

Screening for congenital disorders of glycosylation (CDG): Transferrin HPLC versus isoelectric focusing (IEF).

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OBJECTIVES: Transferrin isoelectrofocusing (Tf-IEF) is widely used to screen for Congenital Disorders of Glycosylation (CDG), but it is laborious, time-consuming, and not suitable for automation or accurate quantification. We present our experience and advantages of the implementation of Tf-HPLC.

METHODS: Sera were iron saturated, lipid precipitated and filtrated on Microcon-YM10. Glycoforms were separated by HPLC on a SOURCE 15Q anion-exchange column. Detection was at 470 nm.

RESULTS: We established reference values and validated the HPLC method by analysing samples with abnormal Tf-IEF. Comparison between both methods is described.

CONCLUSIONS: HPLC is useful for CDG screening, especially for laboratories that deal with great number of samples, due to its easy sample processing, the possibility of performing long series of analysis and the advantage of peak quantification, which allows objective interpretations.