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COMPLEXITY OF THE DIAGNOSIS OF CONGENITAL DISORDERS OF GLYCOSYLATION

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Introduction: Congenital Glycosylation Disorders (CDG) is a group of pathologies caused by the disorder of the glycosylation process of glycoproteins and glycoconjugates with various disabling multisystem impairment mimicking other pathologies.

Keywords: CDG, IEFT, rare disease

Purpose: The implementation of the diagnostic algorithm and identify cases of CDG in the cohort of Moldovan patients.

Material and methods: serum of 40 patients suspected for CDG were analyzed by isoelectric focusing of transferrin (IEFT) and urine by NMR spectroscopy.

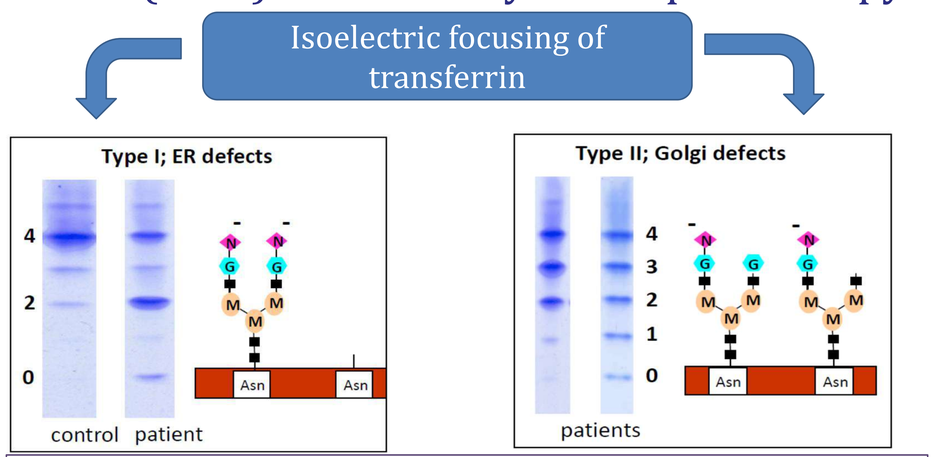

Fig.2 Principles of IEFT

Results: The clinical manifestations of the patients were: hypotonia, hepatomegaly, mild hypoglycemia, increased transaminases, abnormal brain MRI, dysmorphic features, failure to thrive and neurological manifestations.

Fig. 1. IEF of transferrin: 1-3 abnormal profile suggestive for CDG I;

37 serum – normal IEFT profiles,

3– abnormal IEFT profile:

1. Fructosemia
2. Galactosemia
3. Possible CDG needed additional investigations.

Conclusions: The variety of symptoms in CDG lead to missdiagnosis other pathologies. In the process of diagnosing CDG it is mandatory to exclude secondary abnormalities of glycosylation.