-I affected individual

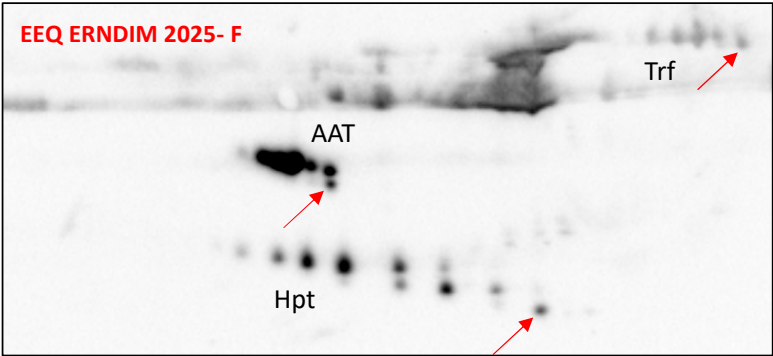
2D electrophoresis of glycoproteins



Suggestive of CDG-I



Not suggestive of CDG  
Possible transferrin protein variant  
(without impact on CDG screening)



Suggestive of CDG-I

AAT: alpha-antitrypsin  
Hpt: haptoglobin  
Trf: transferrin