Hypermobile Ehlers-Danlos syndrome (hEDS) is the most common type of Ehlers-Danlos syndrome and taking into account the prevalence of this condition (1/5000 newborns), it is one of the most common of all genetic disorders. According to one of the best researchers and diagnosticians of Ehlers-Danlos syndrome prof. Roodney Grahame hEDS it is one of the most neglected disorders in history of modern medicine. Questions about genetic etiology of this disease were first raised in 1986, when the first nosology of EDS was established. Up to now gene mutations underlying other types of EDS were determined, but the hypermobile EDS type is still left without molecular genetic diagnosis.

Clinical manifestations include: disfunction of skin (hyperextensibility, smooth, velvety skin, easy bruising, hernias, diastemas) and systems such as: musculoskeletal (joint hypermobility with recurrent joint dislocations), gastrointestinal (gastroesophageal reflux, gastroesophageal hernia, irritable bowel syndrome), cardiovascular (cardiac arrhythmias), nervous (chronic fatigue, postural orthostatic tachycardia syndrome (POTS), urogenital as well as psychiatric disorders and chronic pain.

Due to the wide spectrum of symptoms and their variety, most patients need many years to obtain proper diagnosis (often by exclusion of other connective tissue disorders). This situation is a result of undetermined genetic etiology of this disorder.

Aim of the present study is to establish genetic background of hEDS. Availability of molecular diagnostics of hEDS will allow the implementation of individual therapies, that may help improving quality of life of hEDS patients. Accurate and quick diagnosis will give them a chance for adequate multispecialty care.