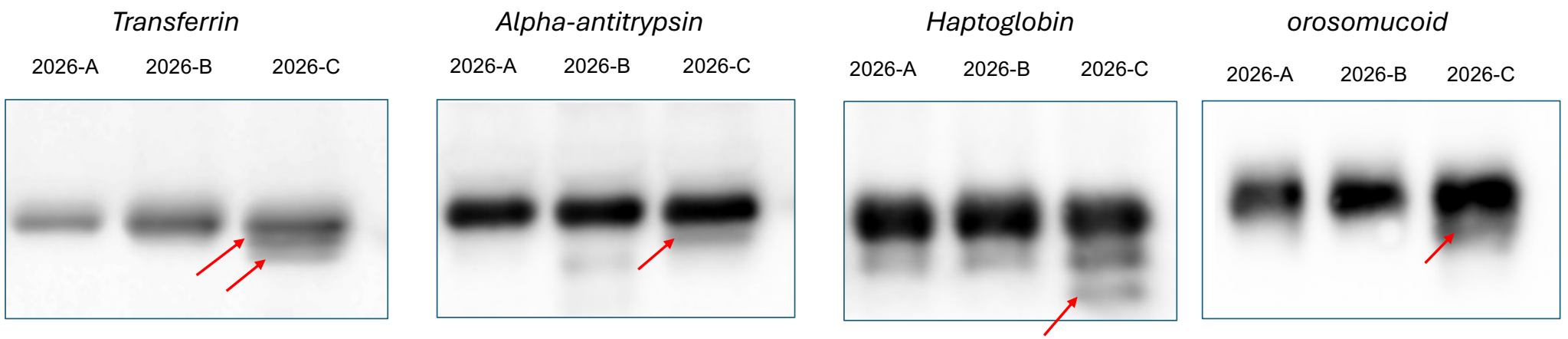
CDG-PP-2026-A: M; 2 years; Microcephaly, feeding problems, hypotonia
CDG-PP-2026-B: F; 15 years; Epilepsy, intellectual disability, nonverbal
CDG-PP-2026-C: F; 68 years; Retinitis pigmentosa, marked gait imbalance, peripheral neuropathy



CDG-PP-2026-A: not suggestive for CDG

Normal profiles for all tested serum/plasma glycoproteins. 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. With the technique we used (Western-blot after SDS-PAGE), a possible Transferrin protein variant (with no impact on the CDG screening) cannot be explored.

CDG-PP-2026-B: not suggestive for CDG – same recommendations as CDG-PP-2026-A

CDG-PP-2026-C: suggestive for CDG-I

CDG-I profiles for all tested serum/plasma glycoproteins. Secondary causes of CDG should be excluded (hereditary fructose intolerance, galactosemia, liver disease). Ask for EDTA blood sample and/or skin fibroblasts for molecular sequencing (CDG gene panels, WES...). In the clinical context, PMM2-CDG, DPAGT1-CDG, DHDDS-CDG can be suspected and will be notably explored. Ask for an informed consent for genetic studies.