World Academy of Science, Engineering and Technology
International Journal of Biomedical and Biological Engineering
Vol:16, No:12, 2022

Genomic Imprinting as a Possible Epigenetic Cause of Esophageal Atresia

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Abstract : Introduction: The cause of the isolated form of esophageal atresia has been yet unknown. Objectives: The primary objective of this study was to indicate epigenetic factors which may play an important role in the etiopathogenesis of esophageal atresia. Methods: We recruited a group of 6 pairs of twins, among whom one of the twins developed EA. The selection of such a group for testing allows for excluding external factors (e.g., infections, drugs, toxins) as the cause of the birth defect. The analyzes were performed with the use of genetic material isolated from the whole blood and esophagus tissue of a patient with EA. The reduced representation bisulphite sequencing (RRBS) technique was used to study the change in the genomic imprinting -a change in the expression of genes, which may be the epigenetic cause of EA. Results: In the course of the analyzes, significant hypomethylation and hypermethylation regions were identified. 65 genes with probably increased expression and 65 with decreased expression were selected. These genes have not been marked in literature as possibly pathogenic in esophageal atresia. However, their participation in the pathogenesis of esophageal atresia cannot be clearly excluded. Conclusion: We suggest a role of hypomethylation or hypermethylation of selected genes as one of the possible epigenetic factors in EA pathogenesis. The use of the RRBS technique in the search for the cause of EA is pioneer research; therefore, it seems necessary to extend the research group to new patients with EA. Acknowledgment: The work was supported by the National Science Centre, Poland, under research project 2016/21/N/NZ5/01927.

Keywords : esophageal atresia, epigenetics, embryonic development, surgery, genes expression, twins
Conference Title : ICECT 2022 : International Conference on Epigenetics, Chromatin and Transcription
Conference Location : Istanbul, Türkiye
Conference Dates : December 20-21, 2022