# Role of epigenetic factors in etiopathogenesis of esophageal atresia.

The causes of isolated esophageal atresia remain unknown. Current researches show that environmental factors may play an important role in development of EA. Molecular tests conducted on patients with EA in order to analyze structure of their genome did not identify genes responsible for EA development. Collected data shows that EA occurs 2-3 times more often in one of the twins than in the overall population. The goal of planned research is to analyze epigenetic factors in terms of etiology of EA development. We plan to analyze environmental factors, affecting parents of the child with EA, which may disrupt developmental processes of gametes and fetus. In planned research we will analyze genetic material in terms of its structure, as well as, in terms of function of particular genes. Research will be conducted using latest technology used in molecular genetics. Method will be evaluated in terms of its usefulness in finding causes of isolated birth defects.

Six pairs of monozygotic twins will be examined in this research. In each pair one of the siblings has the EA. Patients with EA will create the research group, whereas their healthy siblings will create the control. In the first phase medical history of people from both control and research group will be thoroughly analyzed and physical examination will be performed on all participants. Parents will fill in the original survey concerning their exposure on environmental factors before pregnancy and during its early period. During next phases peripheral blood will be collected in order to conduct detailed molecular analysis. Esophagus tissue extracted from patients from group with EA during the repair of esophagus will also be used. We will analyze differences between research and control groups and find correlation between nature of this differences and occurrence of EA. Moreover, we will attempt to further the knowledge about mechanisms playing an important role in development of the defect.

It will be a pioneer study on a global scale. It's results may help in understanding basic molecular causes of birth defects.