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MOLECULAR GENETIC RESEARCH IN THE DIAGNOSIS OF CONGENITAL DYSFUNCTION OF THE ADRENAL CORTEX

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Annotation

Molecular genetic methods are a series of medical tests to detect changes in genes, chromosomes, and proteins. In the modern world, molecular genetic research is increasingly being introduced into the standards of medical care for patients with endocrine pathology[1,2,3].These methods are necessary not only for diagnosis and diagnosis, but also for the selection of personalized therapy. Molecular genetic studies play a separate role in the diagnosis of endocrine diseases in children and adolescents, as they determine the prognosis, treatment strategy and quality of life of these patients[4,5]. Purpose: To evaluate the diagnostic value of molecular genetic research methods in diagnosing a patient with suspected CDCN. Congenital adrenal hyperplasia (CAH) is characterized by impaired biosynthesis of cortisol. 21-hydroxylase deficiency is the most common cause of CAH affecting 1 in 10000-15000 live births over the world[6,7,8,10]. The frequency of the disorder is very high in Iran due to frequent consanguineous marriages. Although biochemical tests are used to confirm the clinical diagnosis, molecular methods could help to define accurate diagnosis of the genetic defect[9,11,12]. Recent molecular approaches such as polymerase chain reaction based methods could be used to detect carriers and identify different genotypes of the affected individuals in Iran which may cause variable degrees of clinical expression of the condition. Molecular tests are also applied for prenatal diagnosis, and genetic counseling of the affected families[13,19,20]. Here, we are willing to delineate mechanisms underlying the disease, genetic causes of CAH, genetic approaches being used in the country and recommendations for health care improvement on the basis of the molecular and clinical genetics to control and diminish such a high prevalent disorder in Iran[15,17,18]. Also, the previous studies on CAH in Iran are gathered and a diagnostic algorithm for the genetic causes is proposed[14,16,17].

Keywords

Congenital Adrenal Hyperplasia, 21 Hydroxylase Deficiency, Iranian population, Molecular Genetics.

Materials and methods: The clinical history of a male patient, age 1 year 7 months, was analyzed. Child from the 6th pregnancy, delivery by cesarean section at the 37th week of gestation. Apgar 8/9. After birth, the condition is satisfactory. Jaundice from 3 days. Echo-KS: open oval window. Ultrasound of the urinary system: diffuse changes in the kidneys. Pyeelectasia on the left. Discharged on the 5th day. Then he was observed on an outpatient basis. The condition is satisfactory. According to the results of neonatal screening, 17OPS was increased to 82 nmol/l. Sent for hospitalization to the Multidisciplinary Hospital in Samarkand. Objectively: the condition is satisfactory. The mother had no complaints about vomiting or regurgitation. I gained adequate weight. The reaction to the examination was adequate, calm, and did not scream. Motor activity is sufficient, muscle tone is physiological, innate reflexes are evoked. There are laboratory changes in hormonal status[14,15]. A blood test showed the following results: cortisol: 19.31 nmol/l (normal range is 28–70 nmol/l); 17OH-progesterone 18.6 ng/ml (normal 0.96–10.46 ng/ml), adrenocorticotropic hormone (ACTH) 119 pg/ml (normal less than 80 pg/ml), renin in blood plasma 127.4 pg/ml. Ultrasound of the adrenal glands without pathology. Based on the results of the study (a significant increase in ACTH, renin, a decrease in cortisol levels, an increase in OPS 17), hypocortisolism of adrenal origin cannot be excluded. Hormone replacement therapy was prescribed: hydrocortisone at the rate of 13 mg/m² - 1 mg 3 times a day. A telemedicine consultation was conducted with specialists from the National Medical Research Center for Endocrinology of the Ministry of Health of the Republic of Uzbekistan in order to resolve the issue of DNA diagnostics of 21-hydroxylase deficiency. The result of TMK - the diagnosis was not in doubt; It is recommended to conduct a molecular genetic study (detection of CYP21 gene mutations) and continue therapy with Cortef and Cortinef at the calculated dose[10,11]..

Results: A molecular genetic study was carried out (detection of CYP-21 gene mutations). The material was collected at the age of 1 month. 2 weeks; final report received at 4 months of age. According to the results of the study, congenital dysfunction of the adrenal cortex was excluded; mutations in the CYP-21 A2 gene were not identified. With the results of the study, the child was sent for further examination to the Republican Scientific Research Center for Emergency Medicine, during which a decision was made to discontinue therapy with hydrocortisone and fludrocortisone.

Conclusions: Molecular genetic research methods are a necessary diagnostic method for making a diagnosis and choosing treatment tactics for patients.

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