USING THE METHOD OF GAS CHROMATOGRAPHY-MASS SPECTROMETRY DURING CONFIRMATORY DIAGNOSTIKS OF HOMOGENTISURIA

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Introduction. Among the genetic children's diseases much more common widespread hereditary metabolic disease or metabolic diseases - with monogenic disease genes in which mutations lead to pathochemical disorders leading to clinical and biochemical picture of the disease.

Aim. The object of work was to determine the optimal methods of diagnosis in patients with homogentisuria.

Materials and methods. Clinical research methods, medical history, physical examination of patients with homogentisuria.Laboratory methods included: gas chromatography, mass spectrometry, urine screening diagnostics, analysis of synovial fluid. We used rapid tests using urine analyzer and urine Arkray test strips (Aution Sticks AE 10).

Results and discussion According to the 2016 GC-MS, we have examined 924 patients, performed 243,313 investigations, of which in 2 were found homogentisuria sick child. The study was fulfilled at the HSMHTS-CA (O) C. When the biological materials were accepted in the laboratory we performed selective screening urine. Most often detected changes were in the proportion (increasing in 34.8% of patients, decreasing -in 2.5%); increasing of sulphites - in 16.2%; the presence of reducing substances - 1.8 (in trace amounts - in 27.5% patients). After receiving the total ion chromatograms, we have carried out the identification of detected organic compounds, searching trivial items adopted in the clinical practice and determine the individual characteristics of each organic acid release time (RT) for total ion chromatogram and retention index (RI). In the course of this work we have identified 117 organic compounds. On the basis of preparation of the mother liquor and preparation working solutions of organic acids graphs of calibration dependence were made for 25 organic acids (OK).

Conclusion. Analysis of urine organic acids- a highly informative method of laboratory diagnosis of metabolic disorders. This allows to doctor can get accurate information about the quantitative characteristics of the available biochemical markers of hereditary metabolic disorders and various exogenous toxins.