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Universitatea de Stat de Medicină și Farmacie „Nicolae Testemițanu”
Societatea de Pediatrie din Republica Moldova



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MTHFR GENE TESTING IN JIA: CLINICAL INDICATIONS

Vladimir Iacomi, Ninel Revenco

INTRODUCTION

Estimation of efficacy in methotrexate (MTX) therapy is investigated by multiple prediction models. One of them is the assessment of pathological polymorphisms of the MTHFR gene.

SCOPE

Elucidation of the presence of mutations in the MTHFR gene in children with JIA following MTX. Assessment of the DAS 28 and JADAS 71 score and the relationship with the investigated polymorphisms.

METHODS

In the analytical, case-control study, with the evaluation of 68 patients with JIA, the genetic polymorphism was tested using the Real Time PCR technique. By ROC analysis, the relationship between DAS 28 and JADAS 71 scores with MTHFR was established.

CONCLUSION

The ROC analysis, based on the sensitivity-specificity relationship, with relevant values for AUC, confirms the recommendation of testing the genetic polymorphism of the MTHFR gene in the case of MTX treatment and the persistence of a high degree of disease activity in patients with JIA.

RESULTS

The sample included 68 children in which MTHFR mutations were recorded in 33.8% (23 cases). JADAS 71 value ≥ 11 has been determined to be a threshold value that can be used to identify potential cases with MTHFR mutation. Likewise, DAS28 value ≥ 3.245 was determined to be a threshold value that can be used to identify potential cases with the mutation.

