10. CLINICAL-GENETICS ANALYSIS OF EHLERS-DANLOS SYNDROME TYPE IV

Author: Bolfosu Elena

Scientific adviser: Mariana Sprincean, PhD, Associate Professor, Department of Molecular Biology and Human Genetics, *Nicolae Testemitanu* State University of Medicine and Pharmacy of the Republic of Moldova.

Introduction. Ehlers-Danlos syndrome type IV, vascular type, represents mutations in the procollagen type III (COL3A1) gene. Confirmation of the presence of a COL3A1 mutation and its nature can help evaluate the risks of complications.

Aim of study. Elucidating the clinical-genetics aspects of Ehlers-Danlos syndrome type IV.

Methods and materials. To achieve the proposed objective, a synthesis of the literature published from 2009 to 2021 was performed using 30 bibliographic sources, including electronic libraries such as PubMed, Medscape.

Results. Arterial events were defined by symptomatic dissections, aneurysms or spontaneous arterial ruptures. Arterial ruptures or dissections are responsible for the majority of deaths. In childhood many individuals with vascular type are first thought to have coagulation disorders. In adulthood, four main clinical findings, including a striking facial appearance, easy bruising, the skin is abnormally thin and pale, it is smooth, soft and velvety with multiple venous lesions.

Conclusion. Most affected patients survive the first and second severe complications, but Ehlers-Danlos syndrome type IV leads to sudden premature death. The diagnosis should be assumed in young people presenting to the doctor because of uterine rupture during pregnancy or arterial or visceral rupture. Early diagnosis is particularly important for surgeons, radiologists and obstetricians because knowing the diagnosis can help manage complications.