many inherited metabolic disorders. Current trends in the treatment are aimed at only symptomatic therapy. During the period from 2011 to 2014, in Moldova were examined children with different metabolic disorders using the following methods: fluid chromatography, NMR and mass spectrometry methods. In base of obtained data the National Register of rare diseases was elaborated. It includes 12 metabolic diseases: methylmalonic aciduria, glutaric aciduria, galactosemia, alcaptonuria, glycogen accumulation diseases, lysosomal diseases, mitochondrial diseases and others. Genetic diagnosis methods include PCR analysis, DNA sequencing, Southern blot method, and allow to reveal the problem at an early stage of development.

Conclusion: The elaboration of the National Register of the rare diseases and introduction into medical practice of the molecular methods of diagnostic of inborn errors of metabolism will help to reduce the mortality and morbidity in children due to early detection of problems and their early treatment.

284. GENETIC STUDY OF CLINICAL VARIABILITY IN THE CRANIO VERTEBRAL JUNCTION ANOMALIES

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Introduction: Cranio-vertebral anomalies represent defects of the development in the structures which are located in the transition zone between mobile cranium and relatively rigid spinal column and can involve brain, spinal cord causing various neurological clinic. Study of these conditions is very important and actual in connection with the development of direction in manual therapy as cranio-sacral therapy.

Materials and methods: Theoretical and methodological basis of the study is scientific aspects studied in the domain of congenital vertebrology. The most important part of analysis is based on material of publications which are containing specific studies from the other countries and international statistics.

Results of this research: There were determined health and development particularities of people with cranio-vertebral disorders; substantiated the main concepts in the occurrence of cranio-vertebral anomalies showing controversies regarding the dynamics of its development; found value and interaction of different factors of influence on the development of cranio-vertebral region; gave reasons for the early identification of developmental points anomaly risk of cranio- cervical junction. Investigated data suggests that malformations in the cranio-vertebral region are quite common among patients in the department of neurology. Among patients who come in the neuro-surgery department with atlanto-axial dislocation 25% have congenital variant of displacement. Clinical polymorphism correlates with a variety of changes at the genetic level.GDF3,GDF6 and MEOX1 genes are involved in bone development and mutations in these genes cause heterogeneity in Klippel-Feil syndrome(KFS).

Klippel-Feil syndrome is clinically characterized by a short neck, low posterior hairline and limited neck movement.

Conclusions: The present study provides sufficient evidence that KFS is caused by a mutation in the MEOX1 and GDF3,GDF6 genes. This issue which has a scientific and clinical interest require an interdisciplinary approach that will ensure efficient planning of resources with involving of a performance type of management aimed to improve the situation in this category of patients as soon as possible.

Key words: cranio-vertebral anomalies, variability

285. ALDOSTERONE SYNTHASE GENE CYPIIB2 -344C/T POLYMORPHISM AND GENDER ASPECTS OF ANTIHYPERTENSIVE TREATMENT EFFICACY

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Background. There is growing evidence that high interindividual variability in response to blood-lowering medications is partially explained by genetic factors. Multiple genes, encoding blood pressure-regulating drug receptors and receptor response mechanisms are Associated with different results in achieving target BP values under antihypertensive treatment. Despite some consistent research, showing that various genetic single-nucleotide polymorphisms (SNP) may affect antihypertensive treatment efficacy, study results in this field continue to be conflicting and provide disparate results [1]. Aldosterone is the key mineralocorticoid rennin-angiotensin-aldosteron system (RAAS) hormone, affecting distal nephron to regulate sodium resorption, excretion of potassium, and intravascular volume. So the associations between aldosterone synthase gene polymorphism and hypertension would thus be of significant interest. Studies about the potential role of aldosterone synthase gene CYP11B2 (-344T/C) polymorphism and primary hypertension demonstrated controversial results. Some results indicate that -344T/C polymorphism has an impact on hypertensive target organ damage and the response to antihypertensive drugs [2 YP41.B2 (-344T/C) gene. For example, studies have polymorphism is Associated with the antihypertensive response to diuretics and RAAS-inhibitors [5 □7]. Due to small study samples and controversial results, even in conditions of one population, it remains unclear, whether CYP11B2 -344T/C single-nucleotide polymorphism (SNP) affects antihypertensive treatment response and long-term treatment outcomes.

Gender-related aspects of hypertension is a research field based on physiological tendency of men to have higher BP values during the whole lifespan, regardless of race or ethnicity. Men also tend to have more modifiable risk factors, such as excessive alcohol consumption, smoking, poor diet, sedentary lifestyle, etc. [8] Highlighting mechanisms, underlying sex differences in hypertension may lead to development of tailored therapeutic strategies, adaptive to specific gender-related variables, thus improving treatment outcomes [9 □10].

Aim. Current study aimed to evaluate gender aspects of interindividual response to antihypertensive treatment along with the role of SNP CYPIIB2 -344C/T in achieving target BP levels.